Overcoming Barriers, Finding Solutions: Parent and School Staff Perspectives on the School Experiences of Children with Cystinosis

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Table of Contents

List of Tables and Figures

Abstract

Over 300 million people live with rare conditions worldwide, 70% of which occur during childhood. The rare disease community has emphasised the need to shift beyond a medical focus towards a broader understanding of the day-to-day challenges and experiences of people living with rare conditions. Cystinosis is a rare, chronic, multi-systems condition affecting 1 in 200,000 people in Ireland. Through a systematic literature review and qualitative empirical research, this thesis aims to explore strengths and needs of children with cystinosis and the impact of living with cystinosis on children's school experiences.

Underpinned by Bronfenbrenner's bioecological theory, the systematic literature review found that while children and young people with cystinosis may be at risk for social-emotional difficulties, there is a significant lack of qualitative research that contextualises these risks. There is very little focus on the experiences of children with cystinosis in the context of school. The empirical research explored the perceptions of parents (N= 5) and school staff (N =7) on the psychosocial strengths and needs of children with cystinosis (aged 4 to 16 years) as well as the factors impacting their experiences in the school system. Results highlighted their resilience and strengths, while also acknowledging that children's needs change over time as they face new challenges. The research highlights the importance of collaboration at a child, school, and multidisciplinary level. There is a need for increased knowledge about cystinosis at a broad level, combined with an understanding of the individual needs of the child in the context of school. Implications for psychological practice and education are outlined.

Keywords: children; cystinosis; psychosocial; wellbeing; school; multidisciplinary.

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Dedication

I dedicate this thesis to my husband Dave, for your constant unwavering belief in me. It goes without saying that none of this would have been possible without your support and encouragement. I hope you know that I see the sacrifices you have made to make this possible. Thank you for keeping the show on the road, for taking our boys on “daddy adventures”, and for your patience, pep-talks, advice and jokes. Finally, after six years, thank you for the champagne.

Statement of Original Authorship

I hereby certify that the submitted work is my own work, was completed while registered as a candidate for the degree stated on the Title Page, and I have not obtained a degree elsewhere on the basis of the research presented in this submitted work.

“Alone we are rare, together we are strong.”
National Organisation for Rare Disorders (NORD, 2024a)

Chapter 1: Introduction

The European classification of a rare disease or disorder is an occurrence of less than 1 in 2,000 people (Rare Disease Ireland, 2024b). While there are over 6,000 different identified rare diseases, some commonalities exist between them in that they are chronic, severe and/or progressive, and present with various degrees of sensory, motor, physical and/or intellectual disability (Aymé, 2012; Rosselló, 2018). Approximately 70% of rare diseases have their onset in childhood, a time of critical development (Nguengong et al., 2020). For many children with rare diseases, medical advancements and increased survival rates mean many children with rare diseases can participate in mainstream education (Sommer & Klug, 2024). While this is a welcome development, it also poses challenges at both the individual and systemic level. At the individual level, children with rare diseases face many of the same challenges as others in navigating educational and social situations; however these challenges are further exacerbated when managing a rare condition. Factors such as medical and care needs, irregular attendance, and prolonged absences due to hospitalisation and illness may mean that young people with chronic illness fall behind their peers (Lum et al., 2017; Runions, 2020). A substantial amount of research has linked chronic illness with poor academic outcomes (Lum et al., 2017). Socially, research has shown that children with rare and/or chronic diseases experience social isolation, bullying, and discrimination (Adama et al., 2023; Foster et al., 2022; Lum et al., 2017).

At a systemic level, current policy and practices strive to encourage inclusion of children with rare diseases in mainstream schools. However, teachers are often unaware of the complexities of rare chronic health conditions (Runions et al., 2020). A general lack of resourcing, training, and clear policy on supporting children with rare diseases may increase challenges for teachers, parents, and students alike (Adama et al., 2023; Foster et al., 2020). Acknowledging this, the National Rare Disease Plan for Ireland 2014-2020 (Department of Health [DoH],

2014) noted a need to integrate additional training on rare diseases into initial teacher education and professional development. This plan also noted a general lack of research with respect to the broader aspects of living with rare conditions including within the education service. At an international level, the Rare 2030 Foresight Study (Kole et al., 2021) emphasises the need to move beyond physiological symptoms of rare diseases and focus on an integrated support for mental health alongside education and training for healthcare professionals.

Overview of Cystinosis

Cystinosis is a rare, chronic, multi-system metabolic condition which causes an accumulation of the amino acid cystine in the organs and tissues of the body, causing widespread tissue and organ damage (Doyle & Werner-Lin, 2015; NORD, 2023). Current estimates indicate that cystinosis occurs in approximately 1 in 200,000 live births within developed countries (Cystinosis Ireland, 2023). Management of the condition is complex and requires treatment with several drugs with a strict dosage schedule (Ariceta et al., 2016; Ariceta et al., 2019). A primary treatment for cystinosis is cystine depletion therapy (Cysteamine) which slows the development and progression of kidney damage and enhances physical growth (NORD, 2024). Cysteamine has an unpleasant smell and taste and side effects include nausea, vomiting, gastric reflux, and an unpleasant odour from the body and breath (NORD, 2024; O'Connell et al., 2022). Many children have difficulty consuming enough calories to support their nutrition due to insatiable thirst resulting from kidney defects, and frequent vomiting. Placement of a gastronomy tube (G-tube) or jejunostomy tube (j-tube) may therefore be recommended to support nutrition and the administration of medication (Cystinosis Research Network, 2018). Complications of cystinosis can include extreme light sensitivity (photophobia), hypothyroidism, heat exhaustion due to impaired sweating, short stature, and muscle weakness (O'Connell et al., 2010; Nesterova & Gahl, 2013; Wilmer et al., 2011). People with cystinosis will develop end stage renal disease (ESRD) where dialysis and kidney transplantation are required, however, early treatment combined with strict adherence can delay ESRD (Ariceta et al., 2019; Nesterova & Gahl, 2013; O'Connell et al., 2010). While essential research into the medical treatment of cystinosis continues, attention has also shifted towards the psychosocial impact of living with the disease. A participatory multiphase design study by Somanadhan et al. (2020) identified the psychosocial

impact of living with a rare disease as a top research priority for people with rare diseases in Ireland.

School Experiences and Cystinosis

While research indicates that children and young people with cystinosis may be at risk of social, emotional, and behavioural difficulties (Aly et al., 2014; Ariceta et al., 2015; Delgado et al., 2005; Spilkin & Ballantyne, 2007; Ulmer et al., 2009), this research is limited in its ability to contextualise these risks in order to provide a detailed understanding of strengths and needs, particularly in relation to their education and school experiences. Schools provide a setting for children that may exacerbate or alleviate challenges to children's mental health and wellbeing (Baker & Maupin, 2009; Runions et al., 2020; DES, 2019). Despite research showing that children with chronic and rare health conditions are at risk for adverse school experiences (Adama et al., 2023; Foster et al., 2022; Lum et al., 2017) very little is known about the school experiences of children with cystinosis.

Wellbeing is challenging concept to define as there is no universally accepted definition (Svane et al., 2019; Tynan & Nohilly, 2021). It is a broad concept that has been noted to include many subsets such as emotional wellbeing, social wellbeing, psychological wellbeing and mental health (Svane et al., 2019). In Ireland, the Department of Education and Skills (DES) has also adopted this multidimensional conceptualisation of wellbeing in the Wellbeing Policy Statement and Framework for Practice (DES, 2019). This policy also highlights that wellbeing is a fluid concept that is experienced at a personal level and is associated with a range of risk and protective factors operating across various contexts. The World Health Organisation defines mental health as a "state of mental wellbeing that enables people to cope with the stresses of life, to realise their abilities, to learn well and work well and to contribute to their communities" (WHO, 2022, p.8). This conceptualisation suggests that positive mental health goes beyond the absence of mental disorders and that mental health is an integral component of wellbeing.

Positive school experiences such as a sense of school belonging and connectedness, opportunities for success, positive relationships with teachers and peers and systems that support children at times of difficulty are important protective factors for children's wellbeing (Baker & Maupin, 2009; DES, 2015; 2019). Conversely, adverse experiences such as relationship and peer difficulties,

absenteeism, alienation and disengagement are potential risk factors for positive school experiences and overall wellbeing. Shedding light on the experiences of children with cystinosis in the school system and identifying potential facilitators and barriers present is an essential step in promoting positive school experiences and protecting and enhancing wellbeing.

Cystinosis and School Experiences: A Bioecological Framework

Bronfenbrenner's bioecological theory emphasises the influence of interactions that occur within a system and the role of four factors on children's development: Person, Process, Context, and Time (PPCT) (Bronfenbrenner & Morris, 2006). The PPCT model emphasises the dynamic relationships between each of these factors and highlights the complex nature of child development and the importance of considering both immediate and distant factors on child development over time. The PPCT model provides a useful framework for contextualising the school experiences of children with cystinosis. Within the model, "Person" refers to the individual, in this case the child with cystinosis, and their individual characteristics. These characteristics are divided into three categories: demand (age, gender, race etc), resource (intelligence, health, housing etc), and force (temperament, resilience, motivation etc.) (Tudge et al., 2008). Process refers to what Bronfenbrenner terms "proximal processes", the interactions between the developing person and the people, objects, or symbols in their environment. In the current study, "Process" contextualises the school context and interactions between teachers and children, between children and peers, between children and parents, and between the child and school activities such as sports, academic tasks, and play. These proximal processes are considered the primary mechanism of child development. In order to be effective, they must occur on a regular basis, over an extended period of time and become increasingly complex over time (Bronfenbrenner & Morris, 2006; Hayes, 2017). While Bronfenbrenner focused primarily on the role of proximal processes in positive developmental outcomes, researchers have highlighted that where negative interactions occur regularly over time, these may be considered "inverse proximal processes" that contribute to adverse outcomes or dysfunction (Merçon Vargas et al., 2020). This expanded view of proximal processes highlights the importance of identifying facilitators and barriers to children's positive school experiences. Given the synergistic nature of the PPCT model, proximal processes

must be understood in relation to their relationship and interdependence with other factors, that is, person, context, and time (Merçon Vargas et al., 2020). In relation to the aforementioned person characteristics, Tudge et al. (2008) note that individual person characteristics impact proximal processes as they determine expectations placed on a person. In the context of cystinosis, the individual physical impact of the condition (e.g. side effects of medication, personal characteristics of the condition), age, and resilience may impact the proximal processes at play. Similarly, children's ability to take part in school-based activities and interactions may be impacted by their condition, for example, muscle weakness, fatigue, and light sensitivity may limit physical and outdoor activities.

"Context" refers to four interrelated systems. The microsystem consists of the person's immediate environment and is the context within which proximal processes occur. Relevant microsystems include the classroom or school, the family, and peers. The mesosystem refers to the inter-relations between two of the child's microsystems, for example, the interrelations between home and school, or between hospital and school. Positive or negative interrelations in the mesosystem can indirectly impact proximal processes at play within a child's microsystem. For example, a lack of communication between health-care providers and school may result in teachers not having the required information to support a child's medical or therapeutic needs. The exosystem includes processes that have an indirect influence on the child through their influence on a person in the child's microsystem (Runions et al., 2020). In the context of the current study, this could include access to professional development for school staff and school policies. The final system, the macrosystem, refers to the shared culture, norms, policies, and values that influences other factors (El Zaatari & Maalouf 2022; Lehman et al., 2017). Factors such as Department of Education policies, and availability of allied health care supports and staff all indirectly impact the child's experiences in school.

The final component of PPCT, "Time", relates to the influence of changes over time on the dynamic interactions at play (Bronfenbrenner, 1994; El Zaatari & Maalouf, 2022). "Time" within this theory highlights the importance of flexible, individualised, and responsive support for children with cystinosis. Bronfenbrenner specified three distinct ways in which time impacts proximal processes during development. Microtime refers to what is happening during a specific activity or

interaction. Meso-time refers to the consistency of interactions and activities. For children with cystinosis, absenteeism due to ill health and hospitalisations may interrupt the consistency of proximal processes over time. Macro-time refers to the broader changing events and expectations in wider society, or the macrosystem, that take place as a child reaches a particular age (Bronfenbrenner and Morris, 1998; Hayes, 2017). Internationally changing societal beliefs have contributed to an emphasis on promoting inclusive practices for all children in schools. In 2021, for example, a united nations (UN) resolution “addressing the challenges of persons living with rare diseases and their families” was adopted. This focuses on the importance of non-discrimination and promotion of the rights of everyone living with a rare disease, including within the education system. For children with cystinosis, these cultural shifts within the macrosystem may impact proximal processes as the broader community strive to understand their experiences and advocate for change in policy and practice. Macro-time also includes specific times in a child’s life such as transitions in the school system and developmental stages (e.g. adolescence) as events that occur over time in the environment and influence the developing child’s experiences (Hayes, 2017). For children with cystinosis, individual symptoms can also change significantly over time given the progressive nature of the condition as children’s needs will vary between times of relative consistency and critical periods of increased need.

Research Focus

The importance of understanding the psychosocial impact of living with rare diseases has been emphasised in recent policy documents such as the National Rare Disease Plan for Ireland 2014-2018 (Department of Health, 2014), international reports (Kole et al., 2021) and research (Somanadhan et al., 2020; Verger et al., 2020). For children with cystinosis, while research indicates that they are at risk of social, emotional, and behavioural challenges, a predominantly quantitative research methods focus makes it difficult to contextualise these risks. Children’s experiences in school are integral in promoting positive mental health, quality of life, social relationships, and overall wellbeing (DES, 2019; Lum et al., 2017; Runions et al., 2020). Previous research has shown that children with rare and/or chronic health conditions are at risk of adverse school experiences (Runions et al., 2020; Foster et al., 2023; Lum et al., 2017). Despite this, little is known about the school experiences of children with cystinosis. A better

understanding of the psychosocial strengths and needs of children with cystinosis and identification of the factors that influence their school experiences is integral in informing practices and policy.

The research presented in this doctoral thesis formed part of a broader study, funded by Cystinosis Ireland, that aimed to identify the psychosocial and cognitive impact of cystinosis for children and young people the condition. This thesis consists of two papers: firstly a systematic literature review (SLR) which explores the existing research pertaining to the psychosocial and educational experiences of children and young people with cystinosis and, secondly, an empirical journal article (EJA) on a qualitative study of the perceptions of parents and school staff on the psychosocial strengths and needs of children with cystinosis and the potential factors impacting their school experiences. Given the pivotal role of school in children's psychosocial development and wellbeing, it is hoped that the insights gained from this doctoral research will provide guidance to practitioners within educational psychology and related professional fields regarding how to best promote positive school experiences for students with cystinosis, and also highlight important considerations for practice and policy.

Significance of the Research

The findings of this doctoral research have significant implications for practice, research, and policy. People with rare diseases, their families, and the broader community have emphasised the need for broader awareness of the implications of living with rare diseases (Kole et al., 2021; Somandahan et al., 2020). Wellbeing is at the forefront of educational policy (e.g. DES, 2019). Care and support for people with rare diseases has been identified as a priority at a national (DoH, 2014) and international (Kole et al., 2021) level.

The findings of the SLR suggested that there is limited research pertaining to the psychosocial and educational experiences of children with cystinosis. The available research is primarily quantitative and lacks theoretical framework to support understanding of the complexities of cystinosis and children's school experiences. Thus, while the available research tells us that children with cystinosis are at risk of social and emotional difficulties, it does little to contextualise these risks and what can be done to mitigate them. The EJA highlights the personal strengths of children with cystinosis, but emphasises the importance of monitoring their needs over time. Results suggest that it is important

for parents, school staff, and allied health professionals such as educational psychologists to be cognisant of challenging periods such as educational transitions, adolescence, and times of increased medical need. Working together to find solutions is essential in promoting and supporting positive school experiences. Participants discussed the importance of collaboration and communication at a child, school, and multidisciplinary level; however they highlighted a lack of multidisciplinary consultation with schools. There is a need for increased knowledge of the impact of cystinosis, particularly for teachers.

Positionality

In line with the constructionist epistemology of this research and the qualitative nature of enquiry, it was important to consider how my own values, beliefs, and experiences shaped how I conducted and interpreted this research (Byrne, 2022; Wilson, 2022). My initial interest in the research topic stemmed from professional experiences. Throughout my professional career I have had the opportunity to support children with complex medical needs, in particular during my six years as a behavioural psychologist with the Central Remedial Clinic. This service provides medical, therapeutic, and educational supports for children with physical disabilities and complex medical needs. I have always been astounded by children's resilience and determination in the face of significant and ongoing challenges. I have worked alongside, and been inspired by, teachers supporting these children; teachers who understand that their role is not confined to supporting children's academic attainment and recognise their pivotal role in a child's social, emotional, and physical development. Often, these teachers work in challenging and under-resourced environments supporting children with a range of complex and varied needs. I believe that teachers and school staff are constantly striving for best practice and advocating for the needs of children in their classroom, but their voices are often unheard. I hoped to address this by including them in my research.

Over the course of my professional training as an educational psychologist (EP), I had the opportunity to expand my knowledge of child and adolescent development and theoretical frameworks and broaden my experience through professional placements (e.g. National Educational Psychological Service [NEPS], Disability Services, HSE Primary Care). During my placement with NEPS, for example, I joined a Chronic Illness Working Group. The group aimed to gain a

greater understanding of the impact of chronic illnesses in school, and the consultative role of NEPS with schools. These experiences have broadened my awareness of the multi-level factors and systems impacting on a child's development and influenced my choice of theoretical framework for this study.

From a personal perspective, I was diagnosed with a rare disease in my early twenties. While I have been fortunate that the symptoms and progression of this condition have been mild, I found myself identifying with some shared experiences most notably, the lack of information and knowledge regarding rare diseases and the increased sense of concern and apprehension at times of transition. However, I was also mindful of the diversity of experiences and the importance of portraying the participants' meaning from the current research. Having been diagnosed as a young adult, my condition had no impact on my education or social experiences to that point and I did not have to navigate my childhood or adolescence through this lens. I have reflected numerous times during this research journey that earlier identification may have altered my experiences and the opportunities I have been afforded and the potential this would have had to impact on my own wellbeing.

As a parent to two young children, I also considered my own experiences in relation to those of the parent participants in this study. I cannot possibly fully understand their experiences as parents to children with rare diseases, but as a parent myself I strived at all times to accurately and honestly convey the strengths, needs, and challenges faced by their children, as I would hope someone would for my own.

Background to the Systematic Literature Review

The systematic literature review aimed to synthesise research pertaining to the psychosocial and educational experiences of children and young adults (under 25) with cystinosis. A systematic search of relevant electronic databases was conducted, and eight empirical studies met the inclusion criteria and were analysed using narrative synthesis. The findings indicated that children and young adults with cystinosis are at risk for social, emotional, and behavioural challenges. The review also highlighted several gaps in the current literature. Notably that there was a significant lack of qualitative research and very few studies which referenced the impact of cystinosis on school experiences. None of the studies included data from teachers or school staff supporting children with cystinosis. The

review also highlighted a lack of theoretical frameworks in conceptualising and understanding the psychosocial and educational impact of living with cystinosis.

Rationale

Research in rare disease is often focused on the biomedical implications of the condition and overlooks the psychosocial challenges faced by people with rare diseases (Cohen & Biesecker, 2010; DoH, 2014). People with rare diseases have emphasised a need to highlight and understand the psychosocial challenges they face as a result of their condition (Kole et al., Somanadhan et al., 2022). Medical advances have significantly improved outcomes for children with cystinosis and many people survive well into adulthood. However, the day-to-day challenges of living with the condition are not well understood. For children and young people with cystinosis, the school environment forms a significant part of their experiences. Previous research has shown that children with rare and/or chronic health conditions are at risk of adverse school experiences (Runions et al., 2020; Foster et al., 2023; Lum et al., 2017). By identifying the current literature on the psychosocial and educational experiences of children with cystinosis, it was hoped that the findings would raise awareness for educators, health professionals, and parents in planning and coordinating support for children and young people. The SLR also aimed to identify gaps in the current literature to inform future research.

Research Journey

In the early scoping stages of the review, I was invited to attend the annual Cystinosis Conference in Dublin where I was afforded the opportunity to listen to families' views on areas of need. Families discussed a general lack of understanding within the education system, questions about the physical impact of the condition in school, difficulties accessing support within the education system and concerns regarding children's psychosocial wellbeing. With this information, I decided to focus my review on the psychosocial and educational impact of children and young people with cystinosis. I recall being surprised by the volume of medical research on cystinosis that my search returned, and the lack of research pertaining to wellbeing and psychosocial factors. While I did find that there were some additional articles focusing on the psychosocial implications of cystinosis in relation to adults, given my role as a trainee EP and the emphasis that parents placed on understanding the psychosocial challenges faced by their children, I made the decision to focus on children and young people in the SLR.

Background to the Empirical Journal Article

As stated previously, the current empirical journal article (EJA) formed part of a broader study, funded by Cystinosis Ireland, that aimed to identify the psychosocial and cognitive impact of cystinosis for children and young people with Cystinosis. The study reported in this thesis is a qualitative empirical study which aimed to explore parent and school staff perspectives on psychosocial strengths and needs of children with cystinosis as well as to identify the factors influencing their school experiences. Underpinned by Bronfenbrenner's Process, Person, Context, Time (PPCT) model (Bronfenbrenner & Morris, 2006), the EJA involved semi-structured interviews with parents and school staff (teachers and SNAs) of children with cystinosis. Reflexive thematic analysis (TA) was used to analyse the interview data.

Rationale

For children with young people with cystinosis, research indicates that they are at risk of social, emotional, and behavioural challenges (Aly et al., 2014; Ariceta et al., 2015; Delgado et al., 2005; Spilkin & Ballantyne, 2007; Ulmer et al., 2009). Despite the fact that most children with cystinosis can attend mainstream school, there is a significant lack of research that explores their school experiences. This is significant given the integral role that schools play in supporting children's wellbeing (DES, 2019; Runions, 2020). The majority of the current research on cystinosis utilises quantitative methodologies which may be limited in gaining deeper insights into the psychological, emotional and social impact of rare diseases (Lippe et al., 2020). Adopting a qualitative paradigm, the current research aims to add deeper understanding of the experiences and processes at play that impact on wellbeing for children with cystinosis in the context of school.

Research Journey

My interest in the area of rare disease was influenced by both professional and personal circumstances. When I first started this journey, I was initially interested in researching the resilience of parents of children with rare disease. This interest initially stemmed from my reflections on the resilience of parents of children that I had supported through my professional work. At this time, Cystinosis Ireland, the national advocacy group for people with cystinosis in Ireland engaged with staff on the doctoral programme at UCD to discuss the need for research into the psychosocial experiences of people with cystinosis. As I

started to engage more deeply in the research process and, importantly, as I heard from parents and advocates of children with cystinosis, it became clear that it was the psychosocial experiences of children that mattered most to families. I had hoped initially to focus more broadly on both the family and school contexts, however, the more I learned about cystinosis and wellbeing in general it became apparent that this was too broad. Given the lack of research focusing on the school environment and the relevance of this to educational psychology practice, I decided to narrow the research focus to the school context. This decision was also influenced by my experience during my placement in NEPS, where I joined a newly formed Chronic Illness Working Group. During meetings, psychologists discussed the complexities of supporting children with chronic illness and rare diseases in schools, the lack of resources for teachers, and the lack of clarity regarding the roles of different services (i.e. NEPS, CDNT, Primary Care) in supporting these children in school, further strengthening my desire to focus my research on school contexts.

I had hoped to include the voice of children with cystinosis in my research. As noted previously, the current research forms part of a wider research project being conducted by UCD with Cystinosis Ireland which will also explore the cognitive profiles of children with cystinosis. As part of the larger research project, a researcher met with children to conduct psychoeducational assessments. Reflecting on this, I was cognisant that children with cystinosis and their families interact with multiple professionals as part of their overall care. Due to the low prevalence of cystinosis, people with cystinosis are often asked to take part in research. Cystinosis Ireland stressed the importance of valuing families' time and being mindful of the children's energy levels and overall health when planning research. Similarly, in a survey by Rare Disease Europe, people with rare diseases noted the importance of developing a positive relationship with researchers, not wanting to be considered only as research subjects (Eurodis, 2018). Rather than including children in my study, the decision was made for the researcher conducting the psychoeducational assessments to also interview children at this time. This was deemed to be a more efficient use of children and families time and limited unnecessary exposure of children to additional testing by unfamiliar professionals.

Reflections

Gibbs reflective cycle (1988) was adopted to guide my professional and personal learning throughout my research journey. Gibb's cycle consists of six distinct stages: Description, Feelings, Evaluation, Analysis, Conclusion, and Action Plan. The individual is prompted to examine their experiences at each stage to incite deep and critical reflection. The completion of this thesis, while at times challenging, has been a positive, rewarding experience. As I reflect on the entire research experience, I feel that I have developed analytical skills and competencies that will be very valuable to my professional practice and role as a reflective scientist practitioner.

When I reflect on the parent interviews, I feel that the skills that I gained through my supervised professional placements were extremely valuable in guiding the interviews and helping to develop a rapport with participants. In the early stages of my training I would have been nervous discussing sensitive and emotive topics with parents and teachers. However, I recall feeling confident in my ability to respectfully guide participants through these interviews. My ability to guide participants through the interviews was also strengthened by preparation, which is an important consideration for professional practice. Having completed the SLR, attended conferences on cystinosis, researched the condition, and developed a thorough understanding of the theoretical framework, I felt I was well equipped to guide the semi-structured flow of the interview, allowing flexibility but maintaining the focus of the interview on the research topic.

It is crucially important that EPs are committed to promoting current evidence-based practice. The research process has also enhanced my competency as a researcher-practitioner. I have learned valuable skills in critically evaluating research, searching literature, and synthesising available research which I plan to utilise throughout my career. Regular supervision was essential to the research process. At times, the complexities of the research and the topic could feel overwhelming. I found that regular supervision allowed a space for me to consolidate my thinking, which in turn was supportive for my overall wellbeing and in reducing stress. Supervision also enhanced my practice in reflexivity through opportunities to reflect on how my training, values, and experiences impacted the research process.

Overall, the completion of this thesis has been of great benefit to my research, professional, and personal development. Completing this research has

also strengthened my passion for supporting children and families with complex medical conditions and I hope to be able to bring the skills that I have learned into my practice as an EP.

Dissemination of Findings

A poster presentation on the findings of the SLR was provided at the Cystinosis Ireland Conference in 2023. This involved a 10-minute oral presentation followed by in-depth questions to a panel of international medical experts as well as adults with cystinosis and their parents and families. At the 2024 Cystinosis Ireland Conference, an oral presentation on the findings of the empirical study was given to a national and international audience which included the key international researchers in the field of cystinosis. The findings of this study, along with the findings of the broader research study, will be presented to Cystinosis Ireland in September 2024 and a summary document using infographics will be shared with all parents through Cystinosis Ireland.

Both the SLR and the EJA will be submitted for publication in educational psychology journals such as Educational Psychology in Practice and Educational and Child Psychology as well as transdisciplinary journals such as the International Journal of Environmental Research and Public Health. I also plan to share my findings with the NEPS Chronic Illness Working Group. I hope to disseminate knowledge from this study through my own professional practice and career as an EP. This research has strengthened my passion for supporting children with complex medical conditions and spreading awareness of the importance of wellbeing through school experiences for children and young people managing medical conditions.

Summary

This chapter provided an overview of the rationale for the current research in relation to the psychosocial strengths, needs, and school experiences of children with cystinosis. An overview of the two research components, the SLR and the EJA, was provided. This summarised the background and rationale for the two studies along with a description of my personal research journey and reflections on this process. The SLR provided in the next chapter provides an overview of the current research pertaining to the psychosocial and educational experiences of children with cystinosis. In chapter three, a detailed description of the research methodology for the EJA is presented. The EJA, which consists of a

qualitative exploration of parent and school staff perspectives of the psychosocial strengths and needs of children with cystinosis and the factors influencing their school experiences is outlined in chapter four. Finally, chapter five describes the implications for practice that were identified from the findings of these studies.

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Definition of Key Terms

Cystinosis: Cystinosis is a rare, multisystem disorder that causes the accumulation of an amino acid (cystine) in different tissues and organs in the body. It has an occurrence rate of approximately 1 in 200,000 people.

Rare Disease: Rare diseases are diseases or disorders with an occurrence rate of 1 in 2000 people.

Chronic Illness: A chronic illness is a long-term health condition that may not have a cure.

Wellbeing: Wellbeing it is a broad multidimensional concept that can include emotional wellbeing, social wellbeing, psychological wellbeing, and mental health. In Ireland, the Department of Education and Skills (DES, 2019) describes wellbeing as being present when a person realises their potential, is resilient in dealing with the normal stresses of life, takes care of their physical wellbeing and has a sense of purpose, connection and belonging to a wider community. It is a fluid way of being and needs nurturing throughout life.

Chapter 2: Systematic Literature Review

Psychosocial and Educational Experiences of Children and Young Adults with Cystinosis: A Systematic Literature Review

Abstract

While much research focuses on the medical treatment of cystinosis, less is known about the psychosocial impact of living with the condition. The purpose of this systematic review was to synthesise the current research regarding the psychosocial impact of cystinosis on children and young adults (under 25), particularly in relation to school experiences. The review was conducted in accordance with best practice guidelines and the Preferred Reporting Items for Systematic Reviews (PRISMA). A systematic search of relevant electronic databases including SCOPUS, APA PsycArticles®, APA PsycInfo®, Medline, Australian Education Index, and ERIC was conducted. Screening and data extraction resulted in eight studies that met the inclusion criteria and were quality rated using the CASP checklists. Due to the heterogeneity of the included studies, a narrative synthesis was conducted.

Overall, research indicates that children with cystinosis are at risk for social and emotional difficulties. Very few studies focused directly on the educational experiences of children with cystinosis and how this may have been a factor in their wellbeing. Importantly, none of the studies included information from school professionals. Studies highlight the need for multidisciplinary support to address the broader needs. Findings are relevant for families and those living with cystinosis, policy makers and education and healthcare practitioners.

Keywords: behaviour, children, cystinosis, education, psychosocial, school, wellbeing, young adult

Introduction

Rare Diseases Ireland (2024) defines rare diseases as a disease or disorder when it affects less than 1 in 2,000 individuals. Cystinosis is a rare, chronic, multi-system disorder which causes an accumulation of the amino acid cystine in the organs and tissues of the body, causing widespread tissue and organ damage (Doyle & Werner-Lin, 2015; National Organisation for Rare Diseases [NORD], 2023). Current estimates indicate that cystinosis occurs in approximately 1 in 200,000 live births within developed countries (Cystinosis Ireland, 2024). The condition is characterised by a demanding life-long treatment regime and frequent medical interventions and hospital appointments. Although cystinosis was once considered an exclusively paediatric condition, medical advances have prolonged survival for people with cystinosis to over 40 years of age (Ariceta et al., 2016; Emma et al., 2021; Van Stralen et al., 2011). Management of the condition is complex, and requires treatment with several drugs with a strict dosage schedule (Ariceta et al., 2016; Ariceta et al., 2019). A primary treatment for cystinosis is cysteine depletion therapy (Cysteamine) which slows the development and progression of kidney damage and enhances physical growth (NORD, 2024). Cysteamine has an unpleasant smell and taste and side effects include nausea, vomiting, gastric reflux, and an unpleasant odour from the body and breath (NORD, 2024; O'Connell et al., 2022). Many children have difficulty consuming enough calories to support their nutrition due to insatiable thirst resulting from kidney defects, and frequent vomiting. Placement of a gastronomy tube (G-tube) or jejunostomy tube (j-tube) may therefore be recommended to support nutrition and the administration of medication (Cystinosis Research Network, 2019). Complications of cystinosis can include extreme light sensitivity (photophobia), hypothyroidism, heat exhaustion due to impaired sweating, short stature, and muscle weakness (Nesterova & Gahl, 2013; O'Connell et al., 2010; Wilmer et al., 2011). People with cystinosis develop end stage renal disease (ESRD) where dialysis and kidney transplantation are required, however, early treatment combined with strict adherence can delay ESRD (Ariceta et al., 2019; Nesterova & Gahl, 2013; O'Connell et al., 2010).

While much of the research focuses on the medical treatments for people with cystinosis, less is known about the psychosocial and educational experience of living with the condition. Although children and young people with cystinosis

face many of the same challenges as others in navigating educational and social situations, challenges are further exacerbated when managing a rare condition. It is important that these additional challenges are identified in order to provide the appropriate social, emotional, and educational support for young persons with cystinosis and their families. Researchers have noted that a focus on biomedical aspects of treatment often dominates research for chronic and rare conditions (Elliot & Richardson, 2014; Foster et al., 2021; Pickles, 2018). It has been suggested that this narrow focus results in care and support which can be disconnected from the needs of the person (Pickles, 2018). While there is a dearth of research in the area of psychological, social, and educational experiences for children with cystinosis, the broader literature from other chronic illnesses and rare diseases offers some insight (Gomez et al., 2019).

Challenges associated with the burden of medications and managing physical symptoms and side-effects appear to impact on the daily experiences of many people with medical conditions. In a study examining experiences of adults with cystinosis, Beinart et al. (2015) noted that participants emphasised the physical effects of cystinosis such as short stature, fatigue, absences from work due to ill health. The adverse side-effects of cysteamine, in particular body and breath odour and unpleasant taste, were also reported to impact on relationships, autonomy, and social life. Very little is known about how these challenges impact children's day to day experiences. The aforementioned medical regime required to manage the condition is demanding and complex and may have a significant impact on the day to day experience of children and their families (Doyle & Werner-Lin, 2015). This is particularly relevant during adolescence, when young people begin to take more responsibility for their own medical care (Ariceta et al., 2015b; Doyle & Werner-Lin, 2016). The complex treatment regime and their associated side effects have been reported to cause challenges with adherence amongst the young adult population and result in increased associated concerns for parents (Ariceta et al., 2015b; Doyle & Werner Lin, 2015; Doyle & Werner-Lin, 2016). Transition to adult care has also been identified as an important and challenging time for young people with cystinosis and their families. Some young adults report lapses in their health due to challenges with treatment adherence, changes in care, and a lack of specialist knowledge in adult services (Doyle & Werner Lin, 2015). The physical impact of cystinosis are not confined to the home

for children and young people. Gaining insight into these experiences for school aged children is crucial information for teachers and support staff in planning school activities and educational tasks and providing additional classroom support where necessary.

The importance of social connectedness for people with rare and chronic conditions is widely reported (e.g. Paz-Lourindo et al., 2020, Knudson et al, 2018). However research has shown that children and young adults with rare diseases have to navigate significant social challenges related to their conditions. In a meta-analysis of people's experiences of living with a chronic illness, Eassey et al. (2020) note that people living with chronic illness often report feeling left out by peers due to health related challenges such as missing school days or tiredness. Exploring parental perceptions of the psychosocial impact of rare disease for school aged children, Adama et al. (2023) found that almost half of parents (46%) reported concerns with bullying, including social isolation, cyberbullying, being left out, physical aggression and name calling. Feelings of stigmatisation are also common (Adama et al., 2023; Santos et al., 2016; Witt et al., 2023b) and can result in reluctance to disclose diagnosis (Knudson et al., 2018). While adults with cystinosis have reported being guarded against disclosing their diagnosis to others due to perceived emotional and social risks (Doyle, 2015), there is no research regarding how children and young people manage peer disclosure. While research supports the importance of social support and connection for people with rare and chronic conditions, little is known about this experience for children and young people with a diagnosis of cystinosis. From a psychoeducational perspective, school plays a crucial role in supporting students to develop social relationships and to support children living with cystinosis in feeling part of their community. Verger et al. (2020) report that peer support is integral to feelings of inclusion, particularly when children have missed school due to illness. Gaining insight into the social experiences of children with cystinosis provides valuable information for enhancing social wellbeing.

Living with rare diseases and chronic health conditions is associated with mental health and psychological difficulties (Adama et al., 2023; Cohen & Biesecker, 2010). In a quantitative study of 41 parents of Australian primary and secondary school children with rare diseases (e.g. musculoskeletal/metabolic/genetic diseases), Adama et al. (2023) reported that

43% of parents indicated that their child had mental health difficulties, with 73% noting that these difficulties had been experienced for over a year. In relation to cystinosis, research has evaluated the psychological experience of living with cystinosis from an adult perspective with them describing the need and desire for autonomy and independence while remaining connected to their families (Beinart et al., 2015; Doyle & Werner-Lin, 2015). Similarly, research has shown relatively higher levels of anxiety and depression for adults with cystinosis and people with other chronic illnesses (Beinart et al., 2015; Pao & Bosk, 2011; Zhao et al., 2012). Conversely, Beinart et al. (2015) also noted that some adults with cystinosis report positive personality attributes such as resilience and an enhanced appreciation of life due to their experience of living with cystinosis.

School forms a significant part of children and young people's lives as they begin to develop a sense of identity and belonging through relationships with peers (Ernst et al., 2010; El Zaatari & Ibrahim, 2021). Research shows that children's experience of the education system may be impacted by illness (Foster et al., 2021; Lum et al., 2017; Santos et al., 2016). Factors such as medical and care needs, irregular attendance and prolonged absences due to hospitalisation and illness may mean that young people with chronic illness fall behind their peers (Lum et al., 2017). This may result in increased stress and anxiety as they struggle to keep up with schoolwork. Associated medical needs which impact on school attendance may also have a negative effect on relationships as young people miss out on social opportunities. Santos et al. (2016) reported that substantial numbers of adolescents with chronic illness felt that their disease impacts on participation in school and in leisure activities. Lum et al. (2017) note that children with chronic illness have reported bullying and teasing, concerns about keeping up with peers, and negative attitudes from both teachers and peers. The authors further note reluctance to attend school due to lower sense of confidence and feeling different from peers. Feeling different from peers is a significant concern for many children with other chronic illnesses, such as cystic fibrosis, with children reporting being different as the most stressful daily event experienced (D'Auria, Christian, & Richardson, 1997). Academically, a substantial amount of research has linked chronic illness with poor academic outcomes (Lum et al., 2017). Teachers are often unaware of the complexities of rare chronic health conditions (Runions et al., 2020). A general lack of resourcing, training, and clear policy on

supporting children with chronic illness may increase challenges for teachers, parents and students alike (Adama et al., 2023; Foster et al., 2021). It is crucial that schools are aware of and understand these challenges and that accommodations and supports are provided accordingly such that children with medical conditions can be provided with the right supports to achieve their potential (Hillard et al., 2015; Verger et al., 2021).

The psychosocial and educational experience of rare diseases such as cystinosis encompasses a wide variety of biological, psychological, and social factors, each of which are integrated into both family and educational contexts. Each category provides both unique and interwoven challenges and experiences. We know that people with chronic illnesses and rare disease report challenges in relation to treatment protocols, side effects of medications, and physical impacts of disease (Beinart et al., 2015; Doyle and Werner-Lin, 2016; Pickles et al., 2018). Despite complex treatment regimes and unpleasant side effects of medication for the treatment of cystinosis, little is known about the psychosocial impact of this for children and young people. From a psychological perspective, the experience of chronic illness and rare conditions has been shown to be related to lack of choice and feelings of disempowerment, as well as higher incidence of depression and psychosocial stress (Eassey et al., 2020; Santos et al., 2016; Zhao et al., 2012). Socially, people with chronic and rare medical conditions report difficulties in disclosing their diagnosis, feeling left out, and report challenges within the education system (Eassey et al., 2020; Knudsen et al., 2018; Lum et al., 2017).

Although the research in relation to chronic illness and rare diseases provides some insight into the needs and experiences of children and young people managing these conditions, cystinosis, like other rare conditions, poses unique and individual challenges and therefore understanding its individual impact is crucial to ensuring appropriate support and intervention.

A Theoretical Framework for Conceptualising Cystinosis: PPCT

Bronfenbrenner's Person-Process-Context-Time (PPCT) (Bronfenbrenner and Morris, 2006) was adopted as a framework for this systematic review as this theory recognises the complex and interacting factors that can impact on a child's development, across contexts and over time. Four interrelated factors are defined within the theory Person, Process, Context and Time. In the context of the current review, person refers to the child or young adult with cystinosis, recognising their

unique psychological, social, emotional, and physical strengths and needs in the context of a rare medical condition. Process refers to reciprocal relationships for example, relationships with peers or teachers. Contextual factors are considered in terms of the microsystems included (e.g. home, school). Broader contextual factors meso and exo and macro system are considered in terms of implications for practice. While the final component, “Time”, may have been referenced by including studies that measured psychosocial factors for adults, the purpose of this review was to synthesise the research regarding the psychosocial impact of cystinosis on children and young adults (<25) and therefore this factor was not included.

Objectives

Research has illustrated that the impact of living with a rare condition reaches beyond the physiological implications. Cystinosis is a rare, life-limiting condition that requires invasive treatment and is likely to impact on many aspects of children's wellbeing. The purpose of this review was to synthesise the research regarding the psychosocial impact of cystinosis, particularly as it relates to education and the school years. It is hoped that by collating and examining the current research, opportunities to inform current policy and practice and gaps in current knowledge will be identified. This will be addressed by answering the following four research questions:

1. What are the psychosocial implications of cystinosis for children and young adults? Psychosocial influences included those that were deemed to have a psychological and/or social impact, for example, mood, friendships, community involvement and social connectedness etc.
2. What does current research tell us about the impact of cystinosis on children’s educational experience? Educational experiences included factors such as school attendance, relationships, teacher and peer knowledge, medical management in school, inclusion and acceptance etc.
3. What theoretical or conceptual perspectives are utilised to inform our understanding of the psychosocial and educational experience of living with cystinosis?
4. What are the implications for psychological and educational practice outlined in current studies?

Search Methods

The review methods were informed by Preferred Reporting Items for Systematic Reviews and Meta- Analysis (PRISMA) statement and guidelines (Page et al., 2021).

Selection Criteria

Inclusion Criteria: Studies were eligible for inclusion if they met the following criteria:

- Studies that reported on psychosocial constructs and on the educational experiences of people with cystinosis.
- The sample included studies that reported on current or retrospective experiences as reported by parents or teachers of children or young adults or patients (under 25) with cystinosis.
- Studies included people with cystinosis and co-occurring conditions and/or intellectual disability or learning difficulties.
- Studies utilising qualitative, quantitative, or mixed methods research design.
- Articles written in the English language.

Exclusion Criteria: Studies were excluded if they met the following criteria:

- Studies that did not include data on psychological, social, or educational constructs.
- Sample included other chronic illnesses or rare diseases, but did not differentiate the experience of cystinosis.
- Studies that included only data for participants aged 25 or above.
- Articles not written in the English Language as resources for translating studies written in additional languages were not feasible.

Search Strategy

A systematic, comprehensive literature search of six databases was conducted to identify published and unpublished studies in the field(s) of psychology and education on psychosocial and educational experiences of children with cystinosis. The following databases were selected as they referenced education or psychology: SCOPUS, APA PsycArticles®, APA PsycInfo®, Medline, Australian Education Index, and ERIC. A search using Google Scholar was also conducted to identify any relevant grey literature. In addition, a manual hand-search was conducted of the reference lists of all the included articles in the review to identify any additional eligible studies. These databases were searched

simultaneously using SCOPUS, ProQuest, EBSCO on the first of January 2023 and again on the 23rd of March 2023 and the 26th of April 2024. The search string was tested and refined using scoping searches which resulted in the identification and inclusion of synonyms for psychosocial experiences, and the exclusion of terms to limit irrelevant studies. Wildcards were used to extend word endings and spelling variations (e.g., vs.; vs.) were automatically included.

Search String

AB, TI, IF ("Quality of life" OR Psych* OR wellbeing OR mood OR anxiety OR social OR peer* OR friendship OR community OR connectedness OR stress OR coping OR behavi?r OR attendance OR teacher* OR inclusion OR acceptance OR Transition*) AND (cystinosis OR "Lysomal Storage Disorder") AND (child* OR adolescen* OR youth OR "young adult" OR adult* OR patient* OR school OR student* OR pupil OR parent* OR guardian* OR teacher* OR educator*)

Screening and Selection Process

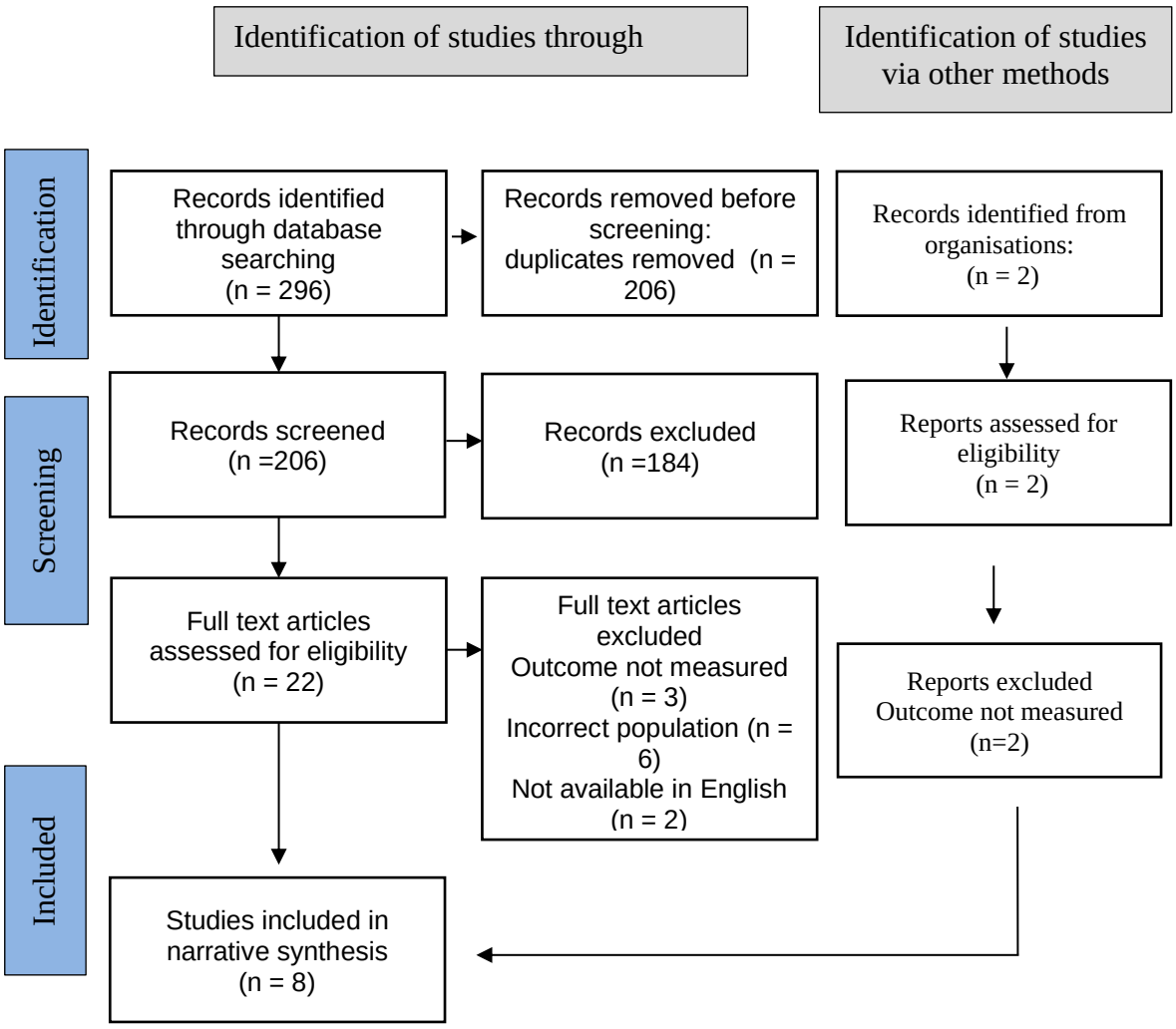
The search returned 296 records for screening against the inclusion criteria. All records were extracted and stored in Endnote reference management software. Ninety duplicate documents were removed. The title and abstracts of the remaining 206 were screened and a further 184 studies were removed. Full texts of the remaining 22 records were reviewed against inclusion criteria. This resulted in the exclusion of 14 studies, the rationale for which is summarised in Table 1.Three studies were not available in English, two studies reported expert recommendations but did not gather data on psychosocial or educational experiences of people with cystinosis, four studies included data outside of the age range, one study reported on the experiences of medical staff only, one study published psychosocial data in a separate paper, one study presented a case study that described the physical presentation of cystinosis, and two studies did not differentiate experience of cystinosis from other diseases. The remaining 8 studies met inclusion criteria. The search process is documented in Figure 1.

Table 1: Reasons for Exclusion of Reviewed Studies

Authors	**Reason for Exclusion**
Ariceta et al., (2016)	Article describes coordination of care from paediatric to adult healthcare teams. No measures of psychosocial wellbeing are included.
Ariceta et al., (2015)	Authors provided recommendations for care, however this did not include data from people with cystinosis, their parents, or their teachers.
Buchanan (2000)	Case study describes treatment and side effects experienced by one young person with cystinosis. No measure of psychosocial experiences
Doyle (2015)	Study did not differentiate between data gathered in relation to children and young people (<25) vs. adults.
Doyle & Werner-Lin, (2015)	Data gathered from adults or parents of adults with cystinosis.
Ehrich et al., (1991)	Details of psychosocial development were published in a separate study which were included in this review (Wolff, 1982)
Henning (1988)	Study reported on quality of life of survivors of end stage renal disease. Seven participants had a diagnosis of cystinosis, however, it was not possible to differentiate the experiences of participants with cystinosis from other participants. Data from adults could not be differentiated from data from children.
Kaufeld et al., (2018).	Study unavailable in English
Kempf & Raimbault, (1974)	Study unavailable in English

Langman et al., (2014)	Using the PedsQL, authors report an improvement in social functioning, school functioning and total function compared to baseline following change in medication. However, it was not possible to determine from results if participants reported significant difficulties with QOL pre or post treatment.
Levtchenko et al., (2022)	Study reported on expert guidance on management including psychological wellbeing; however there was no measure of psychosocial experiences.
Obiagwu et al., (2018)	Did not differentiate experiences of children with cystinosis from those with end stage renal disease.
Winkley (1990)	Study reports on the anxieties and feelings of hospital staff treating a child with cystinosis but does not directly measure the psychosocial or educational impact of the disease for the child.
Witt et al., (2023a)	Literature review on health related quality of life for adults and children with cystinosis.

Figure 1. PRISMA Diagram



Data Extraction

The content of the remaining eight studies was extracted using a data extraction table (Table 2) which was designed specifically for this study. It captured participant characteristics (diagnosis, age range), respondent, as well as first author, year of publication, geographical location, and main findings. Information pertaining to the four research questions: (1) psychosocial factors, (2) educational implications, (3) theoretical background and (4) implications for practice and policy was also collated. Data extraction for each article was performed independently by one reviewer (EM). A second reviewer (JS) cross-checked the extracted data to ensure accuracy and any discrepancies were solved by discussion. Narrative synthesis is an approach to systematic reviews

that relies on the use of words and text to summarise and explain findings (Popay et al., 2006; Snilstveit et al., 2012). Popay et al., (2006) state that narrative synthesis is appropriate for systematic reviews that focus on a wide range of questions and not only those that focus on effectiveness of interventions. A narrative synthesis was chosen due to the considerable heterogeneity among the included studies and the broad research questions guiding the review.

Quality Appraisal Method

All included studies were assessed for quality and risk of bias by two independent reviewers (EM and JS) using the Critical Appraisal Skills Programme (CASP) checklists. The researcher scored each indicator a 1 where the criteria were met, a 0.5 if unsure and a 0 where the criteria was not met. Where no clear information was present in the article a 0 was also scored. Scores for each indicator were combined to give an overall quality score. This quality score was converted to a percentage by dividing the obtained score by the total achievable score. Where questions were not applicable to the study in question, the question was not included in the total achievable score. Once converted, included studies were categorised as scoring high or low according to the CASP criteria. A high score was defined as a score greater than 50% and a low score was defined as a score less than or equal to 50% (Van Der Windt et al., 2000). The results of the critical appraisal were reported in tabular (see Table 2 and Appendix A) and narrative form. None of the studies reviewed were determined to be in the low range.

Overview of the Studies

The search strategy identified a total of eight studies, published between 1982 and 2022, that met the inclusion criteria. Table 2 provides a summary overview of the studies included in the review. Three of the studies were conducted in the United States, two in Egypt, one in Switzerland, one in Spain, and one in Germany. The majority of the studies (n = 6) were quantitative. Sample sizes of included studies ranged from nine (Ulmer et al., 2009) to 186 participants (Delgado et al., 2005).

Demographics

The age range of children with cystinosis across all studies was from 1.5 years to 19.9 years. Ariceta et al. (2015b) grouped participant results into children (<18 years) and adults (>18 years), therefore only the data related to children under 18 years of age is included in this review. All of the studies included parents

and carers as respondents. In one study (Ariceta et al., 2015b) it was not possible to differentiate between the responses of parents versus the children as the results were not separated. One study included children as respondents on psychosocial measures (Ulmer et al., 2009). None of the studies included teachers or school professionals as respondents.

Measures

Three studies used the Achenbach Child Behaviour Checklist (CBCL; Achenbach, 1997) (Aly et al., 2014; Atia et al, 2022; Delgado et al., 2005). The CBCL is a standardised measure that provides parental reports on a child's behaviour. It consists of 120 items that assess internalising (withdrawn, somatic complaints, anxiety/depression, thought problems) and externalising behaviour scales (social problems, attention problems, delinquent and aggressive behaviour). Ulmer et al. (2007) utilised both the CBCL and a quality of life measure (The Netherlands Organisation for Applied Scientific Research Academical Medical Centre Child Quality of Life Questionnaire [TAC-QOL]) which comes in two forms (parent and child) is designed to assess quality of life in children aged 8-15 with chronic diseases across seven scales including physical complaints, cognition, social functioning, autonomy, basic motor function, global positive emotional functioning and global negative emotional functioning. One study (Ballantyne et al., 2013) utilised the Behavioural Rating Inventory of Executive Functioning (BRIEF; Gioia et al., 2000) questionnaire which includes a scale of behavioural regulation reflecting the child's ability to control emotions and behaviour as well as flexibility in thinking. One study (Ariceta et al., 2015b) distributed an anonymous patient/carer survey which consisted of 21 multiple choice questions covering impact of the disease, adherence to cysteamine treatment, monitoring of cysteamine therapy and measures to improve compliance. One study (Spilkin & Ballantyne, 2007) utilised a researcher designed "cystinosis behaviour questionnaire" to provide a narrative account of child characteristics, family adjustment, and school issues. The questionnaire consisted of 10 open ended questions about the child's illness, parental stress and family adjustment, and 13 open ended questions that gathered information on "tantrums" and "mood swings". The final section consisted of 52 statements answered on a 3 point Likert response scale that encompassed child characteristic, family adjustment, school performance and intervention for mental health. In their study,

Wolff et al. (1982) provided an account of children's friendships, relationships, mood, social acceptance, and behaviour obtained during parental interviews and behavioural observations over a period of 22 months (range 4-44 months).

Comparison Groups

Of the eight studies, two (Aly et al., 2014; Ballantyne et al., 2013) utilised control groups consisting of children without diagnosed health conditions. One study included children with chronic kidney disease as a control group (Atia et al., 2022). Delgado et al. (2005) utilised both children without diagnosed health conditions and children with cystic fibrosis as comparison groups. The remaining four studies (Ariceta et al., 2015b; Spilkin & Ballantyne, 2007; Ulmer et al., 2009; Wolff et al., 1982) did not utilise comparison groups. A summary of comparison groups and participant numbers for each study are presented in Table 2.

Table 2. Overview of Included Studies

Study & Location	Condition	Age (years)	Respondents	Data Collection	Psychosocial Factors	Theoretical Framework	Implications for Practice Included?	CASP Quality Score
Aly et al. (2014) Egypt	Cystinosis (n= 13) No Medical Condition (n = 13)	1.5 - 12	Parents/Carers	Quantitative ● CBCL	Cognitive and behavioural profiles	None	Yes	90%
Ariceta et al. (2015b) Spain	Cystinosis (n= 34)	<18 (n= 21) >18 (n= 13)	Patients and/or Parents	Quantitative ● Multiple choice questionnaire	Impact of disease on quality of life	None	None	81%
Atia et al. (2022) Egypt	Cystinosis (n= 20) Chronic Kidney Disease (n = 20) Participants matched for age, set, and CKD stage	Mean 10.97 Mean 9.3	Parents/Carers	Quantitative ● CBCL	Emotional, Social and Behavioural Profiles	None	Yes	95%

Ballantyne et al. (2013). USA	Cystinosis (n = 28) Healthy Controls (n = 24)	8 - 17	Parents (BRIEF)	Quantitative ● BRIEF	Emotional Regulation	None	Yes	90%
Delgado et al. (2005) USA	Cystinosis (N = 64) Typically Developing (n = 101) Cystic Fibrosis (n = 21)	4 - 16	Parents/Carers	Quantitative ● CBCL	Socio Behavioural Characteristics	None	No	95%
Spilkin & Ballantyne (2007) USA	Cystinosis (n = 63)	2 - 17	Parents/Carers	Qualitative ● Cystinosis Behaviour Questionnaire	Child characteristics, family adjustment and school issues	Family Systems Model	Yes	100%
Ulmer et al., (2009) Switzerland	Cystinosis (n = 9)	5.3 - 19.9	Parents/Carers (CBCL, QOL) Children (QOL)	Quantitative ● CBCL ● Quality of Life Questionnaire	Quality of Life Emotional and Behavioural profiles	None	Yes	100%

Wolff et al., (1982), Germany	Cystinosis (n = 12)	8 - 15	Patients and Parents/Carers	Qualitative ● Observations ● Interviews ● Record Review	Friendships, relationships, mood, social acceptance, behaviour	None	No	56%

Systematic Review Results

Findings related to the four research questions were extracted from each study.

Psychosocial Impact

All eight studies measured an aspect of psychosocial experiences e.g. quality of life, social/behavioural difficulties. Results of each study are presented in Table 3. Overall, the data indicates that children with cystinosis are at risk for behavioural, social, and emotional difficulties. Delgado et al. (2005) and Atia et al. (2022) compared children with cystinosis with peers with other medical conditions. Delgado et al. (2005) found that while children with cystinosis were significantly more likely than healthy controls to have challenges related to attention, somatic complaints and aggression, when compared with children with cystic fibrosis they exhibited a significantly higher incidence of social difficulties only. Atia et al. (2022) noted that the mean scores on the CBCL for children with cystinosis were in the clinical range, indicating risk for emotional and behavioural difficulties. However, in this study there was no significant difference in emotional and behavioural problem scores between children with cystinosis and children with chronic kidney disease. When compared with children with no diagnosed health conditions, children and young people with cystinosis have been found to have higher reported levels of total behaviour problems as measured by the CBCL, particularly in relation to social and attention difficulties (Aly et al., 2014; Delgado et al., 2005).

Two studies (Ariceta et al., 2015b; Ulmer et al., 2009) included a quality of life measure. Ulmer et al. (2009) noted that child self-report was in the normal range for six of the seven dimensions measured. Both child and parent reports indicated significantly low positive emotions. However, parents were found to rate their children's quality of life lower than the children, reporting significant impairment in relation to positive emotions, autonomy, social, and cognitive functions. In a study of 34 children (n= 21) and adults (n=13) with cystinosis, Ariceta et al. (2015b) reported that cystinosis led to feeling different for 10% of children with cystinosis and had a negative impact on the social life of 14% of the children. As the study did not differentiate between responses of parents and children with cystinosis it was not possible to ascertain if there was a difference in parent and child perceptions as noted in the Ulmer et al. (2009) study. Conversely, the authors noted that 62% of adults with cystinosis reported feeling different, 31%

required employment leave, 39% expressed professional limitations and 8% referred to compromised social life and rejection. Using the BRIEF questionnaire as a measure of behavioural regulation, Ballantyne et al., (2013) found no significant difficulty in comparison to peers. However, using the researcher designed CBQ with parents of 63 children with cystinosis, Spilkin and Ballantyne (2007) reported that 65% of the parents stated their children with cystinosis had temper outbursts and 36% noted that behaviour was more intense than that of their other children. More than 50% of parents indicated that their child often or sometimes got their feelings hurt, did not enjoy life, and were not self-reliant. While Wolff et al. (1982) reported that parents noted only minor behavioural problems for their children with cystinosis, the authors noted there were some potential signs of psychological difficulties for 9 of the 12 children. However it was unclear how the authors measured or conceptualised these difficulties. The authors further stated that signs of depression appeared to be connected with long hospitalisations and medical treatments. It was unclear from the study how signs of depression was conceptualised.

Educational Impact

Four of the eight studies made reference to the implications of cystinosis on children's educational experiences (Ariceta et al., 2015b; Ballantyne et al., 2013; Spilkin & Ballantyne, 2007; Wolff et al., 1982). Results of each study are presented in Table 3. Ariceta et al. (2015b) noted that 29% of respondents noted that school attendance was negatively impacted by cystinosis whereas 5% reported a negative impact on learning ability. One study (Spilkin & Ballantyne, 2007) measured parental reports of school performance and reported that 30% of parents reported their child with cystinosis (mean age = 8.5 years) had academic difficulties for which they received "special help". There was no further detail of the frequency or type of support children received. Ballantyne et al. (2013) noted that children with cystinosis do not differ from their peers in relation to behavioural regulation (i.e. ability to inhibit, shift between tasks and regulate emotions) but present with more difficulties in metacognitive skills (initiating, planning and organising) than peers. The authors caution that children with cystinosis may be "missed" by school professionals as these difficulties may not be the primary focus of the academic curriculum. In their study with 12 children with cystinosis, Wolff et al. (1982) reported that "most" children functioned well with respect to academic

performance and were well liked by classmates. None of the studies included information from school teachers.

Table 3. Aims & Psychosocial and Educational Findings of Included Studies

Author & Year	Study Title	Aims	Findings: Psychosocial and/or Educational Impact
Aly et al., 2014.	Neurocognitive functions and behavioural profiles in children with nephropathic cystinosis	To assess the cognitive functions and define the behavioural phenotype of children and young people with cystinosis	• Children with cystinosis experienced significantly more behavioural problems (withdrawn, anxious, social problems) than the control group of children without a medical condition
Ariceta et al., (2015b)	Cysteamine (Cystagon) adherence in patients with cystinosis in Spain: successful in children and a challenge in adolescents and adults	To evaluate adherence to cysteamine treatment in a group of patients with cystinosis in Spain in an attempt to identify potential therapy pitfalls and improve overall care of affected individuals.	• Cystinosis was reported to have a negative impact on children's school attendance, (29%), "learning ability" (5%) and social life (10%). • 10% of participants noted that cystinosis results in children "feeling different"
Atia et al., 2022	Cognitive functions and behavioural profiles in children with cystinosis treated with cysteamine and correlation with treatment duration.	To characterise neurobehavioural and neurocognitive changes in children with nephropathic cystinosis and find their temporal relation to cysteamine treatment	• Results of CBCL for children with cystinosis found that: o 85% of children had internalisation symptoms o 75% had somatic complaints o 70% had withdrawn behaviour o 50% were anxious or depressed o 95% of children had a total mean score in the significant range

			• There was no significant difference between children with cystinosis and children with chronic kidney disease on any of the scales.
Ballantyne et al., 2013	Executive function in nephropathic cystinosis	To study executive function in children and adolescents with cystinosis	• The cystinosis and control groups did not differ significantly in terms of behavioural regulation as assessed by the BRIEF. Parents rated the cystinosis group has having significantly more difficulty in on the metacognitive scales compared with control.
Delgado et al., 2005	Behavioural profiles of children with infantile nephropathic cystinosis	To define the behavioural phenotype of children with cystinosis	• There was a significant difference between the cystinosis group and control group on the following scales: Total Problems Summary Scale, Internalising Problems Summary Scale, Social Problems, Somatic Complaints, Attention Problems, Thought Problems and Aggression. • Comparisons between the cystinosis group and CF group revealed that only the social problems scale significantly differed between the two chronic disease groups.
Spilkin & Ballantyne, 2007	Behaviour in children with a chronic illness: A descriptive study of child characteristics, family adjustment and school	To describe behaviour and adjustment in children with cystinosis and their families	• Parents reported numerous family stresses as result of caring for a child with cystinosis • 36% of parents believe their child with cystinosis displays behaviour that is more intense than other children

	issues in children with cystinosis		• 65% of parents report their child with cystinosis experiences occasional mood-swings and outbursts of temper • More than 50% of parents indicated that their child often or sometimes get their feelings hurt, do not enjoy life, and are not self-reliant • 30% of parents report their child often experienced academic difficulties • 80% of children rarely or never see a therapist for support with social or emotional issues
Ulmer et al., 2009	Intellectual and motor performance, quality of life and psychosocial adjustment in children with cystinosis	To assess intellectual performance, motor performance, quality of life and psychosocial adjustment in children and adolescents with cystinosis	• Self-reported QOL was normal on all dimensions apart from positive emotions which was significantly impaired. • Parents reported significantly lower positive emotions, autonomy, social and cognitive functions for children with cystinosis
Wolff et al., 1982	Psychosocial and intellectual development in 12 patients with infantile nephropathic cystinosis	To investigate the psychosocial and intellectual development of 12 children with infantile nephropathic cystinosis longitudinally	• Parents indicated that the children were socially accepted by their peers • Researchers noted signs of potential psychological difficulties however it was unclear how signs were measured or conceptualised • Most children with cystinosis were reported to be functioning well academically

Theoretical Perspectives

Of the eight studies, only one (Spilkin & Ballantyne, 2007) referenced theoretical perspectives. The authors framed the psychosocial impact of cystinosis within the family systems-illness model (Rolland, 1994).

Implications for Practice

Five of the studies made reference to implications for practice (Aly et al, 2014; Atia et al, 2022; Ballantyne et al., 2013; Spilkin & Ballantyne 2007; Ulmer et al. 2009). Implications for practice identified in each study are summarised in Table 4.

Table 4: Implications for Practice in Included Studies

Author & Year	**Implications for Practice**
Aly et al. (2014)	The authors suggest the approach to management should be multidisciplinary and include psychological support in order to formulate comprehensive plans for social and educational support.
Atia et al. (2022)	Physicians should be aware of neurocognitive and behavioural difficulties and utilise tools for assessment to provide multidisciplinary management.
Ballantyne et al. (2013)	The authors highlight a need to design appropriate proactive interventions in relation to executive functioning which may be essential to wellbeing and independence. Reference is made to the impact of executive functioning on school performance and its role in treatment adherence.
Spilkin & Ballantyne (2007)	The authors note that the majority of the focus in care is in medical management. Despite difficulties with behaviour, family adjustment, and academic areas, 80% rarely see a therapist suggesting that the psychological treatment of child and family may be ignored. The authors further postulate that teachers may view children's difficulties as a symptom of illness and therefore be less likely to provide onward referral.

	The authors suggest a variety of potential supports including psychological and behavioural support, social support and support in the development of disease management routines and treatment adherence.
Ulmer et al. (2009)	Results indicated both internalising and externalising difficulties. The authors suggest that the extent of difficulties and complaints reflects the profound effect of cystinosis and underlines the importance of patients receiving adequate supportive and comprehensive treatment.

Quality Appraisal

None of the studies included were judged as being of low methodological quality. The total quality score of all studies ranged from 56% - 100%. Quality scores were calculated by generating an overall quality score based on the CASP criteria and converting this to a percentage by dividing the obtained score by the total achievable score. Where a question was deemed not applicable to the study in question, this was not included in the overall quality rating calculation. A high score was defined as a score greater than 50% and a low score was defined as a score less than or equal to 50% (Van Der Windt et. al., 2000). All studies included a clearly defined focus and each was determined to have used an appropriate means to address the research question. The majority of studies used an appropriate measure of psychosocial constructs (e.g. standardised measure of social-emotional and behavioural presentations, quality of life, parental interviews). Two of the studies provided very limited information on the content of the questionnaires/interviews used. Results of five studies were consistent with the findings of other studies and the results of one study partially aligned with previous studies. The results of one study did not align with previous studies, however, this was the earliest identified study that explored the psychosocial impact of cystinosis for children and it is likely that the experiences of these children was different to that of children in the other studies. Five of the included studies referenced potential confounding factors such as socioeconomic status of parents, individual differences in symptoms of medical conditions, concerns with

quantitative measures, use of non-standardized measures, and impact of parental strain on perspectives.

Discussion

The aim of this review was to systematically identify, synthesise and quality appraise research regarding the psychosocial and educational impact of cystinosis on children and young people alongside information on theoretical models and implications for practice and policy. Eight studies met the inclusion criteria and were quality appraised using the Critical Appraisal Skills Programme checklists. The systematic review highlighted that the research is limited, and the findings somewhat inconsistent.

Psychosocial Implications of Cystinosis

For the most part, research indicates that children with cystinosis are at risk for behavioural problems and difficulties with social adjustment (Atia et al., 2022, Delgado et al., 2005; Spilkin & Ballantyne, 2007; Ulmer et al., 2009). Delgado et al. (2005) noted that while children with cystinosis experienced more problems related to attention, somatic complaints, and aggression than those without cystinosis, they differed from children with other chronic illnesses in relation to social problems only. Conversely, Wolff et al. (1982) found that parents reported only minor behavioural difficulties and that children with cystinosis were generally well liked. However, given that this study was published in 1982, the treatment regime and educational experiences at the time of this study were very different to those experienced by children and young people with cystinosis today. Nonetheless, this qualitative account gives insight into the psychosocial strengths of children with cystinosis. Ulmer et al. (2009) examined health related quality of life as reported by parents and children with cystinosis (N=9). While both parents and children reported lower levels of positive emotions, parents also rated children's autonomy, social, and cognitive functions as being low.

It is interesting to note that all but two studies (i.e. Spilkin & Ballantyne, 2007; Wolff et al., 1982) utilised quantitative measures (e.g. Achenbach Child Behaviour Checklist [CBCL], TACQOL) and research designs. Although measures such as the CBCL are widely used in research and professional practice to provide a measure of emotional and behavioural adjustment, some researchers have cautioned their use in paediatric illness populations. Perrin et al. (1995) noted specific concerns with regard to differences in physical symptoms and social

opportunities that may be experienced by children with rare diseases such as cystinosis. The authors caution that several items that reference physical symptoms may be a symptom of their medical condition rather than indicating psychological differences. As noted by Perrin et al. (1995), there is less opportunity to clarify responses in large scale research than there may be in a clinical context. Of particular importance in the context of childhood medical conditions is the use of CBCL in assessment of social competence. Perrin et al. (1995) cautions that the items on this scale assess children's participation in social activities which may be limited by virtue of the child's illness but should not be interpreted as indicating that the child is less socially competent. The closed-end nature of the questions on such measures may result in the loss of rich, person focused information which can only be gathered by discussion with those with more direct knowledge of the experiences of children with cystinosis as could be obtained in qualitative approaches. Spilkin and Ballantyne (2007) addressed this through the addition of open-ended questions on their researcher designed parental questionnaire. Acknowledging the limitations of their study, however, the authors note that gathering participants' narratives through questionnaires rather than in depth interviews may have limited the insights that they gathered.

Data analysis in the qualitative studies was primarily narrative or descriptive rather than guided by researched or established forms of data analysis (e.g. thematic analysis). It is also important to acknowledge that there have been significant advances in medical treatment and educational practices since Wolff et al.'s 1982 qualitative study. Therefore the results of this study may not be relevant to children with cystinosis today. Spilkin and Ballantyne (2007) note that while their study is limited in terms of data collection methods, it is a valuable step in understanding and illustrating the day to day complexities of life for children with cystinosis and their families. Further in-depth qualitative research will add to our understanding of children's strengths and challenges experienced and is essential in providing a holistic picture of support needs. In the context of the potential psychosocial and educational impact of cystinosis, while quantitative research has been essential to illustrate whether a challenge exists, qualitative research can complement this by exploring the complex processes underlying why challenges exist and crucially, the opinions and voices of people with direct knowledge of the children's strengths and needs (Pope & Mays, 1995).

Educational Implications of Cystinosis

While some research in this systematic review refers to factors that are likely to impact on educational experiences (for example, needs related to attention or aggression), very few specifically evaluated how this translates into the school environment. This is significant given that research has shown that children with chronic and rare illnesses report negative school experiences and poorer academic outcomes (Foster et al., 2021; Lum et al., 2017). Ariceta et al. (2015) noted that school attendance is negatively impacted by cystinosis; however it is unclear how exactly this absenteeism impacts on academic or psychosocial experiences for the participants. Spilkin and Ballantyne (2007) measured parental opinion of academic difficulties, with 43% of parents reporting that their child sometimes or often had academic difficulties for which they required help. There was no further detail regarding the types of academic difficulties or the level of support received. While 65% of parents in the study reported that their children had occasional outbursts of temper or severe mood swings, parents reported children rarely had behavioural difficulties in school. Wolff et al. (1982) reported that parents noted their children were “ambitious” with regard to school performance, socially accepted, and presented little resistance to homework. However this should be interpreted with caution as the children in this study likely had very different educational, social and medical experiences to children with cystinosis today. Importantly, none of the studies include data from teachers, despite the significant role they play in children’s school educational outcomes and mental health and the importance of communication and relationships between parents and school personnel (Hinton & Kirk, 2015; Lum et al., 2017; Runions et al., 2020). Lum et al. (2017) suggest that teachers' knowledge and attitudes are among the most important modifiable factors in educational outcomes for children with chronic illness. Adams and Bourke (2021) report that teachers of children with chronic illness have highlighted the need for broader training related to supporting the educational, social and emotional needs of students in the context of illness. The authors note however that teachers who have experience supporting children with chronic illness also appreciate the importance of learning about the student as an individual rather than learning solely about their condition and medical needs. They note the importance of “working as a team” to include parents, students and medical professionals in the

on-going journey of supporting students' needs (Adams & Bourke, 2021). This may be of particular relevance to children with cystinosis as the impact of the illness can vary at different times and indeed between individuals.

Theoretical Frameworks

Only one study (Spilkin & Ballantyne, 2007) referenced theory underpinning their conceptualisation of the psychosocial experience for children and families in relation to their research. This is an important finding given the complexity of understanding the multiple potential components of and influences on wellbeing. Future research should address this gap by illustrating a theoretical framework of wellbeing in the context of cystinosis.

Implications for Practice

In line with research from the broader literature on rare diseases (e.g. Verger et al., 2021; Kole et al., 2021) the studies included in this review have noted the need for multidisciplinary management and support for children with cystinosis (Atia et al., 2022; Aly et al., 2014; Ballantyne, 2013; Spilkin & Ballantyne, 2007; Ulmer et al., 2009). However, there is limited detail on how or where this support should be provided. Spilkin and Ballantyne (2007) noted that 80% of children rarely receive therapeutic psychological support for social and/or emotional issues, despite reported challenges with behaviour, family adjustment and academic difficulties in the same sample. The authors suggest building on child and family strengths to formulate interventions and help parents and children cope with the challenges of cystinosis. They further note the importance of defining and creating awareness of the unique challenges faced. Ulmer et al. (2009) also highlighted the need for patients to receive supportive and comprehensive treatment in light of the internalising and externalising difficulties expressed. Despite the identified needs, none of the studies gathered families or teachers views regarding what was needed in terms of support for children, or examined the barriers to accessing support. Research in the area would benefit from a strengths-based, solution focused approach to expand our understanding of how to support children with cystinosis, particularly in educational settings.

Limitations of the Review

In analysing the results of this systematic review, it is important that they are considered in the context of limitations present. Firstly, due to resources, it was not possible to include studies that were not published in English. Therefore

some relevant studies and information may not have been included. Secondly, some studies were excluded where it was not possible to differentiate the experiences of children with cystinosis from those of children with other rare diseases. While this may have resulted in some additional information being omitted, it was deemed necessary to differentiate these experiences so as not to obscure those of children and young people with cystinosis.

Conclusion

While research in the area of psychosocial and educational experiences of children with cystinosis is limited and is primarily quantitative, it indicates challenges with behavioural and social and emotional adjustment (Atia et al., 2022; Delgado et al., 2005; Spilkin & Ballantyne, 2007; Ulmer et al., 2009). What remains largely unknown is how the unique challenges and barriers faced by children and young people impact their wellbeing in the context of the education system. The perspectives of teachers regarding the needs of children with cystinosis is notably absent in the research to-date. Furthermore, while researchers point towards the need for multidisciplinary support, the voice of parents and teachers in identifying those supports is lacking in the current literature.

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Chapter 3: Empirical Study Methodology

Chapter Overview

A systematic literature review of the psychosocial and educational impact of cystinosis for children and young people showed that children are at risk for social-emotional and behavioural challenges (Aly et al., 2014; Ariceta et al., 2015; Delgado et al., 2005; Spilkin and Ballantyne, 2007; Ulmer et al., 2009). However, this research is limited, particularly from a qualitative perspective, and offers little to contextualise these risks. Despite research showing that children with rare diseases are at risk for adverse school experiences, there is little known of the school experiences of children with cystinosis. None of the research to date includes the perspectives of teachers or school staff. The current research aimed to explore parents' and school staff perspectives on the psychosocial strengths and needs of children with cystinosis and the factors influencing their school experiences. The research adopted a qualitative paradigm informed by a constructivist epistemological perspective and underpinned by Bronfenbrenner's Bioecological theory (Bronfenbrenner & Morris, 2006). Data was gathered through semi-structured interviews, co-designed through a patient public involvement (PPI) group). Reflexive thematic analysis (TA) was utilised as an analysis strategy. This chapter provides an overview of the methodology used in the empirical study.

Research Questions

The current study aimed to explore parents' and school staff's perceptions of the psychosocial strengths and needs of children with cystinosis and to identify factors influencing their school experiences. The specific research questions are:

1. From the perspective of parents and school staff, what are the psychosocial strengths and needs of children with cystinosis?
2. What do parents and school staff perceive to be the factors influencing the school experiences of children with cystinosis?

Epistemology

Psychological research is underpinned by assumptions of an ontological nature, that is, assumptions about the nature of the world and reality. Ontology asks us what our views are on the nature of reality and the phenomenon we wish to study (Creswell & Plano Clark, 2007; Ponterrotto, 2005). Epistemology is concerned with how knowledge about the nature of the phenomena under study can be acquired. A researcher's epistemological position reflects their

assumptions about what is meaningful and valid knowledge and how this knowledge can be generated and therefore has important implications for research (Braun & Clarke, 2021; 2022; Byrne, 2022). For those who adopt a constructionist epistemology, multiple, constructed realities exist that are subjective and influenced by the participants' experience and perceptions, the social environment, and the interactions between the participant and the researcher (Braun & Clarke, 2022; Terry, 2017; Jha, 2012). Adopting a bio-ecological lens, the current study aimed to explore parents and school staff's perceptions of the psychosocial strengths and needs of children with cystinosis and to identify factors influencing their school experiences. A constructionist framework allowed for the illustration of people's experience in a social context, as interpreted by a researcher.

Research Design

In line with a constructionist epistemology and the research aims, this research adopted a qualitative research design utilising semi-structured interviews with parents and school staff. Qualitative research results in data that includes the voices of participants, reflection and subjectivity on the part of the researcher, description and interpretation of the phenomena being studied and a contribution to literature or policy and procedures (Creswell, 2014; Denzin, 2007; Kelly, 2010). Pope and Mays (2020) describe qualitative research as an interpretive approach, one that is concerned with the meaning that people attach to experiences. Given the richness of this approach it is well suited to understanding the complex and sensitive nature of the research topic. Research in rare diseases can be challenging due to their inherently low prevalence but sample sizes in qualitative research are typically small, which allows for an in depth analysis (Vasileiou et al., 2018). The decision to approach the research questions utilising a qualitative design was also influenced by a lack of qualitative research in relation to cystinosis.

Public and Patient Involvement (PPI) is becoming an important and valued part of research in health and social care. Westerink et al. (2023) note that patient participation brings unique expertise and knowledge, which can be used to improve quality of care, clinical outcomes, patient experiences, increase research relevance and impact, and increase public trust in research. PPI is of particular importance in the context of rare diseases such as cystinosis due to their low

incidence and the unique challenges presented by their conditions (Velvin et al., 2022). In order to ensure that this research captured information of value to families and people with cystinosis and that the research questions aligned with their experiences, a PPI group was established by Cystinosis Ireland to review the interview schedules against the study's research objectives. The PPI group was made up of six contributors and included parents of children with cystinosis and adults with cystinosis.

Participants

Participants in the study consisted of parents (n = 5) and teachers/support staff (n = 7) of children with cystinosis. Purposive sampling was employed for this study. Purposive sampling can be used when researchers want respondents who are knowledgeable about the issues under investigation (Sarfo et al., 2022). Given their insight into the day to day experiences of children with cystinosis, parents and school staff were considered to be well placed to provide a detailed account of their experiences. Access to participants was facilitated by Cystinosis Ireland, the national advocacy group for people with cystinosis in Ireland. The sample size was based on the age of school-going children with a diagnosis of cystinosis in Ireland (N=7). One family who consented to take part was subsequently unable to complete the parent interview due to their child's medical needs at the time of interview but their child's school staff took part. Data from one child with a neurodevelopmental condition was omitted from the study as it was deemed that their experiences, strengths, and needs were unique and best understood in the context of rare illness and neurodiversity. Data were analysed for the remaining six children (five male; one female) with cystinosis. All remaining parent interviews (N=5) were conducted with mothers. Five of the children's schools also participated in the research, including one preschool, three primary schools, and one post-primary school. Two schools were located in Northern Ireland and the remaining schools were in the Republic of Ireland. Three of the interviews were conducted with classroom teachers, two were conducted jointly with teachers and a special needs assistant (SNA), and one interview was conducted with an SNA only. Parent and school staff interviews were conducted separately. Parent/School dyads are summarised in Table 5. To protect anonymity of the participants due to the small sample size, the age and gender of children have been omitted. As

there was only one child attending preschool, this child has been included in the "Primary School" stage of education.

Table 5: Composition of Parent School Dyad's

Child	Education Stage	Parent/s Interviewed	Staff Interviewed
1	Post-Primary	Did not take part	Teacher and SNA
2	Primary School	Mother	Teacher
3	Primary School	Mother	Teacher
4	Primary School	Mother	Teacher and SNA
5	Post-Primary	Mother	Did not take part
6	Primary School	Mother	SNA
Total Interviews		5/6	5/6

Children ranged in age from 4 to 16 years, with an average age of 10 years, 11 months. None of the children in the cohort had received kidney transplants. One child was undergoing dialysis at the time of the study. While the parents consented for this child's teachers to take part, the parents were unable to take part and therefore additional medical information was not available. Of the remaining children, all were diagnosed with cystinosis before the age of three years, with an average age of 22 months. Four children had a gastric tube through which they received medication and primary nutrition. The remaining child had their gastric tube removed in the previous 12 months.

Procedures

An oral presentation on the rationale and aims of the study was provided to parents of children with cystinosis at the Cystinosis Ireland conference in April 2023. Following the conference Cystinosis Ireland emailed the information letters and consent forms to all parents (refer to Appendix B). Parents returned the consent form directly by email to the researcher and also indicated their consent for their child's class teacher and if applicable their special education teacher and special needs assistant to be invited to participate. For children attending post-primary school, the invitation was extended to their year head and if applicable their special education teacher and special needs assistant. Prior to conducting interviews, age-appropriate assent forms (refer to Appendix C and D) were

provided to each of the children to ensure that they understood the purpose of the interviews and assented to their parents and teachers talking about their experiences. For children who were unable to read, parents read the assent forms to their child and described the research and what would be involved in terms they felt their child could understand. On return of parental consent and assent, the researcher contacted the principal/preschool manager by email, attaching information and consent forms for the study (refer to Appendix E), and requesting that the information was shared with teachers/school staff who may be interested in taking part.

All interviews were conducted remotely due to participant choice. Four parent interviews were conducted via an online secure video platform (zoom) and one was conducted via telephone. One school interview was conducted via telephone and the remaining school interviews were conducted via secure video platform (zoom). Parent interviews ranged in length from 62 to 99 minutes with an average duration of 70 minutes. School interviews ranged in length from 44 minutes to 74 minutes with an average of 59 minutes. Interviews were transcribed verbatim for the purpose of analysis and all potentially identifying information omitted.

Data Collection Methods

Information was gathered through semi-structured interviews with parents and school staff. Semi-structured interviews were chosen in order to elicit a detailed, contextualised account of parents and teachers perspectives (Kelly, 2010). Interviews were guided by open-ended questions centred around a set topic. In keeping with qualitative approach and reflexive TA, a flexible and fluid approach was adopted, with prompts and probes used to delve further into participants responses when required (Braun & Clarke, 2022; Kelly, 2010). Two separate interview schedules were devised, one for parents and one for teachers. The interview schedules were informed by research in the area of chronic and rare illness and by professional experience. Overarching questions were divided into seven categories covering: Interests and Strengths, School Experiences, Social Experiences, Medical and Physical Impact, Psychological Wellbeing, Home School Relationships and School Training and Knowledge. Initial interview schedules were piloted through a PPI group established by Cystinosis Ireland. Contributors were provided with a description of the research aims along with the

interview questions. Each PPI participant was asked to review the interview schedules under the following criteria, determined by Cystinosis Ireland.

- *Are the questions easy to understand?*
- *Is the language used appropriate and clear?*
- *Are the questions relevant/suitable?*
- *Will the researchers get what is needed from the parents, the children, and the teachers?*
- *Do you have any suggestions on how the questions might be improved/made more relevant? Please feel free to add/change/remove questions and give your rationale where possible.*

Interview questions were refined and adapted based on feedback from the PPI contributors and a final version of the schedule sent to the group for approval. Where the PPI group suggestions were not included in the final interview schedule, a rationale was provided (i.e. where suggestions for additional questions were not deemed to align with the research questions). Parent and school interview schedules are presented in Appendix F and G respectively.

Data Management

Interviews were audio recorded using an iPad voice recording app which was not connected to the iCloud. Following the interview, the audio recording was transferred to a password protected encrypted laptop and the audio file was deleted from the iPad. Audio files were sent to an external transcription service via a password protected link. All interviews were transcribed into Microsoft word documents and returned to the researcher via email. Transcription documents were downloaded and stored in a password protected file on the researcher's encrypted password protected computer and the email and attachments were deleted. The researcher re-checked all data by listening to the audio recording and reading the transcription. Following this additional check and familiarisation, all audio files were permanently deleted. Anonymised interview transcripts were uploaded to NVivo, a qualitative data analysis computer software. Consent and assent forms were stored separately to transcription documents on the researchers encrypted, password protected computer.

Timeline

Parents were invited to take part in the research in April 2023. All parents who returned consent forms were contacted in June 2023 in order to begin

scheduling the interviews and to obtain contact details for their child's school. With parental consent and child assent, all schools were contacted in June 2023. Due to school holidays, interviews were postponed until September 2023. Parent interviews were conducted between September 2023 and January 2024. School interviews were conducted between October 2023 and January 2024. Data analysis began in January 2024 and was completed in March 2024. The timeline of recruitment, data collection and data analysis is provided in Table 6 below.

Table 6: Timeline of Research Process

Research Stage	**Dates**
Parent participants invited to take part in research	April 2023
Parents contacted to begin scheduling interviews and to obtain school contact details	June 2023
School staff invited to take part in research	June 2023
Parent Interviews	September 2023 – January 2024
School Interviews	October 2023 – January 2024.
Data Analysis	January 2024 – March 2024
Report Write-up	March 2024 - April 2024

Analysis Strategy

Interview transcripts were analysed using Braun and Clarke's reflexive thematic analysis (TA) framework (e.g. Braun & Clarke, 2013; 2022). Reflexive TA is an approach to qualitative data analysis that involves the identification and analysis of patterns, or themes, in data which can produce rich and insightful findings (Byrne, 2022). Reflexive TA provides a flexible approach that can be modified for the needs of research providing a complex understanding of the phenomena under investigation. Reflexivity as the name suggests is central to reflexive TA and acknowledges the researchers active role in the production of knowledge (Braun & Clarke, 2019; 2022). In research, reflexivity is a process involving reflecting on and critically interrogating assumptions, expectations, choices, and actions throughout the research project (Braun & Clarke, 2022;

Finlay & Gough, 2003; Olmos-Vega et al., 2023). As an analysis strategy, reflexive TA embraces subjectivity in meaning making and was therefore considered appropriate for the constructionist epistemology of the study (Braun & Clarke, 2019; Byrne 2022). Reflections were kept in a reflective journal and memos stored in NVivo throughout the research process to identify emerging thoughts and impressions.

Braun and Clarke (2013; 2022) suggest six phases to thematic analysis (see figure 1) which were be used to guide the analysis of data collected in this research: data familiarisation and writing familiarisation notes, systematic coding of data, sorting coded data into initial themes, reviewing themes, defining and naming themes within data and finally writing up themes to tell the story of the data. Although each stage is distinct, the reflexive nature of the analysis called for flexible, recursive movement throughout the stages as the researcher's understanding and thinking developed (Terry, 2017; Braun & Clarke, 2022).

Samples of the analysis process are presented in Appendix H. In phase one of the analysis, the researcher re-read each of the transcripts while simultaneously listening to audio-recordings. Each transcript was then read several times. Initial ideas, feelings, and thoughts were noted using the annotation and memoing functions in NVivo. In the second phase, codes were generated. Coding is considered an iterative, evolving process (Braun & Clarke, 2019; 2022). During the first coding phase, any data that was considered relevant to the research questions were coded. As the researcher's thinking evolved over time with deeper familiarisation with the data, codes were later refined and items re-coded. Meaningfulness of data was of particular importance given the constructionist epistemology of the study (Byrne, 2022). In phase three, codes that shared similar ideas or meanings were clustered into initial "candidate" themes. Distinct themes were identified based on central organising concepts (Braun & Clarke, 2022). The term "candidate themes" was adopted to reflect the ongoing and iterative nature of theme development in this early stage. Candidate themes required additional exploration in relation to their meaning and their relevance to the research questions. Thematic maps were utilised to visually explore the relationships between themes and subthemes (Braun and Clarke, 2019; 2022). Supervision meetings which included reflection on codes, themes, and overall meaning was integral to guiding the process.

Initial themes were reviewed and refined in phase four. Four guiding questions as suggested by Braun and Clarke (2022) were used to guide reflection on themes as follows:

1) Does the theme have clear boundaries? What does it include/not include?

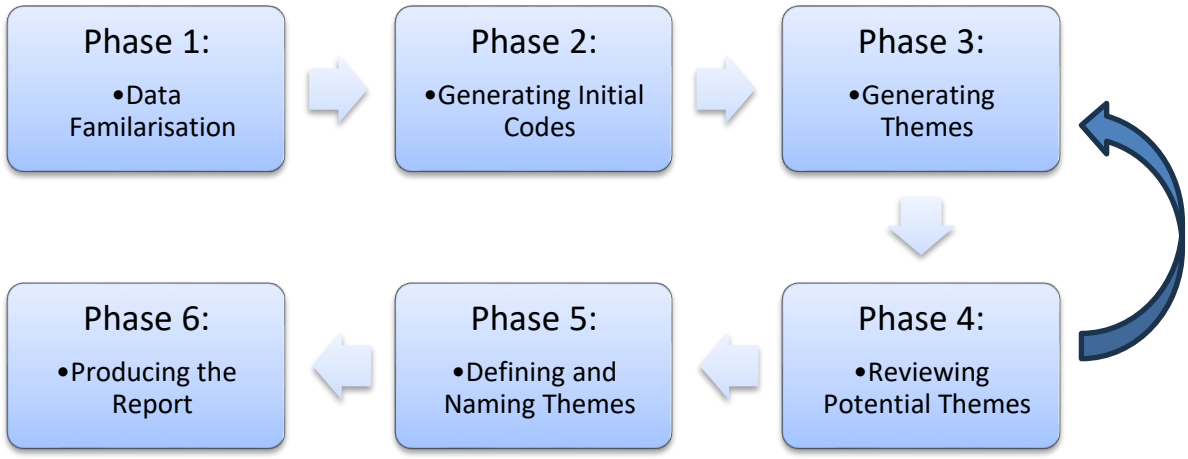
2) Are there enough meaningful data to evidence the theme?

3) Does the theme lack coherence?

4) Does this theme convey something important in relation to the data set and research questions?

The researcher moved between stages three and four in a recursive, reflective pattern until it was felt that the themes captured distinct and important meanings relative to the research questions. Each of the themes were subsequently defined based on the core centralising concept of the theme. Data extracts that provided clear and compelling evidence to illustrate the themes and subthemes were selected and the final report was written.

Figure 2: Phases of Reflexive Thematic Analysis



Validity

Braun and Clarke (2021; 2022) suggest that due to the subjective nature of reflexive TA and its associated theoretical underpinnings, validity and reliability cannot be accurately represented. Rather, they stress the importance of "quality"

and encourage reflection, rigour, a systematic approach and deep engagement over accuracy of coding/theme development. Several steps were taken to ensure quality and integrity of this study in this regard. Both electronic and handwritten tools were utilised in analysis. Reflections and engagement with the data was promoted through the use of a handwritten reflective journal and thematic maps (Braun & Clarke, 2021; 2022). Initial thoughts and observations were recorded throughout the analysis utilising the memoing and annotation functions in NVivo. Interview schedules were reviewed by a PPI group to ensure that they captured data that was relevant and valuable from the perspective of people who have lived experience of the research topic. As an external transcription service was used, all transcripts were reviewed against the audio recordings to ensure accuracy. This process also supported further researcher familiarisation with the data set which may have been lost though outsourcing of transcription. Triangulation refers to a method of enriching understanding of a phenomenon by viewing it from different perspectives (Flick, 1992). This was achieved by gathering data from different groups of people (i.e., teachers, SNAs, parents). Finally, supervision was integral to the analysis process. During supervision, interviews, codes, and themes were discussed in detail, providing an additional space to critically reflect on the meaning of data and the themes identified.

Limitations

It is acknowledged that a significant limitation of this research is that the perspectives of children and young people with cystinosis were not included. However, this was a conscious decision as the current research study forms part of a wider research project being conducted by UCD for Cystinosis Ireland. Part of the overall research project involves psychoeducational assessments and interviews with children in order to identify cognitive and attainment strengths and needs and also to get insight into their school experiences. The researchers were cognisant that children with cystinosis and their families interact with multiple professionals as part of their overall care. Due to the low prevalence of cystinosis, people with cystinosis are often asked to take part in research. Cystinosis Ireland stressed the importance of valuing families' time and being mindful of the children's energy levels and overall health when planning research. In a survey by Rare Disease Europe, people with rare diseases noted the importance of developing a positive relationship with researchers and being seen as more than

just research subjects (Eurodis, 2018). In light of these considerations, the decision was made for the current research to focus on interviewing parents and school staff. This was deemed to be more ethical in terms of placing undue burden on the children and their parents and their family time.

Another methodological limitation of the current study is the low participant numbers. This is a well-documented challenge in the field of rare conditions due to low prevalence of conditions. The current study included data from a parent and/or a teacher/special needs assistant for all children with cystinosis in Ireland and Northern Ireland. This study also highlights the advantages of remote technology to support qualitative research in rare disease, which may facilitate researchers to overcome some of the geographical challenges associated with rare disease research (Dwyer et al., 2022). While data pertaining to one child with cystinosis and a neurodevelopmental condition was omitted, with parental consent the experiences of this child will be shared with Cystinosis Ireland in order to highlight areas of support need for children with rare disease and neurodevelopmental conditions and for future research considerations.

Ethical Assurances

Ethical approval for the current study was granted by the Human Research Ethics Committee, University College Dublin (See Appendix I for HREC ethical approval email). The study was conducted in accordance with the ethical guidelines outlined by the Psychological Society of Ireland (2019). Ethical dilemmas which were considered before the data collection process commenced included ensuring voluntary participation and informed consent/assent, sensitivity of the topic discussed and potential distress to participants, and protecting confidentiality in a small sample size.

Informed consent/assent was required for all participants and children with cystinosis. Information letters (see Appendices B, C, D & E) outlining the purpose, process and the potential outcomes of the study were provided to all participants. Contact details of the researcher and supervisor were provided and participants were advised that they could make contact if they had any questions. An oral presentation regarding the research was provided for parents at the 2023 Cystinosis Ireland conference which facilitated parents an opportunity to ask questions about the research directly. All participants were assured of the voluntary nature of their participation and their right to withdraw their data up until

two weeks after the time of the interview as at that point analysis would have commenced.

As information about the study was provided through Cystinosis Ireland, it was acknowledged that parents may have felt obliged to take part as they receive support and advice from this organisation. Participants were assured that non-participation or withdrawal would have no impact on their access to support from Cystinosis Ireland and that while information regarding the study will be disseminated by Cystinosis Ireland, they will not know which families take part in the research. A further consideration was that parents may have felt obliged to take part in the research in order for their child to receive a psychoeducational assessment. Parents were therefore assured that they could avail of the psychoeducational assessment regardless of whether they wished to take part in the study. Similarly, there was no obligation for parents to take part in the interview in order to avail of the psychoeducational assessment. Age appropriate assent forms (Appendix C and D) were provided to each of the children to ensure that they understood the purpose of the interviews and agreed to the researcher speaking with their parents and teachers about their experiences. Where children were unable to read, parents read the assent forms to their children and described the research and what would be involved in terms they felt their child could understand. Prior to each interview, the transcription process, confidentiality, limits of confidentiality and the researcher's obligations as a mandated person were explained verbally to each participant. Participants were asked to confirm their consent to take part and for the interview to be recorded.

Given the sensitive nature of the topics being discussed during the interview, there was a risk of potential distress to parents as they discussed challenges faced by their children as a result of their condition. As teachers and support staff were also interviewed in relation to the children's experience of school there was similar potential that teachers and SNAs may have become upset if they felt they were struggling to meet the needs of the child in school. It was agreed that interviews would be stopped in the event of a participant becoming distressed. All participants were offered a copy of the interview schedule in advance of the interview to reduce anticipatory stress. This also provided participants with the option to opt out of the research before the interview if there was an area that they were not comfortable discussing. Following the

interview, all participants were given information on support services they could contact in the event of any distress or need for additional information. The contact details for researcher and supervisor were also provided.

Given the small sample size, protection of anonymity was a particular concern and priority. Given the small number of CYP with cystinosis in Ireland and the fact that the sample was recruited from a central support organisation, it is likely that the families knew each other. Strict General Data Protection Regulations (GDPR) were followed to protect the identity of all participants involved in the study. Data was stored in an encrypted format on encrypted devices to ensure anonymity and an ID code was assigned to each participant and the child being referred to. Following interviews, all recordings were sent to an external transcription service via a password protected link. On the return of the transcribed files audio recordings were deleted. As written transcriptions were returned via email, emails were deleted once the files were downloaded. Participants were offered a copy of their de-identified interview transcript. Care was taken during write up not to include quotations where the participant may have been identified (i.e. isolated references to preschool, colloquial phrases/language, location etc). As there was only one female child in the study, participant quotes identifying this child's gender were not included or changed to male pronouns (e.g. he/him) to protect anonymity.

Summary

The objectives of this research was to explore parent and school staff perspectives on psychosocial strengths and needs of children with cystinosis and to identify the factors impacting their school experiences. The research was informed by a constructionist epistemology, positioned within a qualitative paradigm. Semi-structured interviews using a researcher-designed schedule were conducted remotely to gather rich, contextualised information from parents and school staff of six children with cystinosis. Data were analysed using reflexive thematic analysis and steps were taken to ensure that the study was rigorous and robust, including following criteria and guidelines for qualitative research, engaging in regular supervision meetings with the research supervisor and documenting the stages of the research journey. The findings of this empirical study are outlined in the following chapter.

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Chapter 4: Empirical Journal Article

Overcoming Barriers, Finding Solutions: Parent and School Staff Perspectives on the Factors Influencing School Experiences of Children with Cystinosis

Abstract

Cystinosis is a rare, multi-system disorder which causes an accumulation of the amino acid cystine in the organs and tissues of the body, causing widespread tissue and organ damage. While there is limited quantitative research showing that children with a diagnosis of cystinosis are at risk of social and emotional difficulties, there is a significant lack of qualitative research that contextualises these risks. Despite research showing children with rare diseases are at risk for adverse school experiences, there is very limited knowledge of the school experiences of children with cystinosis, with none of the research to date including the perspectives of school staff. Underpinned by bioecological theory, the current qualitative study aimed to provide insight into the strengths and needs of children with cystinosis and identify factors influencing their school experiences. Semi-structured interviews were conducted with the parents (n = 5) and teachers/special needs assistants (n = 7) of six children (aged 4 to 16) with cystinosis. Reflexive thematic highlighted participants' perspective on children's school experiences and emphasised the importance of all stakeholders working together to overcome barriers, particularly at times of increased medical need. Participants identified the unique challenges living with the condition presents as well as the need for increased awareness about cystinosis. Implications for educational psychology practice and opportunities for further research are discussed.

Keywords: cystinosis; education, psychosocial, wellbeing, rare disease, school, teachers

Overcoming Barriers, Finding Solutions: Parent and School Staff Perspectives of the Factors Influencing School Experiences of Children with Cystinosis

Introduction

The European classification of a rare disease or disorder is an occurrence of less than 1 in 2,000 people (Rare Disease Ireland, 2024). While there are over 6,000 identified rare diseases, some common features exist between them, namely that they are chronic, severe and/or progressive, and present with various degrees of sensory, motor, physical and/or intellectual disability (Aymé, 2012; Rosselló 2018). Cystinosis is a rare, chronic, multi-system disorder which causes an accumulation of the amino acid cystine in the organs and tissues of the body, causing widespread tissue and organ damage (Doyle & Werner-Lin, 2015; NORD, 2024). Current estimates indicate that cystinosis occurs in approximately 1 in 200,000 live births within developed countries (Cystinosis Ireland, 2024). The condition is characterised by a demanding life-long treatment regime and frequent medical interventions and hospital appointments. People with cystinosis experience gastrointestinal symptoms such as frequent vomiting, decreased appetite and feeding difficulties, and severe growth restriction. Over time, several organs are affected including eyes, thyroid glands, muscles, liver, lungs, and bones (O'Connell et al., 2022). Without specific treatment, almost all patients with cystinosis will develop end stage renal disease (ESRD) by 10 to 12 years of age. Medical advancements, particularly early identification, availability of kidney transplantation, and long term treatment with cysteine depletion therapy delays progression to ESRD, decreases frequency and severity of complications, increases growth, and is associated with extended life-expectancy (Ariceta et al., 2019; Nesterova & Gahl 2013).

Cystinosis and School Experiences: A Psychosocial Perspective

While much research focuses on the causation, identification, and medical treatment of rare diseases such as cystinosis, there has been a shift towards understanding the day to day reality of living with a rare condition. A participatory multiphase design study by Somanadhan et al. (2020) identified the psychosocial

impact of living with a rare disease as a top research priority for people with rare diseases in Ireland. From a psychosocial perspective, living with rare diseases and chronic health conditions is associated with mental health and psychological difficulties (Adama et al., 2023; Cohen & Biesecker, 2010). Children with rare diseases are at risk of negative experiences such as stigmatization (Bogart, 2014) bullying (Branch Smith et al. 2018; Foster et al., 2022), and social isolation and exclusion (Adama, 2023). For many children with rare diseases, medical advancements and increased survival rates mean many children can participate in mainstream education (Sommer & Klug, 2024). However, very little is known about the psychosocial experiences of children with cystinosis from a school context. This is a significant gap given the pivotal role of education and school experiences in children and young people's lives.

Children's psychosocial experiences in school are integral in promoting positive mental health, quality of life, social relationships, and wellbeing (DES, 2019; Lum et al., 2017; Runions et al., 2020). Wellbeing is challenging to define as there is no universally accepted definition (Svane et al., 2019; Tynan & Nohilly, 2021). It is a broad concept that has been noted to include many subsets such as emotional wellbeing, social wellbeing, psychological wellbeing and mental health (Svane et. al 2019). The World Health Organisation defines mental health as a "state of mental wellbeing that enables people to cope with the stresses of life, to realise their abilities, to learn well and work well and to contribute to their communities" (WHO, 2022 p.8). This conceptualisation suggests that positive mental health goes beyond the absence of mental disorders and that mental health is an integral component of wellbeing. In Ireland, the Department of Education and Skills (DES) has also adopted this multidimensional conceptualisation of wellbeing in the wellbeing policy statement framework for practice (DES, 2019). Importantly, this policy highlights that wellbeing is a fluid concept that is experienced at a personal level and is also associated with a range of risk and protective factors operating across various contexts. Positive school relationships, sense of school belonging and connectedness, opportunities for success and the presence of systems that support children at times of difficulty are among the protective factors for children's wellbeing and mental health (DES, 2015;

2019). On the other hand, adverse experiences such as relationship difficulties, absenteeism, alienation and disengagement may place children's wellbeing at risk. Identifying these factors for children with cystinosis is essential so that the appropriate supports can be provided.

At the individual level, while children with rare conditions face many of the same challenges as others in navigating educational and social situations, these challenges are further exacerbated when managing medical conditions. In a study evaluating health related quality of life (HRQOL) for children with mild-moderate chronic kidney disease (CKD), Gerson et al. (2010) report that children with CKD have poorer HRQOL than healthy children, with the most marked differences evidenced in relation to school functioning. Factors such as medical and care needs, irregular attendance and prolonged absences due to hospitalisation and illness may mean that young people with chronic illness fall behind their peers (Lum et al., 2017; Runions et al., 2020). From a psychological perspective, this may result in increased stress and anxiety as they struggle to keep up with schoolwork. In a quantitative study of 34 paediatric and adult patients with cystinosis in Spain, Ariceta et al. (2015) reported that school attendance was impacted for 29% of children (<18 years), however, the effect of absenteeism on psychological wellbeing was not explored.

Socially, research has shown that children with rare or chronic conditions may experience social isolation, bullying, and discrimination (Adama et al., 2023; Foster et al., 2022; Lum et al., 2017). In a quantitative study of 64 children with cystinosis, Delgado et al. (2007) found that parent-reported scores on the Social Problems scale of the CBCL were significantly higher for children with cystinosis compared to children with cystic fibrosis. Medical needs and school attendance difficulties may have a negative effect on relationships as young people miss out on social opportunities. Santos et al. (2016) reported that substantial numbers of adolescents with chronic illness felt that their disease impacted on participation at school and in leisure activities. For children with cystinosis, muscle weakness can result in fine motor difficulties which may impact on academic performance while gross motor difficulties may negatively impact peer interactions and impair self-esteem (Ulmer et al., 2009). Having a sense of school belonging has been linked to positive outcomes in mental

health, self-concepts and overall health. Children with cystinosis are often of smaller stature than peers, which may impact on their sense of confidence and ability to partake in extra-curricular sports activities. In a study of 402 children with mild-moderate chronic kidney disease (CKD), Gerson et al. (2010) found that short stature is associated with negative impact on quality of life and increases difficulty in physical functioning. Risk of dehydration and light sensitivity means that outdoor activities on hot days can be curtailed, which could lead to feelings of isolation, feeling different, or lost social opportunities. Side effects of cystinosis medication, which include nausea and an unpleasant body odour, may further impact on children's social relationships (Beinart et al., 2014). In a review examining the link between chronic illnesses (asthma, cancer, chronic kidney disease, heart diseases, cystic fibrosis and gastrointestinal diseases) and school experiences, Lum et al. (2017) noted that children with chronic illness reported bullying and teasing, concerns about keeping up with peers, and negative attitudes from both teachers and peers. The authors further noted reluctance to attend school due to lower sense of confidence and feeling different from peers. Feeling different from peers is a significant concern for many children with other chronic illnesses, such as Cystic Fibrosis (D'Auria, Christian & Richardson, 1997). Adults with cystinosis have reported feeling different to other people due to their short stature (Beinart et al., 2015). Adults with cystinosis have reflected on their reluctance to disclose their diagnosis for fear of stigmatisation (Doyle, 2015), however, less is known about how children with cystinosis manage disclosures to peers. This may be of particular relevance during adolescence when children and young people place an increased emphasis on peer relationships and acceptance.

At a systemic level, current policy and practices strive to encourage inclusion of children with rare conditions in mainstream schools. However, teachers are often unaware of the potential complexities of rare and chronic health conditions (Runions et al., 2020). A general lack of resourcing, training, and clear policy on supporting children with medical conditions may increase challenges for teachers, parents, and students alike (Adama et al., 2023; Foster et al. 2022). To-date there are no studies to date that include data from teachers or school staff supporting children with

cystinosis, despite the significant role they play in children's school outcomes and mental health, as well as the importance of communication and relationships between parents and school staff (Hinton & Kirk, 2015; Lum et al., 2017). Lum et al. (2017) suggest that teachers' knowledge and attitudes are among the most important modifiable factors in educational outcomes for children with chronic illness. Adams and Bourke (2023) report that teachers of children with chronic illness have highlighted the need for broader training related to supporting the educational, social, and emotional needs of students managing medical conditions. The authors further note that teachers who have experience supporting children with chronic illness appreciate the importance of learning about the student as an individual rather than learning solely about their condition. They note the importance of "working as a team" to include parents, students and medical professionals in the on-going journey of supporting students' needs (Adams & Bourke, 2023). This may be of particular relevance to children with cystinosis as the impact of the condition can vary at different times and between individuals. It is crucial that schools are aware of and understand these challenges and that accommodations and supports are provided accordingly such that children with medical conditions can be provided with the right psychological, social, and educational supports to achieve their potential (Hillard et al., 2015; Verger et al., 2020).

While research indicates that the children and young people with cystinosis may be at risk of social, emotional, and behavioural difficulties (Aly et al., 2014; Ariceta et al., 2015; Delgado et al., 2005; Spilkin and Ballantyne, 2007; Ulmer et al., 2009), the current research is limited in its ability to contextualise these risks in order to provide an understanding of strengths and needs. The majority of the current studies relied on quantitative measures (i.e. Achenbach Child Behaviour Checklist [CBCL]). Although such quantitative measures are widely used in research and professional practice as a measure of emotional and behavioural adjustment, some researchers have raised concerns about their use for children with medical conditions. Perrin (1995) cautions that the items on the social competence scale of the CBCL assesses children's participation in social activities which may be limited by virtue of the child's condition, and should not be interpreted as indicating that the

child is less socially competent. The closed-end nature of the questions on such measures may result in the loss of rich, person focused information which can only be gathered by discussion with those with more direct knowledge of the experiences of children with cystinosis (Spilkin & Ballantyne, 2007). As such there is a need for qualitative research that can provide a detailed, contextualised account of children's experiences and identify potential risk and protective factors.

Rare Disease and the Role of Educational Psychologists.

To-date there is limited, if any, research on the role of educational psychology in relation to supporting students with rare medical conditions within the school system. Researchers have noted that psychologists working in the education system have historically had a minimal role in the assessment and treatment of children with medical conditions (Brown and DuPaul, 1999). However, as both medical treatments improve and inclusive practices and policy changes result in more children with rare medical conditions attending mainstream school, the role of EPs in systemically supporting children with rare conditions is increasing. While the significant lack of research in educational psychology and rare conditions is notable, and points to the need for increased focus in this area, the limited research in the area of chronic illness offers some guidance. Reilly and Fenton (2013) discuss the role of the educational psychologist (EP) in supporting children with epilepsy and highlight the importance of adopting a broad understanding of children's strengths and needs in the context of their individual medical needs. EPs can have a central consultative role when working at the school systems level and in relation to areas such as the potential impact of medications and collaborating with medical professionals to differentiate between medical and psychological/educational needs. They also have a central role in the identification of potential cognitive, emotional and behavioural challenges and the design, implementation and monitoring of effective, evidence-based interventions to support children and teachers (Reilly & Fenton, 2013). Barraclough & Macheck (2010) further highlight that psychologists also play a role in determining the best educational settings for children with medical needs, identifying required resources, and in informing families and children regarding their educational rights. Educational psychology is also central to the promotion of wellbeing.

Approaching wellbeing from a holistic, systemic perspective, educational psychologists consider the dynamic nature of cognitive, social, emotional, and physical aspects of children's lives and the impact of these on development.

Conceptual Framework

Bronfenbrenner's bioecological theory emphasises the dynamic influence of four factors on children's development, Process, Person, Context, and Time (PPCT) (Bronfenbrenner & Morris, 2006). The PPCT model considers the interdependent relationships between both individual and contextual systems and offers a framework for guiding support at multi-systems levels. The PPCT model provides a useful framework for contextualising the school experiences of children with cystinosis and how these impact on the child's psychosocial experiences in school. The first factor refers to "proximal processes", the reciprocal interactions that occur between individuals and other significant people, objects, and symbols in their environment. For Bronfenbrenner, proximal processes were considered the most powerful predictor of human development (Eriksson, Ghazinour & Hammarström, 2018; Tudge 2008). In the current study, parent-child, child-peer, school staff-child interactions are of particular relevance along with the child's interactions with school based activities such as sports, academic tasks and play. Bronfenbrenner noted that how interactions occur depends in part on the characteristics of the developing person (Tudge, 2017). "Person" characteristics were divided into three categories: demand (age, gender, race etc.), resource (intelligence, health etc) and force (temperament, resilience, motivation etc.) (Tudge et al., 2008). In the context of cystinosis, age, and resilience may impact the proximal processes at play. Similarly, the individual physical impact of the condition (e.g. side effects of medication, individual symptoms of the disease) may impact proximal processes as children's ability to take part in school-based activities and interactions may be impacted by their condition, for example, muscle weakness, fatigue, and light sensitivity may limit physical and outdoor activities.

"Context" involves four interrelated systems of Bronfenbrenner's model. The microsystem is the immediate environment where proximal processes occur. For children with cystinosis, microsystems may include the family, peers, and school

(Lehman et al., 2017; Runions et al., 2020). The mesosystem includes interrelations and interactions among several microsystems in which the person is situated, for example, interactions between school and home, school and medical professionals, and with advocacy groups. The exosystem includes processes that have an indirect influence on the child through their influence on others in the child's microsystem (Runions et al., 2020). In the school context, this could include access to teacher professional development, school policies, and access to psychology supports. The final system, the macrosystem, refers to the shared culture, norms, policies, and values that influences other factors (El Zaatari & Maalouf 2022; Lehman et al., 2017). In the context of the current study, factors such as health and education policies and availability of allied health care supports all indirectly impact the child's experiences. The final component of the PPCT model, "Time", refers to the influences of time, change over time, and events, on people, relationships, and all aspects of the ecosystem (Bronfenbrenner; 1994; El Zaatari & Maalouf, 2022). In the current study, specific times in a child's life such as transitions in the school system and developmental stages (e.g. adolescence) are all events that occur over time in the environment and may influence their school experiences. Given the progressive nature of cystinosis, individual features and symptoms can change significantly over time and children's needs will vary between times of relative consistency and critical periods of increased need, highlighting the importance of flexible, individualised, and responsive support and understanding.

The Current Study

For children and young people, the school environment plays a significant role in shaping their wellbeing. Underpinned by Bronfenbrenner's PPCT model, the aim of the current qualitative study was to explore psychosocial strengths and needs of children with cystinosis and to identify factors influencing their school experiences. This will inform policy and practice in schools in order to support the wellbeing of children with cystinosis and other rare diseases. The research was guided by the following two research questions:

- From the perspective of parents and school staff, what are the psychosocial strengths and needs of children with cystinosis?

- What do parents and school staff perceive to be the factors influencing the school experience of children with cystinosis?

Methods

Participants

In line with a constructionist epistemology, this research adopted a qualitative research design. Participants in the study consisted of parents (n = 5) and teachers/support staff (n = 7) of children with cystinosis. Purposive sampling was employed for this study. Purposive sampling can be used when researchers want respondents who are knowledgeable about the issues under investigation (Sarfo et al., 2022). Given their insight into the day to day experiences of children with cystinosis, parents and school staff were considered to be well placed to provide a detailed account of their experiences. The sample size was based on the age of school-going children with a diagnosis of cystinosis in Ireland (N=7). Access to participants was facilitated by Cystinosis Ireland, the national advocacy group for people with cystinosis in Ireland. One family who consented to take part was subsequently unable to complete the parent interview due to their child's medical needs at the time of interview but their child's school staff took part. Data from one child with a neurodevelopmental condition was omitted from the study as it was deemed that their experiences, strengths, and needs were unique and best understood in the context of rare illness and neurodiversity. Data were analysed for the remaining six children (five male; one female) with cystinosis. All remaining parent interviews (N=5) were conducted with mothers. Five of the children's schools also participated in the research, including one preschool, three primary schools, and one post-primary school. Two schools were located in Northern Ireland and the remaining schools were in the Republic of Ireland. Three of the interviews were conducted with classroom teachers, two were conducted jointly with teachers and a special needs assistant (SNA), and one interview was conducted with an SNA only. Parent and school staff interviews were conducted separately. Parent/School Dyads are summarised in Table 1 below. To protect anonymity of the participants due to the small sample size, the age and gender of children have been omitted. Additionally,

as there was only one preschool child in the cohort, this child was included in the "primary school" stage of education.

Table 1: Composition of Parent School Dyad's

Child	Education stage	Parent/s Interviewed	Staff Interviewed
1	Post-Primary	Did not take part	Teacher and SNA
2	Primary School	Mother	Teacher
3	Primary School	Mother	Teacher
4	Primary School	Mother	Teacher and SNA
5	Post-Primary	Mother	Did not take part
6	Primary School	Mother	SNA
Total Interviews		5/6	5/6

Children ranged in age from 4 to 16 years, with an average age of 10 years, 11 months. None of the children in the cohort had received kidney transplants. One child was undergoing dialysis at the time of the study. While parents consented for this child's teachers to take part, parents were unable to take part and therefore additional medical information was not available. Of the remaining children, all were diagnosed with cystinosis before the age of three years, with an average age of 22 months. Four children had a gastric tube through which they received medication and primary nutrition. The remaining child had their gastric tube removed in the previous 12 months.

Procedures

An oral presentation on the rationale and aims of the study was provided to parents of children with cystinosis at the Cystinosis Ireland conference in April 2023. Following the conference Cystinosis Ireland emailed the information letters and consent forms to all parents. Parents returned the consent form directly to the researcher and also indicated their consent for their child's class teacher and if applicable their special education teacher and special needs assistant to be invited to participate. For children attending post-primary school, the invitation was extended to their year head and if applicable their special education teacher and special needs

assistant. Prior to conducting interviews, age appropriate assent forms (refer to Appendix C & D) were provided to each of the children to ensure that they understood the purpose of the interviews and assented to their parents and teachers talking about their experiences. For children who were unable to read, parents read the assent forms to their child and described the research and what would be involved in terms they felt their child could understand.

All interviews were conducted remotely due to participant choice. Four parent interviews were conducted via an online secure video platform (zoom) and one was conducted via telephone. One school interview was conducted via telephone and the remaining school interviews were conducted via secure video platform (zoom). Parent interviews ranged in length from 62 to 99 minutes with an average duration of 70 minutes. School interviews ranged in length from 44 minutes to 74 minutes with an average of 59 minutes. Interviews were transcribed verbatim for the purpose of analysis and all potentially identifying information omitted.

Measures

Information was gathered through researcher-devised semi-structured interview schedules (see Appendix F & G) which were informed by research in the area of chronic and rare conditions, professional experience, and co-designed with a patient and public involvement (PPI) group established by Cystinosis Ireland. The PPI group consisted of six contributors and included parents of children with cystinosis and adults with cystinosis. Overarching questions were divided into seven categories covering: Interests and Strengths, School Experiences, Social Experiences, Medical and Physical Impact, Psychological Wellbeing, Home School Relationships and School Training and Knowledge. Additional prompts were used in a flexible manner when the researcher felt additional information was required.

Analysis Strategy

Interview transcripts were analysed using Braun and Clarke's reflexive thematic analysis (TA) framework (Braun & Clarke, 2013; 2021b; 2022). Reflexive TA is an approach to qualitative data analysis that involves the identification and analysis of patterns, or themes, in data which can produce rich and insightful findings (Byrne, 2022). Reflexive TA provides a flexible approach that can be modified for the needs

of research providing a rich understanding of the phenomena under investigation. Reflexivity is central to reflexive TA and acknowledges the researchers active role in the production of knowledge (Braun & Clarke, 2019; Braun & Clarke, 2022) As an analysis strategy, reflexive TA embraces subjectivity in meaning making and was therefore considered appropriate for the underlying theoretical underpinnings of this study.

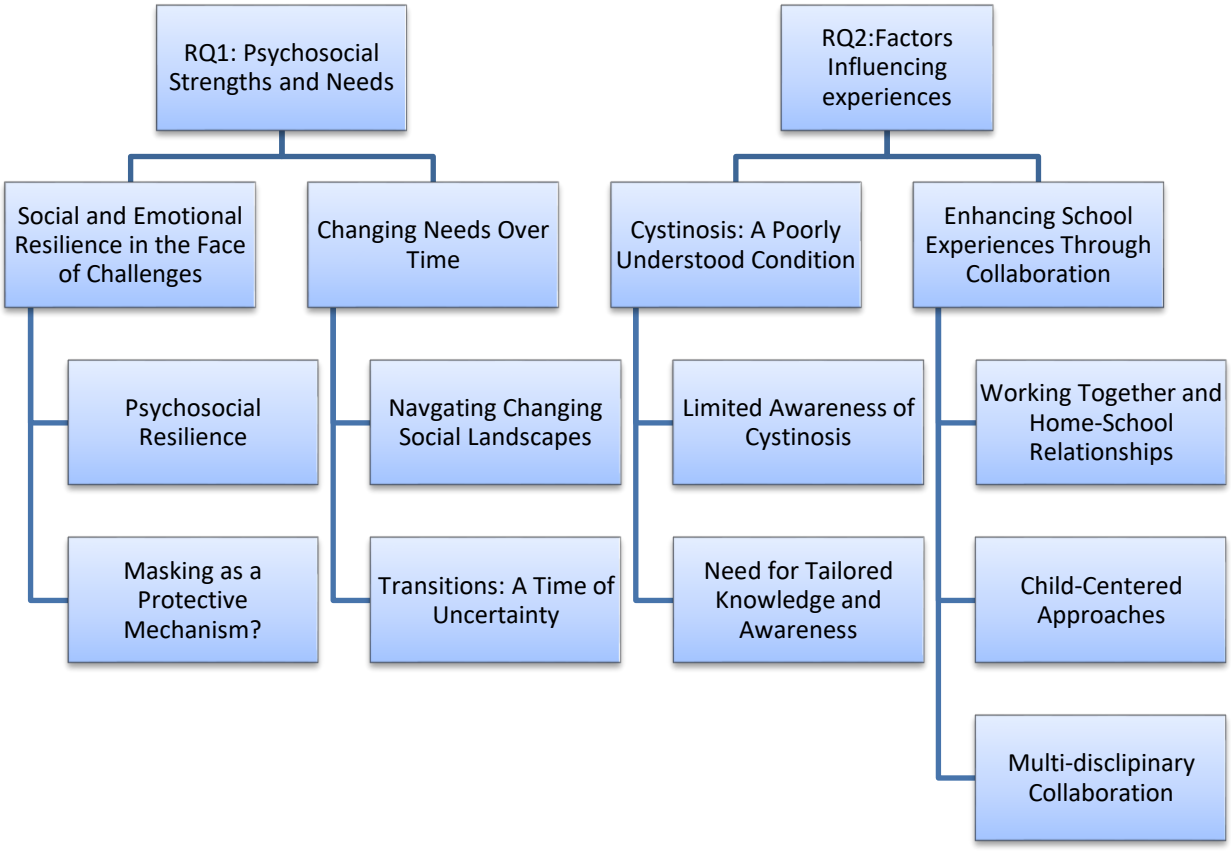
Braun and Clarke (2013; 2022) suggest six phases to thematic analysis which were used to guide the data analysis. Although each stage is distinct, the reflexive nature of the analysis called for flexible, recursive movement throughout the stages as the researcher's understanding and thinking developed (Braun & Clarke, 2022; Terry, 2017). The researcher utilised both electronic (NVivo) and hand-written (reflexive journal, mind-maps) tools as part of the analysis process to identify emerging thoughts and impressions. In the first phase of analysis, the researcher re-read each of the transcripts while simultaneously listening to audio-recordings. Each transcript was then read several times. Initial ideas, feelings, and thoughts were noted. Following the initial familiarisation phase, initial code labels were generated. Coding is considered an iterative, evolving process (Braun & Clarke, 2019; 2022) During the first coding phase, any data that was considered relevant to the research questions were coded. As the researcher's thinking evolved over time with deeper familiarisation with the data, codes were later refined and items re-coded. Codes that shared similar ideas or meanings were clustered into initial candidate themes. Distinct themes were identified based on central organising concepts (Braun & Clarke, 2022). Thematic maps were utilised to visually explore the relationships between themes and subthemes (Braun & Clarke, 2019; 2022). Supervision meetings which included reflection on codes, themes, and overall meaning were integral to guiding the process. Initial themes were reviewed and refined in relation to the full data set and research questions (Braun & Clarke, 2022). The researcher moved between stages in a recursive, reflective pattern until it was felt that the themes captured distinct and important meanings relative to the research questions. Each of the themes were subsequently defined based on the core centralising concept of the theme (Braun &

Clarke, 2022). Finally, a narrative account of the analysis was produced in phase 6.

Results

Four main themes were identified following the reflexive thematic analysis. Themes were grouped according to research questions and divided into subthemes as outlined in Figure 1. Descriptions of themes are provided, along with illustrative quotes from participants. As there was only one female child, all children will be referred to as "he" to protect anonymity. Quotes are referenced using a code which identifies them as a parent (P) or school staff (S) along with participant number. References to "classroom assistants" a term used in the Northern Irish school system were changed to "SNA".

Figure 1: Research Questions, Themes and Subthemes



Research Question 1

From the perspective of parents and school staff, what are the psychosocial strengths and needs of children with cystinosis?

Theme 1. Social and Emotional Resilience in the Face of Challenges

"He sort of takes it, owns it. You know, he owns the condition"

This theme, Social and Emotional Resilience in the Face of Challenges, encapsulates the psychological and social strengths demonstrated by the children despite the challenges experienced due to their condition while acknowledging participants' views that children's needs change over time as they face new challenges. Two subthemes were identified, Psychosocial Resilience and Masking as a Protective Mechanism.

Psychosocial Resilience

Participants highlighted the children's positive psychological wellbeing despite the many challenges their condition brings. Some participants noted that children had to manage health related responsibilities that were not typical for other children their age "*Who's doing my meds is a worry that a normal child wouldn't have"* (P3). One parent reported that their child's mood and behaviour was impacted by how they were feeling physically "*you know, whenever he's not feeling well, that he would like sort of lash out with words that you're going where did that come from or… But you know that it's because he's not feeling, you know, right."* (P5) However, in general the children's general mood was described in positive terms by all participants. One parent, commenting on her child's positive disposition, noted that "*he's always smiling you know even with everything he's been through.*" (P6) This sentiment was echoed by teachers as illustrated by one teacher's comment that, despite the child presenting with significant medical needs, "*He never comes across as being stressed, or anxious*", and "*he never complains.*" (S1) Overall, children's behaviour at home and at school was reported to be positive and typical for their age.

Parents and school staff emphasised that the children did not let their condition impact on their engagement with school or extracurricular activities, *"Some mornings he doesn't feel great, but he fights through it and like if he has to be sick, he goes into the bathroom, is sick, comes out and gets on with it."* (P5) A teacher provided a

similar description about another child, "*he's not someone who wallows and, you know, is feeling very sorry for himself. He's just… he gets on with the disability that he has, and it doesn't hold him back."* (S3) The majority of parents and teachers noted that children's school attendance was very positive, as reflected in this parent's statement*, "He actually would have good attendance. Like I think last year his attendance was over 90% which is really good.*" (P5) Two children had missed a significant amount of school in the past year due to hospitalisations and symptoms associated with cystinosis. One parent noted that their child had missed a lot of school during periods of ill health when he were younger,

> "*more so when he was younger, because for years we were in hospital at least once a month during the bad years. Now, I mean, it must be a year or two since he's actually been staying in hospital. But when he was younger, he did miss a lot of time because of that*. (P3)

One teacher felt while absenteeism would be unlikely to impact on a child in preschool it could be a cause for concern at primary level. Where children missed a significant period of school at a post primary level, school staff reported that this impacted on their ability to make friends, "*I think he's missed loads of opportunities to make friends, just because he hasn't been able to be here fully, and even when he is here, like he's not able to be here fully, because he's unwell in himself*"(S1) and their academic progress *"I would say because of how much he's missed out, and it's a significant amount of schooldays, he's not really able to reach that potential as such, you know"*(S1). For the remaining children, absences from school were typically due to hospital appointments, but where possible parents ensured that children returned to school that day, "*If mum takes him out for a medical appointment she tries to get him back into school for the rest of the day, so he's in school as much as he can be in school"* (S3). The majority of participants noted that children were not impacted academically by these short absences from school, but they did not like to miss out on social opportunities or special days as illustrated by this teacher "*say a visitor coming in or something and if he had heard about it, I think he'll feel that."* (S2) Reflecting on her child missing a no uniform day, this parent explained, "*it's only one day like he'll catch up on his homework*" but her child was upset to miss out on the

social aspect "*he's disappointed because you know, it's a fun day and he doesn't get the chance to go.*" (P6) In a similar vein, most participants reported that children disliked leaving the class for fear of missing out as illustrated in this comment from a parent, "*[Child's name] doesn't like being taken out anyway. Oh yeah. He hates to miss whatever the rest of them are doing."* (P3) Children's resilience and determination was also evident in their approach to sports with the children making great efforts to participate despite the physical challenges associated with cystinosis. "*He does take part in everything but he's just not strong enough. You know? That would be a big thing probably with him, you know? But he does put everything he has into it."* (P5)

Masking as a Protective Mechanism?

Some parents and teachers were surprised by the children's resilience and positive disposition, and wondered whether children were masking potential emotions, as highlighted by this parent:

> *"Is it normal that he's just so you know I was like, is he holding something in, can you find out is he like you know, dying inside because he's just so happy all the time, he never, he just does, goes with the flow, it's like yeah, whereas the others would complain."*(P4)

This sentiment was also echoed by teachers who noted that while the children appeared outwardly resilient, they questioned if there may be some underlying difficulties *"I would imagine surely it must… it must impact him, but he's not voicing that, do you know?"*(S1) Speaking about difficulties being selected for school sports teams, one parent noted that while her child *"still loves to go and try and try"* this can be emotionally challenging, "*We've already had tears about that. And I'd say, sure you don't really mind if you don't make the team. And he says, I sort of mind. And the tears would be rolling down his face."* (P3) Reflecting on concerns, one parent contextualised her concerns regarding wellbeing as being informed by negative descriptions of children's wellbeing in the literature, *"You're reading all these things that children also suffer from depression and it's terrible and you know this black deep future it's horrible"* (P4) rather than on her child's presentation, "*My fears, my fears absolutely, yeah. He has never, he's as happy as Larry."* (P4) One child in the

cohort was referred to primary care psychology services by their parents, but was discharged following screening.

Participants highlighted that it was important to the children that they felt the same as their peers as reflected in this parental quote, "*he doesn't want to be treated any different."* (P5) Some children made efforts to conceal their medical needs from their peers, for example, one parent commented that, "*he is private about it, you know he won't, if the friends were over for example he would go, come on into another room and do the meds, whereas normally here in the house it's just in the kitchen"* (P4) while parent 3 said, "*If he's with a group of people that he doesn't know, he would try to hide it".* Teachers reported similar strategies in school, as reflected in this comment:

> "*[Child's name] has requested this year that the bag be taken out when he's not in the classroom, when the other children are not in the class, that the bag itself is not visible, it sounds like as he's getting older he's obviously looking for it to be more discreet even than it has been before."* (S4)

For some children this was evident during social activities, where they would pretend to eat *"he'll sit down for every meal, he'll bring a lunchbox to school, he'll pretend to eat with his friends."*(P2) Parents noted that their children had developed these strategies independently as reflected in these statements, *"He has figured out it's something to hide or he wants to hide"* (P4), and another stating, *"that's something he did himself."*(P2)

Theme 2: Changing Needs Over Time

The theme, "Changing Needs Over Time" encapsulates parents' and teachers' reflections that while the children had many psychosocial strengths they acknowledge that children may face new challenges may over time.

Navigating Changing Social Landscapes

The majority of participants reported that the children had developed friendships in school, as highlighted in this comment from one parent, *"I don't really have an issue with him making friends, he's really social."* (P5) The majority of teachers noted similar positive peer relationships as seen in this comment from a primary school teacher *"He's definitely a very sociable boy. He's got lots of friends*

and is very well liked in the class" (P3). Some parents observed that although their child participated in structured and planned activities such as sports, they felt their children were sometimes excluded from more informal social activities, particularly as they got older. One parent noted that her child " *goes to all the training and everything but it's the sort of… The non-official things he misses out on."*(P3) Similarly, another parent noted that their child had noticed they were not always invited to activities due to their condition, *"He's like, they didn't ask me for sleepovers because I have to get so many meds'* (P6). Parents expressed concerns about their child encountering new social challenges as a result of their cystinosis as they approached adolescence, *"You know yourself as teenagers, sometimes when people fall out, they go for the jugular, what hurts most, and that could be the child's condition."*(P6) Parents and teachers noted that the children were becoming more aware and conscious of "*differences*" as they grew older. Commenting on this, one mother discussed how her typically resilient child was upset by this, *"Oh I'm different, I've a hole in my stomach.*" (P6) Similarly, teachers noted *" you know, he's short for his age and he's quite thin, you know, so physically I think he sees the changes that are going on."* (S3)

Transitions: A Time of Uncertainty

Parents and teachers reflected on both their past experiences and future concerns regarding the child transitioning through the school system, "*Secondary school is absolutely something I'm nervous and worried about."*(P4) Parents reported a variety of concerns about aspects of future transitions, including bullying "*it is when all the bullying could start"* (P4), *"I'm afraid he's going to get bullied or that he's just going to be so small and weak compared to the others."* (P3), academic attainment *"I was just thinking, oh my God, what's it going to be like for [Child's name] when he's doing his Leaving Cert? He'll never be able to sit there for three hours and write."*(P3) and independence, *"He has to manage himself and get from class to class and organise his books, I genuinely don't know if he has it in him."* (P4), to practical aspects such as balancing administration of medication with activities and classes *"they've PE days, there'll be the days they go out for activities. Like, the timing of all that will be key to whether he'll get sick after medication in school or not."* (P2)

Some parents discussed their fears that their child would not get a school place due to their medical needs, *"My biggest fear is that the school will have said no to him first day, that there's too much medical needs, that they wouldn't be able to cover it"* (P2) and *"We're just so grateful that he has school to go to."* (P3) Participants noted that despite their fears, transitions had been relatively smooth. One parent, reflecting on their child's transition into primary school stated,*" It was quite a straightforward transition he had."* (P5) while another stated, *"You read everything that they're all you know going to experience so much difficulty so I was really concerned but it didn't really transpire to be a thing so we just let it go."* (P4) A teacher, discussing a child's transition into school remarked, *"Not a bother. He told [Mother's name] and [fathers name] goodbye and took his school bag from them that morning."* (S2)

Reflecting on their child's transition into school, three parents and two teachers noted that schools were apprehensive about the child starting and how they, or the child, would manage the school day but the reality was more positive as reflected in this parent's statement*:*

> *"When we went on the first day and explained [child's name] to them, they'd the fear of God, which I don't know is that a good or a bad thing. We had to stay in the school because they were afraid to mind [child's name]and then they were like, actually there's nothing to him, he's just the same as everyone else"* (P2).

A teacher also openly recalled their apprehension, " *Definitely coming in we thought that, you know, he would be impacted, but by… he definitely doesn't let it kind of interfere with anything really, to be honest"(S3).*

For parents who were planning their child's transition into the next stage of education, they noted a general lack of information and clarity on how to manage this transition "*It's a minefield, I don't know how to… I don't know where to even start"* (P2). One parent discussed her initial meeting with her child's school and explained that she had nowhere to go to support her in answering the school's questions stating, *"All the questions the school had, I've no one to go and ask"* (P2). In keeping with this, another parent similarly reflected on how difficult it was to access

information and guidance when her child was starting school, "*I have a friend who's a teacher, who was able to say to me, oh no you need to get that a year before he starts, you need to do this and that, there's not a lot of guidance on for parents."* (P6)

Research Question 2

What do parents and school staff perceive to be the factors influencing the school experience of children with cystinosis?

Theme 1: Cystinosis: A Poorly Understood Condition

Participants provided insights into peer and school staff knowledge of cystinosis as being a significant factor in children's school experiences. While stressing the importance of awareness and understanding of cystinosis, participants also noted challenges associated with accessing relevant information and training. Two subthemes were identified, Limited Awareness of Cystinosis and Awareness as a Facilitator

Limited Awareness of Cystinosis

Both parents and school staff felt that although school staff had a general awareness of what cystinosis is, there was uncertainty regarding how well they understood the condition. Commenting on knowledge of cystinosis, one teacher stated, *"We only know a certain amount, and I'd say the amount we know is actually a fraction."*(S1) Similarly, this parent commented "*I don't know, being honest with you, how much they totally understand it."*(P4) Several parents felt that despite teachers' general knowledge of cystinosis, they lacked a deeper understanding of the individual impact that the condition has on the child, particularly outside of school. Parents noted that for many children with cystinosis they may be struggling with many adverse effects that are not visible in school, "*They see a fun-loving child that comes in the door, and they see nothing of what he actually does then when he comes home."* (P2) and *"If they only knew that he had vomited twice before he even got to school, you know."* (P3)

In relation to information and training, the majority of participants noted that schools had been given formal training regarding medical care needs *"that was only for the peg feeding, we didn't receive… you know, that was only for the peg feeding."* (S4) Participants noted that the responsibility for staff training regarding what

cystinosis is fell to the parents as discussed by this teacher, *"His parents actually came in and they trained us kind of how to, you know, feed, but they also came in with an information pack about cystinosis."* (P3) This parent described her role in training school staff , "*I did that then, myself and [father's name] went up and sat down with them and I kind of made out a pack. I know I'm not probably trained to do that now, but just an understanding sheet of cystinosis."* (P2) School staff also discussed researching the condition in their own time, "*I have to be honest that when we found out that we were having a child coming in with cystinosis, mine was Google. I just googled everything, tried to learn as much as I could, took notes, that was my learning, and that's how I learnt everything that I knew about (Child's name) before we started looking after him."* (S1)

In relation to peer awareness, the majority of participants noted that peers were not provided with any formal information about the child's condition from the school, *"I would imagine there are probably a handful of kids in the school that probably have a good level of awareness of what's going on from him, but that hasn't come through us by any means."* (S1) Participants noted that while peers were aware that the child had a medical condition, they were not aware of what the condition was, or how it impacted them "*They wouldn't really know what it is. They would just know that he's got something that he needs tablets for. I don't think they'd really know what the actual condition is."* (P5) Peer awareness of the child's condition came through general exposure to the child receiving medication in school, or in the case of three children, from the child showing them their gastric tube or telling peers themselves as illustrated by these participants, "*I know he has told a couple of the lads"* (P4) and *"They know that he gets medications with me, because he's told them that."*(S6)

Need for Tailored Knowledge and Awareness

All school staff discussed their desire to have more formal, accessible training about cystinosis. Teachers reflected on a need for broad training that included wellbeing, "*an overall wellbeing approach"* (S1), changing needs over time "*if there was some sort of a guide or something like…I don't know, for different ages, for preschool child with cystinosis and a child in junior school, in primary school but the*

junior side of it, the senior side in secondary school " (S2), communicating with parents, " *how to be able to broach conversations that may be difficult with the parents,"* (S1*)* and the potential impact on academic attainment *" the academic impact"* (S3*).* Teachers emphasised the need for general knowledge about the condition for all staff, "*more general and not specific to [Child's name], that we'd all have an understanding, and then like you were saying, that we would have an understanding of [Child's name] in Fourth Class 'This is what we should be looking out for."* (S4) as well as the importance of understanding the individualised nature of the condition as seen in this suggestion regarding training "*How it impacts children differently/individually, you know."* (S1) Teachers emphasised that having this knowledge early would be beneficial *"it would have been very beneficial on day one for… if someone had come and spoken about the disease and what to expect and what not to expect."* (S4), and that frequent continuing professional development would be beneficial *"more frequent training."* (S3)

Promoting school awareness was considered important for most parents. For this parent, it was important that the school understood the non-visible impact of the condition "*a full understanding of the impact it has on him, and that yes, physically he looks like the boy sitting next to him, but what's going on inside [child's name]'s body is completely different."* (P2) Another parent felt that increasing teacher awareness could prompt teachers to be cognisant of potential areas of difficulty "*I do because maybe they could watch out for things that are not coming up now."* (P4) Some parents noted that formal training would be helpful as it reduces the onus on parents to ensure schools are aware of the child's challenges as seen in this comment:

> "*I guess if they did have some form of training or just even a cystinosis information pack or something, you know? Because I don't want to be going down there kind of beating my drum trying to tell them. But I don't know. I mean, if they have 100 kids coming in all with different issues or whatever, they're not going to want to listen to every single mother who feels that her child has an issue."* (P3)

While parents viewed training as necessary, some parents noted a reluctance to highlight to their difficulties as they did not want their child to be treated differently

"*But I don't necessarily want them to know either because I want them to try and treat him the same. Because if they were saying, oh, [Child's name], poor sick little [Child's name]. You sit over here. I don't want that either, you know?"* (P3)

For some parents, as long as their children's medical needs were being met in the school context, they were unsure if additional training would be beneficial.

"I'm not sure, to be honest. I'm not sure, I think his SNA knows his medical needs and that's being met. I suppose other than a yearly kind of meeting with the vice principal, tick a box and fill in a form you know I don't think they can do anything." (P6)

In relation to formally informing peers, views varied between participants, with some feeling this would be very beneficial as illustrated by this mother "*I'd love his peers to have a level of understanding."* (P2) and teacher *"someone to come and talk to the class even about it, do you know?"* (S3) Some parents believed that providing formal training peers was not necessary unless the condition was impacting on the child's day to day experiences, "*Maybe if it were causing problems that would be the right thing to do but for us it just hasn't, he has slotted in so well."* (P4) or *"if the school was like if [Child's name]was being teased, I would think then maybe we would."* (P6) Participants felt that it was important that children and young people are given the choice as to who they told, particularly as they entered adolescence as reflected by this mother, *"now it's up to him who he wants to tell"* (P5) and this teacher *"If (Child's name) thought it would be, then like by all means we'd be more than happy to broach that with the kids."* (S1)

Theme 3: Enhancing School Experiences Through Collaboration

The importance of collaboration between parents, teachers, and children in promoting positive school experiences for children with cystinosis was emphasised. In relation to this area, three subthemes were identified: Home-school collaboration, Child Centred Approach, and Multi-disciplinary Collaboration.

Working Together and Home-School Relationships

Parents and teachers emphasised positive relationships, partnership and cooperation between parents and school staff as a facilitator to children's positive

school experiences. The positive relationships developed between home and school were reflected in this teacher comment "*the parents I think feel quite safe… they feel that (Child's name) is quite safe in our hands here in school"* (S4) and this parental comment "*I know there's always somebody there who cares as much as I care."* (P5) They spoke about working together to overcome barriers as highlighted by this teacher "*any problem that we've kind of been faced with has always kind of… we've kind of come around it, whether the teachers have come up with a solution, or whether his parents kind of have come up with a solution, we all kind of just aim to kind of make sure that he's included and that he's not feeling left out".* (S3) Parents echoed this sentiment, as seen in this statement "*you work with them. You don't work against them"* (P5). In working to support children's engagement in school, participants identified strategies parents implemented, for example, making changes to medication schedules to reduce the impact on the child's school day, despite this meaning they would *"take the brunt*" of the medication administration and associated side effects at home or attending sports activities to cater for medical needs.

Open and clear communication was seen as key to this effective collaboration by both parents and teachers. This parent described how communication was a facilitator *"I think it's just us building a rapport with them, knowing that we're approachable as parents and that they're approachable as a school."* (P2) Similarly, this teacher discussed the ease with which they could contact parents to discuss solutions if needed, *"Yeah, if there's something in it changes, or something that might not run to how we'd normally run in school, [SNA's name] would ring home and check with Mam how we could work around that."* (P4)

Parents and school staff typically communicated informally, as needed. This informal communication occurred primarily with the support staff (e.g. SNA, classroom assistant). Support staff were considered fundamental facilitators to both successful collaboration and inclusion of the child as illustrated by this parent "*if he was sitting in school and was just being left, obviously maybe he wouldn't be in as much."* (P5) Parents referenced the positive impact of support staff in relieving parent stress as illustrated in this statement *"because the SNA is so brilliant, we don't have to worry".* One teacher noted that having an SNA in the classroom meant that they

could focus on their teaching *SNA manages the medical side of it, which allows me then to just be their teacher."* (S4) Another teacher reported that having dedicated support staff in the classroom was more inclusive, and resulted in less time out of the classroom for the child, *"he'd be leaving the classroom all the time, do you know? Whereas it's just handier for the SNA to leave so he's not missing out on the classwork."* (S3) Participants also referenced the positive impact of support staff for children as they have someone they can trust and go to if they are feeling unwell. Some participants also acknowledged that the presence of an SNA could act as a barrier to social integration and inclusion as seen in this comment "*where is the opportunity for other kids to be able to engage with him then, if there is an SNA next to him."* (S1) Similarly, the parent and SNA of one child discussed how the child was "*attached"* to the SNA and it was felt that this impacted opportunities to initiate play with other children. Highlighting the importance of communication, the child's parents emphasised that this concern was resolved through open discussion with the school.

Despite the positive relationships and rapport, some parents also reflected on challenges and barriers associated with advocating for their child's needs, *"as a parent you just have to not be afraid to voice your concerns and raise and be assertive and just remember that you're the voice for your child and advocate, which sometimes can be difficult, if it's not in your nature."* (P5) Another parent felt that a liaison person would be a helpful support at times like this "*if I were concerned that he's not being looked after well enough or he's not keeping up or he's not happy, I'm so thankful I haven't had to do it, I just know I would be quite nervous to do it, so maybe if there were an intermediary maybe there between the school it would be a lot easier."* (P4).

While medical care plans, which all of the children had, also facilitated communication and collaboration, these were solely in relation to medical needs. Two children had student support plan but their parents were unsure of their purpose or content. While all parents met formally with the school through, for example, parent teacher meetings, the format and content of what was reviewed in meetings or plans varied between schools. This parent noted

"*Every year at the start of the year they ask us to come in just for a very quick meeting. It might be like, ten minutes and then they get us to sign something and to be honest, we don't really know what we're signing or… I've never investigated what we're entitled to or anything like that. As long as [Child's name] is happy and they're happy with him and I say, oh yeah, that sounds fine", and I sign."*(P3)

One SNA specified that while discussions in care plan meetings were centred around the child's medical needs, their psychosocial wellbeing was also discussed. Another parent noted that "*the word cystinosis has never come up*"(P4) in parent teacher meetings.

Child-Centred Approaches

Parents and teachers spoke of the importance of respecting children's choices and autonomy while providing encouragement and support as needed. They discussed instances of how they followed the child's lead in relation to how they organised and provided assistance in school, for example, in relation to participation in school outings, this teacher noted *"It could be up to him, but like as a standard across the board for us in terms of procedure, like any kids that have medical needs, or additional needs, we always have a Support Teacher that is like linked with that child on an outing. So, you know, they would be fully supported whilst they're out of the school and participating in an activity… and it would not be kind of forced, or strong-armed into participating either. They could be there as an observer."* (S1) Another teacher noted *"we always follow his lead and the request. It's always like, you know you help me with how we do this, that's how we always conduct our day."* (S4)

Most of the children were reportedly not directly involved in setting goals or formally asked their perspectives. Discussing her child's involvement in his care plan, this parent stated "*I don't think he'd even know there is one*" (P4). One parent noted that a child's care plan was updated yearly and the child's views were gathered as part of this process "*it hasn't really changed over the years on his medical needs and then [Child's name]is given a form to fill in you know what he feels is going good you*

know we've just bit of what he thinks is going strong and stuff and then they send home form for us to fill in and then we just meet and discuss and tick boxes"

Parents and teachers highlighted the importance of maintaining the balance between self-determination and encouragement, recognising when to allow children to make their own decisions and be guided by them, and when to step in and encourage children to take part, to try new activities, and to meet their potential in school. This parent reflected on this saying

> *"He'd come in and go, oh, I'm so tired, mam, I must sit down and if you let him sit down for the whole evening, he will sit down for the whole evening. You know? It's very much like you can't let him slide down like that. You have to say, come on now, [Child's name], get out the homework quick. And on we go".* (P3)

School staff also acknowledged this balance saying, *"there's a really nice family dynamic. Yeah, and it's no nonsense at the same time. It's like 'Get on with it.' you know? you know 'Come on, we can do it. Let's go."* (S4) This parent recounted instances where they needed to motivate their children to overcome challenges with a solution-oriented thinking:

> *"he'll say I can't do that, and I'm like why can you not do that and he says because of my condition and I'm like oh so I try and say but why, why are you thinking that and I'm like oh sure what can we do to make it easier, so I'm always trying to promote him like, don't be looking at what you can't do, there's solutions."* (P6)

Mult-disciplinary Collaboration

Parents and teachers noted a general lack of consultation and support from external agencies in relation to children's needs in school. The majority of participants noted that there had been no direct involvement from allied health professionals such as occupational therapy, speech and language therapy, or psychology services, including the National Educational Psychology Service (NEPS). This teacher reflected on how school staff collaborate and share knowledge with each other, but receive no additional guidance or support in school specific to the child *"If we're working with him, we would just be telling one another how (Child's name) is doing that particular*

day, but other than that we have nothing from outside that's helping us." (S1) This parent described parent and teacher doubts about how to best meet the child's needs in school without support and consultation from allied health services *"they might ask me what do I think or am I happy with this or whatever. But because I don't really know either what he needs, we're kind of muddling along."* (P3)

Where resources and recommendations were provided, this information was passed on by parents to teachers rather than school staff being directly consulted "*His parents took him maybe to someone, maybe to do like this food therapy, and this was the recommendation that we got, so yeah, as a result then we kind of started doing it in his resource."* (S3) While one child accessed tuition in the hospital, the school noted that there was no link between hospital and school staff, which the school would have welcomed.

Parents and teachers felt that consultation with allied health or medical staff would be beneficial as stated by this teacher:

> *"Having some kind of contact from the hospital, or a GP, or somebody that is working with him, to know, you know, if we're doing everything that we should be, or if there's more we can be doing, or even just in terms of how he's progressing."* (S1)

Participants also highlighted that support should be responsive to a child's need and acknowledged that not all children will require the same level of support, *"he is doing so well but yeah if things were different or you have a child who is I suppose not managing as well, absolutely."* (P4)

Discussion

While research suggests that children with cystinosis are at risk of psychosocial difficulties (Aly et al., 2014; Ariceta et al., 2015; Atia et al. 2022; Delgado et al., 2005; Spilkin & Ballantyne, 2007; Ulmer et al., 2009), it does little to contextualise these risks or, crucially, to identify what can be done to mitigate them. Despite the fact that many children with cystinosis attend school on a regular basis, there is very limited research to date that explores their experiences in this context and none that includes the perspective of school staff. The current qualitative study aimed to explore children's experiences of school and in particular their strengths and needs in order to

identify the challenges, barriers and facilitators in the education system from the perspective of their parents and teachers. Bronfenbrenner's PPCT model provided an illustration of the complex interactions between individual and contextual processes over time. The findings from this study are illustrated within this model in Figure 3.

Figure 3: Findings applied to Bronfenbrenner's PPCT model

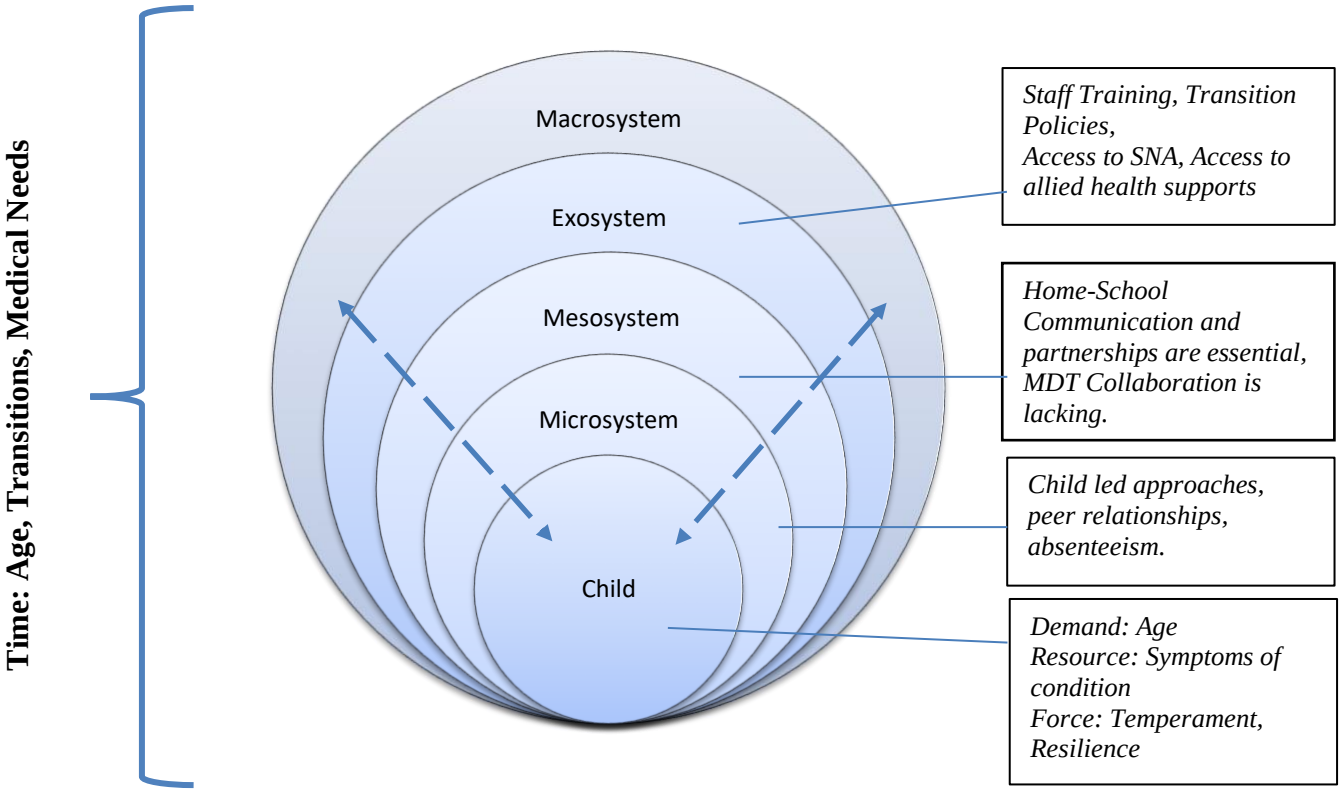


Figure adapted from Murtagh & Seoige, (2022)

Children are Resilient but Face New Challenges Over Time

For the children in the current study, parents and teachers identified many individual social and emotional strengths which were not emphasised in previous studies including sociability, good relationships with peers, positive behavioural and emotional dispositions, and resilience. Relating this to the PPCT model, various "person" characteristics (Bronfenbrenner & Morris, 2006) warrant consideration. Acknowledging that children do not develop in isolation, Hayes (2017) notes that what is important about these person characteristics is that they both shape and are

shaped by experiences. At a “demand” level, age is an important consideration for the current cohort as they ranged in age from 4-16. Hayes (2017) states that children’s age influences how they experience the world. The demands present in the environment and the proximal processes (e.g. interactions with peers or teachers) at play are likely to have varied at different ages. At a “resource” level, all of the children were impacted by the biological and physical effects of cystinosis and its associated treatment. Some children experienced higher levels of medical need including dialysis and more demanding treatment regimes, which may have impacted on proximal processes. At a “force” level, all the children showed significant social and emotional resilience in the face of the significant demands and challenges presented by their medical condition.

Despite the strengths identified, a complex interwoven sense of change was present. Wellbeing can be considered a fluid state, and may change depending on personal needs, risks, and protective factors (DES, 2019; Nohilly & Tynan, 2022). In the current study, participants identified adolescence, educational transitions, and times of higher medical need as critical periods of increased need. Bronfenbrenner’s PPCT model provides a lens for understanding these developing changes. Proximal processes constitute progressively complex and reciprocal interactions between the developing person and other people in their environments and are described as the “engines of development” (Bronfenbrenner & Morris, 2006). At a child-peer level, children in the current study were described as highly sociable and the majority had developed good friendships. While these friendships may have operated as facilitators to wellbeing (Tynan & Nohilly, 2021), more research on the importance of peer relationships for children with rare diseases is needed (Runions, 2020). In terms of “time”, parents reflected on concerns that their child may experience bullying or social isolation in adolescence, particularly as they moved from the relatively close knit environment of primary school into post primary school. The adolescent period ranges from age 8 to 19 (Salemela-Aro, 2011), a time during which children experience physical, psychological, and emotional changes, alongside changes in school contexts such as change of support staff, school, and changes in peer relationships. Positive peer relationships and close friendships are central to a young

person's adjustment at this time (Brown, 2004; Poulou & Norwich, 2019). However, for some children, friendships that develop during primary school are at risk as children attend different schools, classes, and meet new friends. Participants in the current study emphasised children's strong desire to feel the same and be treated the same as their peers and reported that children were becoming more aware of their "differences" as they got older. Children disliked when their medication schedules or medical appointments interrupted social activities or resulted in them "missing out" on aspects of the school day. Physical symptoms of the condition such as tiredness and weakness impacted on sporting ability. For some children in this cohort, there was a developing sense of being excluded from unstructured social activities due to their medical needs. While some children openly revealed their concerns to parents for others, this was seen in an increasing effort to mask aspects of their condition.

For the majority of children in this study their condition was relatively stable and well managed. Ariceta et al. (2015) reported that adults with cystinosis report a greater negative impact on quality of life than children, and posit that this is likely related to the progressive nature of the condition. In the current study, parents and teachers were able to navigate challenges to ensure that children were able to take part in activities and their school attendance was not hugely impacted which may have been a strong factor in facilitating and supporting their wellbeing (Valero-Moreno et al., 2020). A sense of connectedness and school belonging is an important aspect of wellbeing (Arslan, 2021; DES, 2019). However, where children did miss a significant amount of school or their participation in activities was curtailed due to their medical conditions, participants noted that this impacted their friendships and academic progress. It is acknowledged that there was only one participant in the current study who experienced significant absenteeism beyond preschool/early primary where it was considered that absenteeism would not have had such a significant impact. Nonetheless, this is an important consideration for schools and families as children's school attendance will be dependent on their medical needs which may change if the child's condition deteriorates or necessitates more intensive treatment (e.g. dialysis, kidney transplants). The National Institute for Health and Care Excellence (NICE) guidelines identify support with educational transitions and

other life changes as being integral to social, emotional and mental wellbeing in primary and secondary education (NICE, 2022). It is crucial that parents, teachers, and allied healthcare teams are aware of potential challenges experienced at these crucial times so that support can be put in place.

Knowledge and Awareness as a Facilitator

Teacher knowledge and awareness of cystinosis was identified as an important factor in the experiences of children in school. These results are consistent with previous studies that have identified teacher knowledge, initial teacher education, and professional development opportunities as essential in promoting positive school experiences for children with medical conditions (Adams & Bourke 2023; Nabors et al., 2008; Verger et al., 2020). Teachers emphasised that continuing professional development should encompass a broad awareness of the condition, its associated side effects, and the potential impact on wellbeing and academic attainment at different time points. In line with other research on chronic illness and rare disease (e.g. Adams & Bourke, 2023; Paz-Lourido et al., 2020) teachers specified that broad knowledge should be combined with understanding at the individual child level. Crucially, school staff noted that training should be provided as early as possible so that teachers can be vigilant and mindful of areas of need, allowing them to observe, respond, and adjust to children's needs as appropriate. There was a sense of apprehension and nervousness from schools when they first learned of the child's medical needs. Increased knowledge has been found to increase teachers' confidence in supporting children with rare conditions in school (Nabors et al., 2008) which may in turn decrease apprehension and concern. Parents highlighted that the non-visible impact of cystinosis such as nausea, dehydration and tiredness are not always evident to teachers and peers and they are not aware of the day-to-day impact of the condition outside of school. When children do not present with visible symptoms or observable difficulties, there can be a risk that they do not receive appropriate support (Paz-Lourido et al., 2020; Verger et al., 2021). A study by Sommer et al., (2024) reported that online seminars for both initial teacher education and professional development leads to greater awareness of rare diseases and increases teacher competence and confidence in supporting affected students. The

teachers in this study noted that online webinars such as those available for children with epilepsy and diabetes would be easily accessible and welcomed.

It was interesting to note that some parents did not feel that increased knowledge or awareness for school staff and peers was necessary, particularly when the child's condition was well managed. In terms of bioecological theory, health can be considered a "person" characteristic that shapes how teachers and peers may respond to children. Participants in this study emphasised that it was important to parents and children that children with cystinosis were not treated differently to their peers. This underscores the importance of sensitive, family centred home-school communication that respects the confidentiality and wishes of the child. For children with medical needs, there may be critical time periods where their condition will necessitate differential treatment. Researchers have posited that reluctance to disclose diagnosis may indicate need for greater focus on inclusivity, awareness, and diversity (Verger et al. 2020). This was not examined in the current study but could be an area for future research.

Collaboration is Key, Relationships Matter

At the microsystem level, parents and teachers reflected on child-school staff and child-parent and relationships. The teacher-student relationship has been identified as one of the most influential variables in determining children's sense of belonging in school (Ibrahim & El Zaatari, 2020). Bronfenbrenner identified the teacher-student dyad as essential for child development (Bronfenbrenner, 1979). Bronfenbrenner suggested that "the developmental impact of a dyad increases as a direct function of the level of reciprocity, mutuality of positive feeling, and a gradual shift of balance of power in favour of the developing person" (Bronfenbrenner, 1979, p.59). In relation to the current cohort, the relationship between the child and their special needs assistant was highlighted, with participants noting that the child had someone they could trust to go to if they were feeling unwell and that access to an SNA facilitated inclusion in school. Some participants noted that although essential for medical needs, the presence of an SNA could act as a social barrier and impact independence. One parent-school dyad reported that access to an SNA had led to a degree of social dependence for a child, but they were able to work together to

overcome this barrier through open communication and the SNA stepping back during social activities. This finding underscores the importance of communication, while also highlighting the importance of balancing the role of the SNA with also promoting independence (Griffin & Blatchford, 2021).

School staff highlighted the importance of listening to the child's preferences with regards to administration of medication, peer awareness of their condition, and participation in activities. This was an encouraging finding in line with key principles of the Wellbeing Policy and Framework for Practice (DES, 2019) and the Junior Cycle Wellbeing Guidelines (NCCA, 2021) which encourages schools to adopt a child-centred approach, including respecting and valuing the voice of students. Previous studies have highlighted the importance of including children with rare diseases in decisions with healthcare teams (Belzer et al., 2022), however little is known about how this is achieved in schools. In the current study, parents and teachers reported incorporating children's preferences into their school day (e.g. altering medication schedules where possible). Parents highlighted a similar child-centred approach that included encouraging the child to overcome barriers, while also respecting their wishes and independence. Despite acknowledging the importance of following children's lead and including their preferences into their school day, children were not directly included in decision making or processes. While the rationale for this was not explored in the study, age and developmental levels as well as complexities and lack of training in eliciting the voice of the child have been cited as perceived barriers to eliciting the voice of the child (Griffin, 2021). Future research could focus on how school teams integrate both the voices and choices of children with rare diseases such as cystinosis into school planning and decision making.

Collaboration at the mesosystem level was also identified as a key facilitator to children's school experiences. Positive and trusting parent-school relationships are known to have a significant positive effect on children's wellbeing (Minke et al., 2014; Santiago et al., 2016; Sheridan et al., 2017). The National Council for Curriculum and Assessment (NCCA) also emphasises positive relationships and communication with parents as integral to wellbeing (NCCA, 2021) and it was clear that the experiences of parents and school staff aligned with this. For the participants in the current study,

clear and open parent-school communication was vital to this relationship. School staff and parents stressed the importance of working together to find solutions to challenges that ensured the child could be included in activities to the best of their ability. The ability to communicate with the child's support staff (i.e. SNA/classroom assistant) was an important feature of positive home-school communication.

Despite the positive relationships identified between home and school, some parents explained that advocating for their children's needs may be challenging should the situation arise. Similarly, one teacher noted that it would be helpful to have more information on how to have "difficult conversations" with parents. For participants in this study communication appeared to occur primarily through informal conversations. This is an important consideration for post-primary school, where identification of a primary liaison person can be more challenging (Verger et al., 2021). For children in the current study, formal planning tended to be "front loaded" at the start of the education journey. Participants felt confident that parents and teachers alike would highlight concerns should they arise, but these were not regularly reviewed or discussed. Other research has highlighted the importance of monitoring wellbeing of "at risk" students to identify early signs of emerging difficulty and timely support (Hooper et al., 2022). Including discussions regarding children's psychosocial experiences and wellbeing into students' care plans may provide a practical, formal way to ensure that these areas are monitored.

Parents and teachers alike identified a lack of coordination between allied health professionals (e.g. psychology, occupational therapy, medical teams) and school as a challenge. Although some children had accessed allied health supports or hospital education services, these services did not directly consult with school. Some parents compensated for the lack of coordination by passing on relevant information to teachers and support staff. A lack of coordination between healthcare providers and schools has been reported internationally (Lum et al., 2017; Paz-Lourido et al., 2020; Tumiene et al., 2022; Verger et al., 2020; 2021). Verger et al. (2021) suggest that health care that does not consider the impact of school and focuses solely on therapeutic interventions does not consider health as a whole. Teachers and schools play significant roles in children's development that goes far

beyond academic attainment. Given the complexities of rare diseases such as cystinosis, a coordinated multidisciplinary approach with a communication protocol is essential, and it is crucial that the role of schools is valued and included in this care (Tumiene et al., 2022; Verger et al. 2021). As noted previously, the needs of individual children will vary across time and it is likely that children will experience critical time periods (e.g. during hospitalisations, transitions) which will necessitate an increase in collaboration. Balancing a focus on the specific needs of the disease and any associated 'medicalisation' of the child against the benefits of the establishment protocols and policies to support collaboration and communication is pivotal. Given educational psychologists' knowledge and training of child development, educational systems as well as the role of interrelated contexts and processes, they would be well placed to support the link between education, health/social care, and the home contexts.

Limitations

It is important to acknowledge the limitations of the current study. Of particular significance is that the perspective of children with cystinosis is not included. Previous research has found that parents and children with cystinosis can have different views on the psychosocial impact of cystinosis (Delgado et al., 2005). While the results of this study underlined the importance of eliciting the child's voice, this research forms part of a wider research study where children's views on school were captured which will add this crucial component to overall understanding. An additional limitation of the current study is that the views of health care staff were not captured. As collaboration with allied health professionals was identified as a barrier, the voice of clinicians and in particular psychologists may have broadened our understanding of the children's wellbeing strengths and needs as well as the role of a systemic based consultation within the school setting for supporting children with rare diseases. As noted by Paz-lourindo et al. (2020), a greater understanding of health professionals' perceptions on the meaning of school for children with rare diseases and the role of school in their development is important to facilitate coordinated care. Nonetheless, the research provides a detailed overview of the perceptions of parents and teachers regarding the factors influencing the school experiences of children with cystinosis. To the

researcher's knowledge, this is the first study to include the views of teachers supporting children with cystinosis.

Conclusion

Previous research has indicated that children with cystinosis are at risk of social, emotional, and behavioural challenges (Ariceta et al., 2015; Aly et al., 2014; Delgado et al., 2005; Spilkin & Ballantyne 2007; Ulmer et al., 2009). School experiences play a significant part in the wellbeing of children. Despite this, there is little known about school experiences of children with cystinosis. Moreover, there is no research to date that examines the factors influencing their experiences from the perspective of the school staff. Underpinned by Bronfenbrenner's PPCT model, the current study highlighted the complex interactions between individual and contextual processes over time and how these may impact on children's experiences. The research points to the importance of collaborative and individualised support which recognises that children's needs will ebb and flow between periods of relative consistency and times of critical need such as adolescence, educational transitions, times of increased medical need. There is a need for increased knowledge at a broad level, combined with an understanding of the individual needs of the child in the context of school. It is important that children are treated the same as their peers, and schools work alongside children and parents to find solutions to barriers to ensure that children can participate in school and extracurricular activities to the best of their ability. Schools would welcome clear links with health professionals and allied health supports and a clear pathway to access this additional support when needed. The findings offer a contribution to our understanding of the psychosocial needs of children with cystinosis in school. Adopting a PPCT lens emphasised the interrelated and changing biological, psychological, and social needs of children with cystinosis and the need for co-ordinated multi-level support across contexts. The findings are therefore important for educational psychologists, teachers, and healthcare teams.

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Chapter 5: Implications for Practice

Introduction

Rare Diseases Ireland defines rare diseases as a disease or disorder when it affects less than 1 in 2,000 individuals (Rare Diseases Ireland, 2024). The impact of rare diseases stretches beyond the physiological aspects of these conditions (Kole et al., 2021; Eurodis, 2017). The importance of understanding the broad impact of living with rare disease has been emphasised in recent policies (DoH, 2014) and research (Belzer et al., 2022; Verger et al., 2020;). The rare disease community have reported that psychological and social impact of living with a rare disease is not routinely considered as part of their overall care and support and have called for an increased emphasis on the day to day challenges experienced (Kole et al., 2021). From an Irish context, a participatory multiphase design study by Somanadhan et al. (2020) identified the psychosocial impact of living with a rare disease as a top research priority for people with rare diseases in Ireland.

Approximately 70% of rare diseases have their onset in childhood, a time of critical development (Nguengong et al., 2020). For many children with rare diseases, medical advancements and increased survival rates mean many children can participate in mainstream education (Sommer & Klug, 2024). Schools provide a setting for children that may exacerbate or alleviate challenges to wellbeing for children with medical conditions (Runions et al., 2020). Children living with rare diseases have to navigate several challenges within the school system however including difficulties with school attendance and participation, stigmatisation, bullying, and lack of teacher knowledge about their illness (Adama et al., 2023; Kole et al., 2021; Foster et al., 2023; Somanadhan et al., 2023). At a national level, Ireland's National Rare Disease Plan highlights the importance of educators understanding the complex impact of children's condition on their educational experiences and mental health (DoH, 2014). Internationally, a recommendation from the 2030 Rare Disease Foresight study was to increase the quality of education through spreading knowledge of the impact of rare diseases to educators and the next generation (Kole et al., 2021).

The current thesis focuses on cystinosis, a rare, chronic, multi-system disorder which causes an accumulation of the amino acid cystine in the organs and tissues of the body, causing widespread tissue and organ damage (Doyle & Werner-Lin, 2015; NORD, 2023). Current estimates indicate that cystinosis occurs in approximately 1 in 200,000 live births within developed countries (Cystinosis Ireland, 2024). The condition is characterised by a demanding life-long treatment regime and frequent medical interventions and hospital appointments. Management of the condition is complex and requires treatment with several drugs with a strict dosage schedule (Ariceta et al., 2016; Ariceta et al., 2019). A primary treatment for cystinosis is cysteine depletion therapy (Cysteamine) which has an unpleasant smell and taste and side effects include nausea, vomiting, gastric reflux, and an unpleasant odour from the body and breath (NORD, 2024; O'Connell et al., 2022). Many children have difficulty consuming enough calories to support their nutrition due to insatiable thirst resulting from kidney defects, and frequent vomiting. Placement of a gastronomy tube (G-tube) or jejunostomy tube (j-tube) may therefore be required to support nutrition and the administration of medication (Cystinosis Research Network, 2018). Complications of cystinosis can include extreme light sensitivity (photophobia), hypothyroidism, heat exhaustion due to impaired sweating, short stature, and muscle weakness (O'Connell et al., 2010; Nesterova & Gahl, 2013; Wilmer et al., 2011). People with cystinosis will develop end stage renal disease (ESRD) where dialysis and kidney transplantation are required; however, early treatment combined with strict adherence can delay ESRD (Ariceta et al., 2019, Nesterova & Gahl, O'Connell et al., 2010). While essential research into the medical treatment of cystinosis continues, in line with shifts towards a broader more holistic understanding of rare diseases, attention has also shifted towards the psychosocial impact of living with cystinosis. Underpinned by Bronfenbrenner's PPCT bioecological theory of development (Bronfenbrenner & Morris, 2006), the overall aim of this doctoral thesis was to explore the psychosocial impact of living with cystinosis for children, and to identify factors influencing their school experiences. This was achieved through the completion of two papers, a systematic literature review (SLR), and an empirical journal article (EJA). The findings from these studies have important implications for

education and psychology. This chapter outlines the results of this research and highlights how the findings may contribute to practice for educational psychologists and other stakeholders supporting children with cystinosis such as educators and multidisciplinary staff.

Summary of the Doctoral Research

A systematic literature review (SLR) was conducted to provide an overview of current research pertaining to the psychosocial and educational impact of cystinosis for children and young people. A second aim of the SLR was to identify gaps in current knowledge in order to inform the empirical research. Results of the SLR revealed a dearth of knowledge on the psychosocial and educational strengths and needs of children and young people with cystinosis, particularly in the school context. The research indicated that children and young people with cystinosis are at risk of psychosocial difficulties (Atia et al., 2022; Delgado et al., 2005; Spilkin & Ballantyne, 2007; Ulmer et al., 2009). There was a significant lack of qualitative research giving context to difficulties identified. Although quantitative measures are widely used in research and professional practice as a measure of emotional and behavioural adjustment, some researchers have raised concerns about their use for children with medical conditions. Perrin (1995) cautions that the items on the social competence scale of the CBCL assesses children's participation in social activities which may be limited by virtue of the child's condition, and should not be interpreted as indicating that the child is less socially competent. The closed-end nature of the questions on such measures may result in the loss of rich, person focused information which can only be gathered by discussion with those with more direct knowledge of the experiences of children with cystinosis (Spilkin & Ballantyne, 2007). As such, there is a need for qualitative research that can provide a detailed, contextualised account of children's school experiences and potential risk and protective factors. The voice of school personnel was notably absent, despite the significant role that school and school staff play in children's school experiences and wellbeing (DES, 2019; Lum et al., 2017; Tynan & Nohilly, 2021).

The empirical research aimed to address these gaps by exploring parents' and school staff perception of strengths and needs of children with cystinosis and factors

influencing their school experiences. Overall, the results highlighted the importance of collaborative and individualised support which recognises that children's needs will ebb and flow between periods of relative consistency and times of critical need. There is a need for increased knowledge at a broad level, combined with an understanding of the individual needs of the child in the context of the school system. It is important that children are treated the same as their peers, and schools work alongside children and parents to find solutions to barriers to ensure that children can participate in school and extracurricular activities to the best of their ability. Schools would welcome clear links with allied health professionals and supports and a clear pathway to access this additional support when needed. The role of educational psychologists (EP) is that of a scientist-practitioner who utilises psychological skills, knowledge, and understanding to support the needs of children through consultation, assessment, intervention, research and training at an individual, group, or organisational level (Fallon et al., 2010; Kennedy, 2009). In line with the broad role of an EP and the bioecological perspective of this research, implications for practice were evident across contexts.

Microsystem Level: Placing the Child at the Centre

Bronfenbrenner identified the microsystem as the direct environments in which children have immediate contact with other people, objects or symbols (Bronfenbrenner & Morris, 2006; Tudge et al., 2017). The school environment was the primary focus in the current study. In Ireland, support for children's psychosocial wellbeing in school is guided by the Department of Education continuum of support model (DES, 2019). Within this model, support is divided into three tiers: whole school and classroom support for all, school support for some, which includes identification, targeted prevention and early intervention for those at risk and, school support for few which includes targeted intervention for pupils with complex and enduring needs (DES, 2019).

In relation to the children in the current study, parents and teachers emphasised the children's resilience and how, despite the additional challenges and at times the limitations presented by their condition, they were generally happy and well-adjusted. However, the results also highlighted that children with cystinosis may

face increasing challenges over time. Some parents and teachers noted that they were unsure if children were experiencing psychosocial difficulties which they were masking and not voicing. The NICE (2023) social-emotional and mental health guidelines for schools noted that it is important that schools are cognisant that some children and young people may internalise their distress and can be more difficult to identify. Despite the children's outward resilience, the results suggest that it is crucial that educators and allied healthcare professionals are aware of potential risks to the children's wellbeing and actively monitor wellbeing overtime and particularly at times of transitions during the school years. In terms of student voice, parents and school staff reported that some children voiced their preferences informally rather than being directly included in planning meetings, particularly in relation to their care plans. In line with Lundy's (2007) model of participation and the rights of children to participate in decision-making and education rights, the use of resources and psychological tools to facilitate parents and teachers to access student voice are recommended so that children can be provided with appropriate methods of communicating their emotions and feelings. Despite the recognition of the importance of pupil voice at a policy level (e.g. DCYA, 2019), there is a lack of guidance and training for school staff with regards to how to practically elicit and include the voice of the child (Griffin, 2021). Psychologists can play an integral role in both advising about and facilitating this process.

Adopting a Person Centred Planning (PCP) approach where the child is directly involved in planning school supports is one way that children could be given additional opportunities to share their thoughts and feelings and directly impact how support is provided in school. PCP focuses on identifying individual strengths rather than deficiencies, placing individuals at the centre of planning and decision making, and co-constructing solutions (Gray & Woods, 2022). This process involves gathering information centred around the child, exploring what is working and not working from different perspectives, and generating goals and actions (Kilbane & Sanderson, 2004; Gray & Woods, 2022). While additional research on the use of PCP in improving outcomes and school experiences for children with rare diseases is warranted, PCP has been suggested to improve relationships and interactions at both the

mesosystem (e.g. parents, teachers) and micro-system levels (child-teacher) levels (Gray & Woods, 2022). Additionally, when considering critical time periods identified in this study, the role of PCP may be particularly beneficial at times of transition (Hayes, 2004) and returning to school following periods of absenteeism (Corrigan, 2014). EPs are ideally suited to act as both facilitators in PCP meetings and in training and empowering others to utilise person-centred practices within educational settings (Corrigan, 2014; Gray & Woods, 2022).

These findings also have specific implications for practice for EPs as they have expertise in assessment and intervention in mental health and can facilitate dialogue with children and assess psychosocial wellbeing in a developmentally appropriate way. Psychoeducation for parents and teachers regarding adolescent mental health, resilience, and coping strategies with a special focus on rare disease could also help to address and alleviate some of the concerns expressed by participants. Anderson et al. (2003) suggest that psychologists working in school settings can provide vital support to children with chronic illness through providing intervention in areas such as problem solving, educational intervention strategies and coping strategies.

Bridging the Mesosystem Gap: The Consultative Role of Educational Psychologists

As medical treatment improves and inclusive practices and policy changes support children with medical conditions to attend mainstream school, recognising and understanding the important role of EPs in supporting children with complex medical needs is essential (Anderson, 2021; Brown and DuPaul, 1999). Given that EPs have historically had minimal involvement with children with rare conditions in mainstream schools, this is an area that warrants considerable additional research and focus.

Promoting collaboration between different stakeholders is one important aspect of an EP's role. The mesosystem consists of inter-relations among several microsystems in which the developing person is situated (Bronfenbrenner & Morris, 2006). According to the bioecological theory, children's development is optimal when relationships between contexts (i.e. the mesosystem) are strengthened (Sheridan et al., 2017). The current study identified collaboration between parents, teachers, and

other professionals (i.e. allied health professionals including psychologists, medical staff) supporting children with cystinosis as a strong facilitator in their school experiences. In terms of the home-school interrelations, participants highlighted positive, trusting relationships characterised by clear and open communication. Despite this, some teachers noted concerns about broaching “difficult conversations” with parents, while parents similarly noted the stress and challenges associated with advocating for their children’s needs. The importance of collaborative holistic care for children with rare conditions such as cystinosis is well documented (e.g. Belzer et al., 2022; Ariecta et al., 2015; Kole et al., 2021; Verger et al. 2020). National policies and frameworks for practice have highlighted the need for communication between healthcare and education sectors for people with rare disease (DoH, 2014). Despite this, participants in the current study noted that there was no direct link between healthcare teams and schools. While the children were generally doing well, multi-disciplinary supports in relation to particular stages (e.g. transitions) is recommended. Establishing a clear communication protocol between services would be of benefit.

EPs have both specialist knowledge of education and the school system, combined with a comprehensive understanding of child development. As such, they are well placed to promote and strengthen collaboration between different stakeholders at the mesosystem level (i.e. parents, school, and health and psychology services) across different developmental periods (Barraclough & Machek, 2010; Wodrich, 2004).

NEPS promotes the use of a consultation model to encourage change (Department of Education and Skills & National Educational Psychological Service, 2010). The consultation model recognises the unique expertise of people in the child’s microsystem and emphasises the value of working together to find solutions (Wagner, 2000; Sheridan et al., 2017). Sheridan et al. (2017) state that consultation builds on strengths in the mesosystems to allow for individual planning that is unique to student’s needs. This model may be of particular relevance in the rare disease community where access to information is scarce. Through consultation, EPs bring stakeholders together to find solutions, building on each team member's expertise

and strength thus facilitating a more holistic approach to addressing children's wellbeing.

Emphasising Time: Responding to Needs Over Time

Support for Educational Transitions

In terms of time, educational transitions (i.e. from preschool-primary/primary to post-primary) were identified as a critical period where families, children, and schools required additional support. Parents noted a lack of clear information regarding the transition process, and reported heightened stress and worry about this period. Participants also noted schools' apprehension when first learning of a children's medical needs. For participants in this study, there was no specific transition protocol in place to support children with a rare illness through educational transitions. The onus was on parents to fill this gap by providing training, developing information packs, advocating for their child's needs, and researching supports available for their child. The transition from paediatric to adult healthcare services has received significant attention with an emphasis on the importance of co-ordinated multidisciplinary care and increasing autotomy of adolescents (e.g. Raina et al., 2017; Cystinosis Research Network, 2022), However, there is no information and advice that focuses on educational transitions for children with rare diseases in the Irish context. The National Council for Special Education (NCSE, 2016; 2023) has developed guidelines for supporting transitions for children with special educational needs. While such resources may be useful as a general tool, the results of the study suggest the need for an educational transition guide designed for children with cystinosis and/or rare disease. In addition to providing information on medical needs, the findings of this study (e.g. the importance of individualised understanding of needs, monitoring needs over time, the challenges of adolescence and the importance of collaboration) inform the essential components of such a resource. Given their knowledge of child development and the educational system, EPs are ideally placed to inform the development of this guide. As participants highlighted the importance of child-centred care, it would also be pivotal to involve children with cystinosis and their families in the development of a transition resource. Additionally, EPs would be well placed to provide a link between primary and post-primary

schools. Barraclough & Macheck (2010) highlight that psychologists also play an integral role in determining the most appropriate educational settings for children with medical needs, identifying required resources, and informing families and children of their educational rights.

Changing Health Needs and Absenteeism

The majority of children in the current study did not experience significant school absenteeism. However, it was noted that this could change over time based on health needs. For children who experienced absenteeism in post primary school, participants felt that absenteeism impacted their relationships with peers and academic progress. Given that this experience was only evident for one child in the current cohort, more research is necessary. However, it highlights the importance of maintaining peer relationships and connectedness to school during periods of ill health. Previous research has suggested that maintaining regular contact with peers during periods of absenteeism may facilitate smoother transitions back to school (Verger et al., 2020). The use of information technology to maintain a sense of connection to school is one way in which this could be facilitated (Verger et al., 2020). A novel approach presented by Powell et al. (2021) indicated that the use of telepresence robots improved adolescents' sense of connectedness with peers and school during periods of absenteeism for adolescents with cancer. While innovative, the researchers acknowledged that such technology may not be an option for all schools. Simpler approaches such as sending videos, writing cards, or utilising virtual classrooms could be used to maintain a sense of connection. For older children, classmates could play a role in passing on homework (if necessary) through text messages, video calls etc. Previous research has suggested that positive school reintegration experiences can be enhanced through structured communication pathways between home, school and healthcare providers (Lum et. al., 2017). This is particularly important given the lack of school-healthcare collaboration previously noted in this study. Defining a link person to communicate between health and education services, particularly in post primary school may be of particular benefit to promote collaborations and reduce parental stress (Verger et al., 2020).

Adolescence

Approaching adolescence was identified as a time of potential psychosocial challenge for children in this study. Previous research has identified the adolescent period as a time of increased need for children with rare disease (Somanadhan et al., 2023). Parents in this study reported concerns about the changing social demands of adolescence, being treated differently, and bullying. It was important for children to feel the same as their peers and some children concealed aspects of their medical condition (e.g. G-tube). From a whole school perspective, this underscores the need to promote general wellbeing guidelines in school, and develop a culture of inclusivity and respect for diversity as recommended by the NICE social, emotional and mental health guidelines (NICE, 2022) and the Department of Education's wellbeing policy statement and framework for practice (DES, 2019). Through consultation, EPs can play a role in helping teachers to identify and monitor risks and co-ordinate responses across the continuum of support as needed. Where children present with social, emotional, or behavioural needs, EPs can provide support for children and their families through assessment and evidence-based intervention (Anderson et al., 2003).

Knowledge in the Exosystem

At a exosystemic level, participants in this study highlighted the need for increasing teacher awareness regarding the psychological, social, and academic impact of cystinosis for children and young people in schools. This is in line with The National Rare Disease Plan for Ireland 2014-2018 which highlights that there should be increased awareness of rare diseases and the potential impact on learning and emotional wellbeing within the context of initial teacher education and continuing professional development (DoH, 2014). The findings from this study could inform training and professional development as it highlights the interrelated, dynamic factors that are involved in psychosocial wellbeing for children and young people with cystinosis. In addition to focusing on what children's medical needs are, it is crucial that schools know how these impact the child, when times of difficulty may arise, and what is needed to facilitate psychosocial wellbeing. The results emphasise the importance of communication at a child, parent, and multi-disciplinary level and crucially highlights that children's psychosocial wellbeing should be monitored over

time. Awareness of the knowledge and implications of rare conditions has been identified as integral to the inclusion of children with rare conditions in education in previous research (Verger et al., 2020). Given EPs' knowledge of such broad factors, they are well placed to co-create training alongside parents, teachers, children with cystinosis and medical professionals. Anderson et al. (2003) suggest that psychologists are particularly well placed to engage with children in developmentally appropriate ways regarding understanding their medical conditions. Teachers and parents also highlighted the need for awareness at the individual level, further highlighting the importance of person centred planning and understanding individual needs as described previously.

For participants in this study, views on increasing peer awareness of cystinosis were varied. While some felt that this would be beneficial, others were reluctant, but all stressed the importance of respecting the child's wishes. The importance of including the child in decision making was highlighted by parents and teachers. In terms of professional practice, this finding underscores the need to provide sensitive, child-centred support, and including children with cystinosis in decisions about their care and education. Although children may not wish for their peers to be given specific knowledge about cystinosis, continued promotion of whole school approaches to wellbeing, diversity, and inclusion may be a practical way to promote psychosocial wellbeing and positive school experiences.

The Role of the Special Needs Assistant in the Exosystem: Beyond Care Needs

Special Needs Assistants are integral to supporting students who have significant care needs arising from a disability or a medical condition. In Ireland, the SNA role is characterised by a non-teaching care remit to facilitate attendance and inclusion of pupils with additional care needs in school. Examples of primary care needs include assistance with feeding, administration of medication, assistance with toileting, assistance with mobility, non-nursing care needs, and care needs requiring frequent interventions/withdrawals from the classroom (DES, 2014). The children in this cohort required support in many of these areas and all had access to SNAs/classroom assistants. However, it was also clear from the responses from participants that the role of SNAs for children with cystinosis goes beyond primary

care needs. In the current cohort, participants noted that SNAs were integral to facilitating home-school communication and often acted as a primary point of contact for parents. Their positive relationships with children was emphasised and some participants noted children may be more comfortable discussing challenges/concerns with them than with other school staff. SNAs, particularly those who support children over a number of years, would be well positioned to monitor signs of psychosocial vulnerability. Professional development for SNAs regarding how to recognise these signs along and a protocol for responding to concerns may be beneficial.

For some children, parents and teachers noted that access to an SNA/classroom assistant could at times be a barrier to social integration, opportunities for social interaction, and independence. This finding is in line with previous research examining SNA support in Irish schools. Zhao et al. (2021) highlighted that while parents, pupils, SNAs and teachers view the role of the SNA as essential, pupils in particular can feel that the presence of an SNA inhibits social interactions with their peers and result in them feeling different to their classmates Circular 0030/2014 (DES, 2014) highlights the importance of including the viewpoints and preferences of the child regarding the level and extent of SNA support they receive. However, research by Griffin (2018) illustrated that, in practice, pupil voice is lacking in relation to the SNA scheme. As highlighted earlier, eliciting the voice of the child in relation to their care needs and preferences is crucial and their views on SNA support should be included. Given that SNA support is essential for many children with rare diseases, future research could also focus on how the views of children with rare diseases and complex medical needs are integrated into the SNA scheme.

Previous research has shown SNAs frequently engage in academic, social, emotional and behavioural support outside of their prescribed non-teaching role (Griffin & Blatchford, 2021). A report by the NCSE (2018) noted concerns that teachers can become over-reliant on SNAs as the “expert” on children with care needs. While this was not specifically examined in the current study, participants referenced reliance on SNA to transfer information from one class teacher to the next, to communicate with parents and to be aware of child’s medical needs could suggest that teachers take a “back seat” with regards children’s broad school experiences.

While SNAs play an essential role in the meaningful inclusion of children with rare diseases it is also important that teachers remain fully informed of children's care needs and have a primary role in planning and coordinating support alongside parents, children, and SNAs.

Conclusion

The current research aimed to explore the psychosocial strengths and needs of children with cystinosis and identify, from the perspective of parents and school staff, the factors influencing their school experiences. This research has contributed to the evidence and knowledge base regarding school experiences of children with cystinosis and other rare diseases. To the researcher's knowledge, this is the first qualitative study that explores the experiences of children with cystinosis in the school system. The findings from both the systematic literature review and the empirical research having significant implications for educational psychology practice, schools, and healthcare teams. The findings highlight that although children with cystinosis have many psychosocial strengths, it is important for EPs, schools, and healthcare teams to be mindful of the challenges and barriers they experience over time. The findings highlight the importance of collaborative, person-centered practice that is coordinated across health and education sectors and EPs can play a central role in bridging this gap. The findings also highlight the need for increased professional development for teachers (and other key stakeholders in the education system) on the impact of living with cystinosis. It is hoped that highlighting these implications of the current research will promote positive change that facilitates positive school experiences for children with cystinosis and other rare diseases.

"My wish is only that he does get through school the same as he is, the same little happy man".

Parent of primary school child.

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Appendices

Appendix A: Critical Appraisal Skills Programme Checklist

CASP Review Questions 1- 6

Author	Q.1 Did the study address a clearly focused issue?	Q.2 Did the authors use an appropriate method to answer their question?	Q.3 Were the cases recruited in an acceptable way?	Q.4 Were controls selected in an acceptable way?	Q.5 Was the exposure accurately measured to minimise bias?	Q.6 (a) Were the groups treated equally	Q.6 (b) Have the authors taken account of potential confounding factors in the design/analysis
Aly et al., 2014	Yes	Yes	Yes	Yes	Yes	Yes	No
Ariceta et al., 2015b	Yes	Yes	Yes	N/A	Not sure	N/A	No
Atia et al., 2022	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Ballantyne et al., 2013	Yes	Yes	Yes	Yes	Yes	Yes	No
Delgado et al., 2005	Yes	Yes	Yes	Unsure	Yes	Yes	Yes
Spilkin & Ballantyne, 2007	Yes	Yes	Yes	N/A	Yes	N/A	Yes

Author	Q.1 Did the study address a clearly focused issue?	Q.2 Did the authors use an appropriate method to answer their question?	Q.3 Were the cases recruited in an acceptable way?	Q.4 Were controls selected in an acceptable way?	Q.5 Was the exposure accurately measured to minimise bias?	Q.6 (a) Were the groups treated equally	Q.6 (b) Have the authors taken account of potential confounding factors in the design/analysis
Ulmer et al., 2009	Yes	Yes	Yes	N/A	YES	N/A	Yes
Wolff et al., 1988	Yes	Yes	Yes	N/A	Not sure	N/A	Yes

CASP Review Questions 7 - 11

Author	**Q.7** **What are the results of the study**	**Q.8** **How precise are the results**	**Q9.** **Do you believe the results?**	**Q.10** **Can the results be applied to the local population?**	**Q.11** **Do the results fit with other available evidence**	**Score**
Aly et al., 2014	Children with cystinosis experienced significantly more behavioural problems (withdrawn, anxious, social problems) than the control group of children without a medical condition	Statistically significant differences between groups on total behaviour scores.	Yes	Yes	Yes	9/10 90%
Ariceta et al., 2015b	Cystinosis was reported to have a negative impact children's school attendance, (29%), "learning ability" (5%) and social life (10%).	Descriptive statistics used.	Yes	Yes	Yes .	6.5/8 81%

	10% of participants noted that cystinosis results in children “feeling different”					
Atia et al., 2022	Results of CBCL for children with cystinosis found that: ○ 85% of children had internalisation symptoms ○ 75% had somatic complaints ○ 70% had withdrawn behaviour ○ 50% were anxious or depressed ○ 95% of children had a total mean score in the significant range There was no significant difference between children with cystinosis and children with chronic kidney disease on any of the scales.	Statistical analysis revealed no difference between groups.	Yes	Yes	Not sure	9.5/10 95%
Ballantyne et al., 2013	The cystinosis and control groups did not differ significantly in terms of behavioural regulation as assessed by the BRIEF. Significant difference	Statistical analysis	Yes	Yes	Not sure	9/10 90%

	reported in terms of metacognitive skills.					
Delgado et al., 2005	• There was a significant difference between the cystinosis group and control group on the following scales: Total Problems Summary Scale, Internalising Problems Summary Scale, Social Problems, Somatic Complaints, Attention Problems, Thought Problems and Aggression. Comparisons between the cystinosis group and CF group revealed that only the social problems scale significantly differed between the two chronic disease groups.	Statistically significant differences reported	Yes	Yes	Yes	9.5/10 95%
Spilkin & Ballantyne, 2007	• Parents reported numerous family stresses as result of caring for a child with cystinosis • 36% of parents believe their child with cystinosis displays behaviour that is more intense than other children • 65% of parents report their child with cystinosis experiences	Descriptive data used.	Yes	Yes	Yes	8/8 100%

	occasional mood-swings and outbursts of temper • More than 50% of parents indicated that their child often or sometimes get their feelings hurt, do not enjoy life, and are not self-reliant • 30% of parents report their child often experienced academic difficulties 80% of children rarely or never see a therapist for support with social or emotional issues					
Ulmer et al., 2009	• Self-reported QOL was normal on all dimensions apart from positive emotions which was significantly impaired. Parents reported significantly lower positive emotions, autonomy, social and cognitive functions for children with cystinosis	Descriptive statistics used	Yes	Yes	Yes	8/8 100%
Wolff et al., 1982	• Parents indicated that the children were socially accepted by their peers	N/A Qualitative Description	Yes	No Cystinosis treatment is different	No	4.5/8 56%

	• Researchers noted signs of potential psychological difficulties however it was unclear how signs were measured or conceptualised Most children with cystinosis were reported to be functioning well academically			today and children likely have different experiences		

Appendix B: Parent Information Letters and Consent Forms

Note: The information provided in this letter outlines the entire research project being conducted by UCD and Cystinosis Ireland. The research presented in this thesis forms part of this project.

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Parent/Guardian Participant Information Letter
Cognitive Profiles and Psychosocial Experiences of Young People Living with Cystinosis

Dear Parent(s),

My name is Joyce Senior, and I am an educational psychologist and researcher in the School of Education, University College Dublin. I am inviting you to participate in the research that I am undertaking in relation to *Cognitive Profiles and Psychosocial Experiences of Young People Living with Cystinosis*. I am undertaking this research alongside Elaine Mellotte and Agnes Fitzgerald, who are trainee educational psychologists. This letter provides further information on the study and what participation would involve for you. I invite you to read the following information and then decide whether or not to take part.

What is this research about?

The purpose of the proposed research is to identify the unique educational, cognitive (reasoning, problem solving and remembering), emotional, social, and wellbeing challenges, barriers and facilitators faced by young people with cystinosis.

Why am I doing this research?

To date, the vast majority of research in cystinosis has focused on medical aspects of the illness. While children with a diagnosis of cystinosis face many of the same challenges as others in navigating educational and social situations, these challenges may worsen when managing a rare condition. Despite this, little is known about the experiences of children living with this diagnosis, particularly in relation to school.

The limited research data that is available suggests that children with cystinosis have significant deficits in visual processing (how the brain understands what is seen) and visual spatial skills (problem solving using visual information). These challenges can lead to learning difficulties particularly in Mathematics, spellings and Geography.

A further aim of this research is therefore to conduct individual cognitive/psychoeducational assessments with children and young people with cystinosis in order to provide more detail with regard to potential patterns of specific strengths and needs associated with cystinosis. This is essential information so that appropriate supports, recommendations and accommodations can be identified and put in place.

Why have you been invited to take part?

As the parent of a child with cystinosis, your voice is essential to the research. I am contacting all parents of school-aged children with cystinosis in Ireland. Should you wish to take part, this invitation will be extended to your child's teaching and support staff (e.g., SNA if applicable), with your consent.

How will my data be used?

Data generated in the interviews will be analysed to identify themes in relation to the educational and psychosocial experiences of children with cystinosis. It is important that these additional challenges are identified in order to provide the appropriate support and opportunities for young persons with cystinosis and their families. Overall findings and conclusions will be shared with Cystinosis Ireland and will form part of the doctoral theses of the trainee educational psychologists involved in the project.

They may also be published in academic journals and presented at conferences. Personal, identifiable information will not be shared or published. De-identified data (data gathered over the course of the research, with all personal information [names, date of birth, school name] removed) may be archived for future studies by researchers as part of the Doctorate in Educational Psychology. You can indicate on the consent form if you are happy for your data to be used in future studies.

What does participation in this research involve?

Study 1: Parental and Teacher Interview

Participation will involve an interview with you and your child's teacher and support staff (e.g. Special Needs Assistant, if applicable). This will focus on what it is like for your child to have a diagnosis of cystinosis, what barriers they experience, and what is going well and is helpful, particularly in relation to school. The interview should take approximately 60 minutes and will be organised at a place and time that is convenient to you. Interviews can take place in person in a location of choice (e.g., a café, UCD,), via telephone, or via a secure video platform such as Zoom. With your consent, your interview will be audio recorded and audio files will be sent to an external service for transcription.

Study 2: Psychoeducational Assessment

Should you wish to avail of this, a second aspect of the research is a psychoeducational assessment of your child's cognitive ability (ability to learn, problem solve, remember), attainment in literacy and numeracy, independent living skills, and social and emotional wellbeing. This will allow us to gain insight into specific strengths and needs. This will involve an individual assessment with your child in your home and it would take approximately three hours including breaks. It can be spread over a number of days if your child would find it difficult to complete in one day. Following this assessment, your child will be asked some questions which will focus on their experience of school, and their experience of cystinosis in the school setting. If there are teacher and parent reported needs/difficulties, you and your child's teacher may be asked to complete an additional screener on behaviour or

wellbeing (e.g. the BASC, Achenbach or Conners', etc) and/or adaptive and everyday living skills (Vineland or ABAS). Following the assessment, you will be provided with a psychoeducational report which will contain detailed recommendations for you and your child's school.

How will my privacy be protected?

All information which is collected during the course of the research will be kept strictly confidential, unless limitations of confidentiality are breached. This includes concerns of potential or actual harm to you or another person. This will be discussed with you prior to beginning the interview.

On completion of the interview, the interview will be transcribed (converted to written format) via an external transcription service and any identifying information (such as names, date of birth, name of schools) will be removed. Following this the recorded interview will be deleted. I will assign a code to the records and any use of the data will be based on the code number. Only members of the research team will have access to these files.

All data, including that from the psychoeducational assessment if you wish to avail of that aspect of the research, will be stored securely, with any identifiable details (such as names, date of birth, names of schools) removed. All data will be stored in a folder on a password-protected, encrypted computer for the duration of the study. Once the data has been analysed, it will be archived in a password-protected folder on the UCD server. No individual participant will be mentioned in any report or publication of the findings.

Are there any benefits in taking part in the study?

It is hoped that the study will generate a greater understanding and awareness of the experiences of children and young people with cystinosis and their families. It is anticipated that the findings will inform initial and continuing professional training of

teachers and healthcare professionals such as educational psychologists as well as national and international policy and practice.

If a psychoeducational assessment is completed with your child, you will receive a detailed report outlining the results of the assessment. This report will identify and describe your child's strengths and needs, and will recommend accommodations and strategies for use at school and home to support your child academically, socially and emotionally.

Are there any risks involved in participating?

The interview will focus on day to day experiences of children with cystinosis. Some topics may be sensitive and have the potential to cause distress for some participants. If you become upset during the interview, it can be paused or postponed. There is no obligation to continue with the interview and you may withdraw your participation from the study if you wish. When the interview is finished, participants will have the opportunity to reflect on the interview, share their experience of it and ask any questions they may have. You will be provided with information on support services should you require further support.

During the interview, it is possible that you or your child's teacher will share information which may suggest other difficulties or conditions, for example, specific learning difficulties such as dyslexia or mental health difficulties. If you choose to avail of the psychoeducational assessment, the results of this may also show that your child has particular needs or additional conditions and you will be guided to apply for supports.

Can I change my mind at any stage and withdraw from the study?

Yes, you can withdraw your participation at any point before or during the interview or the psychoeducational assessment. If you participate in the interview or the assessment and subsequently wish to withdraw, you may do so for up to two weeks

following the interview and/or assessment. Non-participation or withdrawal from the study will have no impact on the support you receive from Cystinosis Ireland.

What do I do if I wish to take part?

If you would like to take part in the study, please complete the attached consent form and return it to me via the email address below. We would also like to interview your child's class teacher/year head (if in post primary) and special education teacher and SNA (if applicable). Should you wish your child's school to be involved, with your consent we will contact your child's school with information about the study and invite them to participate.

What do I do if I would like to avail of the psychoeducational assessment but I do not wish to take part in the study?

You can choose to avail of the psychoeducational assessment without taking part in the study. You will receive the same professional assessment and report but the data will not be included in the study. There is also no obligation to take part in the parent or school interview if you avail of the psychoeducational assessment.

How will your data be used?

Collected data will be used to inform research and policy and practice. Data will also be archived for research purposes, and may be published in academic journals, used to produce research reports, and presented at conferences.

How will I find out what happens with this project?

A summary of the research findings will be provided following completion of the study. Results of the study will also be shared with Cystinosis Ireland and presented at conferences. Should you wish to receive a copy of the final doctorate research document you may contact me directly. The results will also be used towards the publication of a journal article in an academic journal.

Should you wish to avail of the psychoeducational assessment, you will be provided with a full report.

Contact details for further information.

If you have any further questions, please do not hesitate to contact me at the details provided below:

Email: Joyce.senior@ucd.ie
Telephone: 087 784 0308.

Yours Sincerely,

_____

Joyce Senior, Educational Psychologist

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Consent Form for Parents/Guardians

Cognitive Profiles and Psychosocial Experiences of Young People Living with Cystinosis

Researchers: Joyce Senior, educational psychologist, Agnes Fitzgerald and Elaine Mellotte, trainee educational psychologist.

- I have read and understand the information letter provided for the above study.
- I have had the opportunity to ask questions about the study and received satisfactory answers to any questions posed.
- I understand that my participation in this research is entirely voluntary (my choice) and that I am free to withdraw my consent without explanation at any time before, during, or up to two weeks following my interview and the psychoeducational assessment (if I wish to avail of this).
- I agree for my interview to be audio recorded and sent via a secure link to an external transcription service to be transcribed.
- I understand that my interview data (and the psychoeducational assessment materials) will be stored with an ID number on a password-protected encrypted computer.
- I understand that identifiable details such as the real names of me, my child and the school they attend will not be included in the study.
- I agree that the data collected can be used in publications such as journal articles or presentations at conferences and in doctoral theses.

Please indicate your agreement on each of the statements below:

- I understand the above statements and I agree to take part in this study and an interview YES NO
- I agree for my child to avail of the full psychoeducational assessment YES NO
- I agree for my child's school/teachers to be contacted to take part in this study YES NO
- I agree for my de-identified data to be archived for use in future studies for researchers on the Doctorate in Educational Psychology in UCD YES NO
- I agree that the data collected can be used in a thesis for the degree of Doctorate in Educational Psychology and in publications such as journal articles or presentations at conferences YES NO

Name of Child (in block capitals):

_____

Name of Parent/Guardian (in block capitals):

_____

Relationship to child (please tick)

Mother		Father		Legal Guardian	

Signed: _____

Date: _____

Appendix C: Child Information Letter and Assent Form (Primary School)

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Hello,

Our names are Elaine, Agnes, and Joyce. We are doing some research to learn about what it is like to have Cystinosis.

To do this we would like to talk to you, to your parents, and to your teachers. We would also like to meet with you and do some activities that will help us find out about how you learn, what you are good at, and any areas that you might need more help with. This will involve doing some activities like puzzles, reading, writing, and maths.

We will record and write down things that are important to you, things that are hard, and things that would be helpful for you, especially in school.

We will write a research article about what we learn, but we won't use your name or the name of your parents, teachers, or school.

We hope that we can use what we learn to help people to understand what it is like to have cystinosis, and how teachers and parents can help kids with Cystinosis.

Are you happy for us to talk to your parents?

Are you happy for us to talk to your teachers?

Are you happy for us to talk to you?

Are you happy to complete oral and written activities with us in your home?

Are you happy for us to write down all your answers so that we can write a report?

I understand that it is my choice if I take part. If I do agree to take part, I can change my mind and ask that my answers are not included in the study for up to two weeks afterwards.

My Name: _____

Date: _____

Appendix D: Child Information Letter and Assent Form (Post-Primary School)

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Hello,

Our names are Elaine, Agnes, and Joyce. We are doing some research to learn about what it is like to have Cystinosis.

To do this we would like to talk to you, to your parents, and to your teachers. We would also like to meet with you and do some activities that will help us find out about how you learn, what you are good at, and any areas that you might need more help with. This will involve doing some activities like puzzles, reading, writing, and maths.

We will record and write down things that are important to you, things that are hard, and things that would be helpful for you, especially in school.

We will write a research article about what we learn, but we won't use your name or the name of your parents, teachers, or school. We hope that we can use what we learn to help people to understand what it is like to have cystinosis, and how teachers and parents can help kids with cystinosis.

Are you happy for us to talk to your parents? YES ____ NO ____

Are you happy for us to talk to your teachers? YES ____ NO ____

Are you happy for us to talk to you? YES ____ NO ____

Are you happy to complete oral and written activities with us in your home? YES ____ NO ____

Are you happy for us to write down all your answers so that we can write a report? YES ____ NO ____

I understand that it is my choice if I take part.
If I agree to take part, I can change my mind and ask that my answers are not included in the study for up to two weeks afterwards. YES ____ NO ____

My Name: _____

Date: _____

Appendix E: School Information Letter and Consent Form

Note: The information provided in this letter outlines the entire research project being conducted by UCD and Cystinosis Ireland. The research presented in this thesis forms part of this project.

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Participant Information Letter for Teacher/SNA
Cognitive Profiles and Psychosocial Experiences of Young People Living with Cystinosis

My name is Joyce Senior and I am an educational psychologist and researcher in the School of Education, University College Dublin. I am inviting you to participate in the research that I am undertaking in relation to *Cognitive Profiles and Psychosocial Experiences of Young People Living with Cystinosis*. I am undertaking this research alongside Elaine Mellotte and Agnes Fitzgerald, who are trainee educational psychologists. This letter provides further information on the study and what participation would involve for you. I invite you to read the following information and then decide whether or not to take part.

What is this research about?

The purpose of the proposed research is to identify the unique educational, cognitive (reasoning, problem solving and remembering), emotional, social and wellbeing challenges, barriers and facilitators faced by young people with cystinosis.

Why am I doing this research?

To date, the majority of research into cystinosis has focused on medical aspects of the illness. While children with a diagnosis of cystinosis face many of the same challenges as others in navigating educational and social situations, these challenges may worsen when managing a rare condition. Despite this, little is known about the experiences of children living with this diagnosis, particularly in relation to their engagement and participation in school.

The limited research data that is available suggests that children with cystinosis have significant deficits in visual processing (how the brain understands what is seen) and visual spatial skills (problem solving using visual information). These challenges can lead to learning difficulties particularly in Mathematics, spellings and Geography.

A further aim of this research is to conduct individual cognitive/psychoeducational assessments with children and young people with cystinosis in order to provide more detail with regard to potential patterns of specific strengths and needs associated with cystinosis. This is essential information so that

Why have you been invited to take part?

As educational staff supporting a child with cystinosis your voice is essential to the research. With the permission of the student's parents, I am inviting class teachers, year heads, special education teachers and special needs assistants of school-aged children with cystinosis in Ireland to participate in this study. It is your decision whether or not to participate.

How will my data be used?

Data generated in the interviews will be analysed to identify themes in relation to the psychosocial experiences of children with cystinosis. It will demonstrate what challenges are faced by children with cystinosis as well as what is/would be helpful to

further support them. Overall findings and conclusions will be shared with Cystinosis Ireland and will form part of the doctoral theses of the trainee educational psychologists involved in the project. Personal, identifiable information will not be shared or published. De-identified data (data gathered over the course of the research, with all personal information [names, date of birth, school name] removed, so participants cannot be identified) may be archived for future studies by researchers as part of the Doctorate in Educational Psychology. You can indicate on the consent form if you are happy for your data to be used in future studies.

What does participation in this research involve?

Participation involves completion of an interview. This interview will collect information about the student and their experiences in school in the context of their diagnosis. The interview will focus on how the student's condition impacts on their school experience including what barriers they experience as well as what is going well and is helpful.

The interview should take approximately 60 minutes. Interviews will be organised at a place and time that is convenient to you. Interviews can take place in person, via telephone, or via a secure video platform such as Zoom. With your consent, your interview will be audio recorded for transcription purposes.

If there are teacher and parent reported needs/difficulties, you and the child's parent may be asked to complete an additional screener in relation to wellbeing or behaviour (e.g., the BASC, Achenbach or Conners', etc) and/or everyday living skills (Vineland or ABAS).

How will my privacy be protected?

All information which is collected during the course of the research will be kept strictly confidential, unless limitations of confidentiality are breached. This includes concerns of potential or actual harm to you or another person. This will be discussed with you prior to beginning the interview.

On completion of the interview, the interview will be transcribed (converted to written format) via an external transcription service and any identifying information (such as names, date of birth, name of schools) will be removed. Following this the recorded interview will be deleted. A code will be assigned to the records and any use of the data will be based on the code number.

All data will be stored securely, with any identifiable details (such as names, date of birth, names of schools) removed. All data will be stored in a folder on a password-protected, encrypted computer for the duration of the study. Once the data has been analysed, it will be archived in a password-protected folder on the UCD server. No individual participant will be mentioned in any report or publication on the findings.

Are there any benefits in taking part in the study?

It is hoped that the study will generate a greater understanding and awareness of the experiences of children and young people with cystinosis. It is anticipated that the findings will inform initial and continuing professional training of teachers and healthcare professionals as well as national and international policy and practice.

Are there any risks involved in participating?

The interview will focus on the experiences of children with cystinosis. Some topics may be sensitive and have the potential to cause distress for some participants. If you become upset during the interview, it can be paused or postponed. There is no obligation to continue with the interview and you can withdraw your consent to participate if you wish. When the interview is finished, participants will have the opportunity to reflect on the interview, share their experience of it and ask any questions they may have. You will be provided with information on support services should you require further support.

Can I change my mind at any stage and withdraw from the study?

Yes, you can withdraw your participation at any point before or during the interview. If you participate in the interview and subsequently wish to withdraw, you may do so for up to two weeks following the interview.

What do I do if I wish to take part?

If you would like to take part in the study, please complete the attached consent form and return it to me via email. If you do not wish to take part, no further action is required.

How will your data be used?

Collected data will be used to inform the writing of research papers and policy documents. Data will also be archived for research purposes, and may be published in academic journals, used to produce research reports, and presented at conferences.

How will I find out what happens with this project?

A summary of the research findings will be provided following completion of the study. Results of the study will also be shared with Cystinosis Ireland and presented at conferences. Should you wish to receive a copy of the final doctorate document you may contact me directly. The results will also be used towards the publication of a journal article in an academic journal.

Contact details for further information.

If you have any further questions, please do not hesitate to contact me at the details provided below.

Email: Joyce.senior@ucd.ie
Telephone: 087 784 0308

Yours Sincerely,

_____

Joyce Senior, Educational Psychologist

UCD School of Education

Roebuck Offices Room 0.06
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Belfield, Dublin 4, Ireland

T +353 1 716 7967
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Áras Roebuck
An Coláiste Ollscoile Baile Átha Cliath,
Belfield, BAC 4, Eire

sarah.walshe@ucd.ie

Consent Form for Teachers/SNAs

Cognitive and Psychosocial Profiles of Young People Living with Cystinosis

Researchers: Joyce Senior, educational psychologist, Agnes Fitzgerald and Elaine Mellotte, trainee educational psychologist.

- I have read and understand the information letter provided for the above study.
- I have had the opportunity to ask questions about the study and received satisfactory answers to any questions posed.
- I understand that my participation in this research is entirely voluntary (my choice) and that I am free to withdraw my consent without explanation at any time before, during, or up to two weeks following my interview.
- I agree for my interview to be audio recorded and sent to an external transcription service to be transcribed
- I understand that my interview data will be stored with an ID number on a password-protected encrypted computer.
- I understand that identifiable details such as the real names of me, my student and the school they attend will not be included in the study.
- I agree that the data collected can be used in publications such as journal articles or presentations at conferences and in doctoral theses.

Please indicate your agreement on each of the statements below:

- I understand the above statements and I agree to take part in this study and an interview YES NO
- I agree for my de-identified data to be archived for use in future studies for researchers on the Doctorate in Educational Psychology in UCD YES NO
- I agree that the data collected can be used in a thesis for the degree of Doctorate in Educational Psychology and in publications such as journal articles or presentations at conferences YES NO

Name of Student (in block capitals): _____

Name of Staff Member (in block capitals): _____

Relationship to child (please tick)

Class Teacher		Year Head (Post Primary)		Special Education Teacher	
Special Needs Assistant		Other (Please Specify			

Signed: _____

Date: _____

Appendix F: Parent Interview Schedule

UCD School of Education
Roebuck Offices Room 0.06
University College Dublin,
Belfield, Dublin 4, Ireland

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F +353 1 716 1143

Scoil an Oideachais
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An Coláiste Ollscoile Baile Átha Cliath,
Belfield, BAC 4, Eire

sarah.walshe@ucd.ie

Parent Interview Schedule

Introduce self and aims of the research project.

My name is Elaine Mellotte, I am a trainee educational psychologist from University College Dublin.

The aim of the project is to explore the unique strengths and needs of children with cystinosis and identify barriers and facilitators faced in the context of the education system. This will be ascertained through interviews with parents, teachers and special needs assistants (if applicable) of children with cystinosis.

I am undertaking this research under the supervision of Dr. Joyce Senior, an educational psychologist and Director of the Professional Doctorate in Educational Psychology Training Programme in UCD.

Explain limits of confidentiality.

All information collected during the course of the research will be kept strictly confidential, unless limitations of confidentiality are breached. As a mandated person there are circumstances where I may be obliged to break confidentiality. This may occur in two ways:

1. If a strong belief exists that there is a serious risk of harm or danger to your child or another individual.
2. Occasions when disclosure is required as part of a legal process or Garda investigation.

In such instances information may be disclosed to significant others or appropriate third parties without permission being sought. Where possible a full explanation will be given to you regarding the necessary procedures and intended actions that may need to be taken.

Opportunities for questions and provision of consent

Ask participant/s if they have any questions about the research or data collection and answer any questions.

Check that they consent to the interview being audio recorded.

Check that they consent to their data being transcribed (converted into written format) by a third party company.

Ask if they are happy to proceed. If yes, inform them that you will begin recording.

Section A: Interests and Strengths

- Tell me a little about your child. What does he/she like to do to have fun?
- What are his/her strengths (e.g.. kind, creative, funny, sporty, etc)?

Section B: School Experiences

Attendance and Engagement in School

- What, if any, impact does your child's condition have on their ability to attend school?
    - *If yes - How do you think this impacts him/her? E.g. Falling behind in schoolwork/stress in catching up/social opportunities/hospital education services*
    - *How many days? Access to home tuition? Summer provision?*
- Has Cystinosis impacted on his/her ability to participate in activities at school? (academic/physical/social/extracurricular)

- Does your child like to go to school?
    - *What do you think helps here/ What makes this difficult?*
- Have there been any involvement in school from outside agencies e.g. NEPS in relation to your child's needs?
- Have they had any challenges with transitions e.g. starting school/changing teachers

Academic Attainment

- What subjects does your child enjoy most?
- Are there any they find more challenging?
- Do you have any concerns about your child's learning?
- Does your child receive any additional teaching support (e.g. special education teaching/learning support)?
- Do they have a Student Support Plan/IEP?
    - *If Yes, were you and your child involved in setting the targets?*
- Do you have formal review meetings for parents and school staff e.g. care planning meetings?
    - If yes, who is involved in these?
- Do you have any concerns regarding
    - ability to organise themselves or manage the school routine?
    - follow instruction?
    - concentration/attention to tasks?
    - independence?

- What is homework like for him/her? Is it manageable? Is it adapted to their needs both academically and physically (e.g. tiredness, feeling sick etc).

Section C: Social Experiences in School

- What are their relationships with peers in school like? (e.g. strong network of friends/are they excluded/treated the same by peers and parents, concerns regarding teasing/bullying/being different)

    - *Does cystinosis impact on his/her ability to make friends? If yes, how so?*
    - *What helps/would help your child to engage socially in school*
- Do your child's peers know about and understand their condition?
    - *If peers have been informed of cystinosis, how was this done and by who? If not, why?*
    - *How does your child feel about them knowing/not knowing - have they ever raised any concerns about this?*
- Does their condition preclude them from engaging in some activities with peers in school? E.g. school trips, extracurricular activities, parties in school etc.
    - *How is this managed?*

Section D: Medical & Physical Impact

- Can you tell me about your child's current treatment.
- Can you tell me about the physical impact of cystinosis/treatment for your child and how this impacts on his/her educational experience?
- Does medication need to be administered in school?
    - *If yes, who administers it?*
    - *Are they reluctant to take medication in school?*
    - *Has the school staff been provided with specific training regarding administration of medication? If yes, who provided this?*
- Does your child ever have to be withdrawn from class due to aspects of their condition e.g. medication administration, breaks, side effects (nausea etc) etc.
    - *Do you think this has any impact on them (socially/emotionally/academically?)*
    - *Is there anything you think that would be helpful to manage this?/What do you think helps with this?*

Section E: Psychological Wellbeing

How would you describe your child's typical mood/temperament?

- *Do they report any feelings of anxiety, sadness, anger, feeling judged or different etc related to his/her diagnosis, particularly in relation to school.*
- *Have they received any psychological support? What about in school?*

How would you describe their behaviour at home?

- If there are challenges, what are they in relation to?
- What helps here?

Do you have any concerns about their behaviour in school?

- If there are challenges, what are they in relation to?
- What helps here?

Section F: Home-School Relationship

- How would you describe the home-school relationship?
    - *What has been/would be helpful to develop this relationship?*
- What kind of a relationship does your child have with school staff?
    - *What has been/would be helpful to develop this relationship*?

Section G: School Training and Knowledge

- Have the school received any support/consultation through primary care/NEPS/hospital in relation to supporting the them or learning more about their condition?
- Has the school been provided with an opportunity to learn about cystinosis and the impact that this has on your child?
- Do you feel that school understands your child's diagnosis and how this impacts on them at an individual level?
- What training would you like to see offered in the future?
- What supports or resources do you think would be helpful to improve your child's school experience?

Section H: Closing Questions

- Is there anything else that you would like to add in relation to your child's experiences in school?
- Do you have any questions?

Interview Closure

Thank participants for taking part. Explain that as some participants may experience stress or upset speaking about the wellbeing of their child, a follow up letter will be provided immediately following the interview containing contact information for support organisations. This letter also contains the contact details of the interviewer and research supervisor should they have any questions or feedback regarding the research.

Appendix G: School Interview Schedule

UCD School of Education

Roebuck Offices Room 0.06
University College Dublin,
Belfield, Dublin 4, Ireland

T +353 1 716 7967
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Scoil an Oideachais

Áras Roebuck
An Coláiste Ollscoile Baile Átha Cliath,
Belfield, BAC 4, Eire

sarah.walshe@ucd.ie

Teacher/SNA Interview Schedule

Introduce self and aims of the research project.

My name is Elaine Mellotte, I am a trainee educational psychologist in University College Dublin.

The aim of the project is to explore the unique strengths and needs of children with cystinosis and identify barriers and facilitators faced in the context of the education system. This will be ascertained through interviews with parents, teachers and special needs assistants (if applicable) of children with Cystinosis.

I am undertaking this research under the supervision of Dr. Joyce Senior, an educational psychologist and Director of the Professional Doctorate in Educational Psychology Training Programme in UCD.

Explain limits of confidentiality.

All information which is collected during the course of the research will be kept strictly confidential, unless limitations of confidentiality are breached. As a mandated person there are circumstances where I may be professionally obliged to break confidentiality. This may occur in two ways:

1. If a strong belief exists that there is a serious risk of harm or danger to the pupil or another individual.
2. Occasions when disclosure is required as part of a legal process or Garda investigation.

In such instances information may be disclosed to significant others or appropriate third parties without permission being sought. Where possible a full explanation will be given to you regarding the necessary procedures and intended actions that may need to be taken.

Opportunities for questions and provision of consent.

Ask participant/s if they have any questions about the research or data collection and answer any questions.

Check that they consent to the interview being audio recorded.

Check that they consent to their data being transcribed (converted into written format) by a third party company if required.

Ask if they are happy to proceed. If yes, inform them that you will begin recording.

Section A: Interests and Strengths

- Tell me a little about the pupil. What does he/she like to do to have fun?
- What are his/her strengths (e.g.. kind, creative, funny, sporty, etc)?

Section B: School Experiences

Attendance and Engagement in School

- What, if any, impact does the pupil's condition have on their ability to attend school?
    - *If yes - How do you think this impacts him/her? E.g. Falling behind in schoolwork/stress in catching up/social opportunities/hospital education services*
    - *How many days? Do they have access to home tuition? Summer provision?*

- Has cystinosis impacted on his/her ability to participate in activities at school? (academic/physical/social/extracurricular)**.**
- Does the pupil like to go to school?
    - *What do you think helps here/ What makes this difficult?*
- Has there been any involvement in school from outside agencies e.g. NEPS in relation to the pupils needs?
- Have there been any challenges with transitions e.g. starting school/changing teachers

Academic Attainment

- What subjects does the pupil enjoy most?
- Are there any they find more challenging?
- Do you have any concerns about the pupil's academic attainments?
- Does the pupil receive any additional teaching support (e.g. special education teaching/learning support)?
- Do they have a Student Support Plan/IEP?
    - *If Yes, who was involved in setting the targets?*
- Do you have formal review meetings for parents and school staff e.g. care planning meetings?
    - If yes, who is involved in these?
- Do you have any concerns regarding
    - ability to organise themselves or manage the school routine?
    - follow instruction?
    - concentration/attention to tasks?
    - independence?

- Do you know what is homework like for him/her? Does it need to be adapted to their needs academically and physically (e.g. tiredness, feeling sick etc)?

Section C: Social Experiences in School

- What are their relationships with peers in school like? (e.g. strong network of friends/are they excluded/treated the same by peers and parents, concerns regarding teasing/bullying/being different)
    - *Does cystinosis impact on his/her ability to make friends? If yes, how so?*
    - *What helps/would help them to engage socially in school?*
- Do the pupils' peers know about and understand their condition?
    - *If peers have been informed of cystinosis, how was this done and by who? If not, why?*
    - *How do you think the pupil feels about them knowing/not knowing - have they ever raised any concerns about this?*
    - *Have their peers ever asked questions about their condition?*
- Does their condition preclude them from engaging in some activities with peers in school? E.g. school trips, extracurricular activities, parties in school etc.
    - *How is this managed?*

Section D: Medical & Physical Impact

- Can you tell me about how the physical effects of cystinosis and/or treatment impacts on his/her educational experience?
- Does medication need to be administered in school?
    - *If yes, who administers it?*
    - *Are they reluctant to take medication in school?*
    - *Have the school staff been provided with specific training regarding administration of medication? If yes, who provided this?*
- Does the pupil ever have to be withdrawn from class due to aspects of their condition e.g. medication administration, breaks, side effects (nausea etc) etc.
    - *Do you think this has any impact on them (socially/emotionally/academically?)*
    - *Is there anything you think that would be helpful to manage this?/What do you think helps with this?*

Section E: Psychological Well Being

How would you describe the pupil's typical mood/temperament

- *Do they report any feelings of anxiety, sadness, anger, feeling judged or different etc related to his/her diagnosis?*
- *Have they received any support for psychological wellbeing in school (e.g. NEPS, groups in school etc.)*

How would you describe their behaviour in school ?

- If there are challenges, what are they in relation to?
- What helps here?

Section F: Home-School Relationship

- How would you describe the home-school relationship?
    - *What has been or would be helpful to develop this relationship?*
- What kind of a relationship does the pupil have with school staff?
    - *What has been or would be helpful to develop this relationship?*

Section G: School Training and Knowledge

- Has the school received any support/consultation through primary care/NEPS/hospital services in relation to supporting the student or learning more about their condition?
- Has the school been provided with an opportunity to learn about Cystinosis and the impact that this has on the pupil?
- Do you feel that school understands your child's diagnosis and how this impacts on them at an individual level?
- What training would you like to see offered in the future?
- What support or resources do you think would be helpful to improve the pupil's school experience?

Section H: Closing Questions

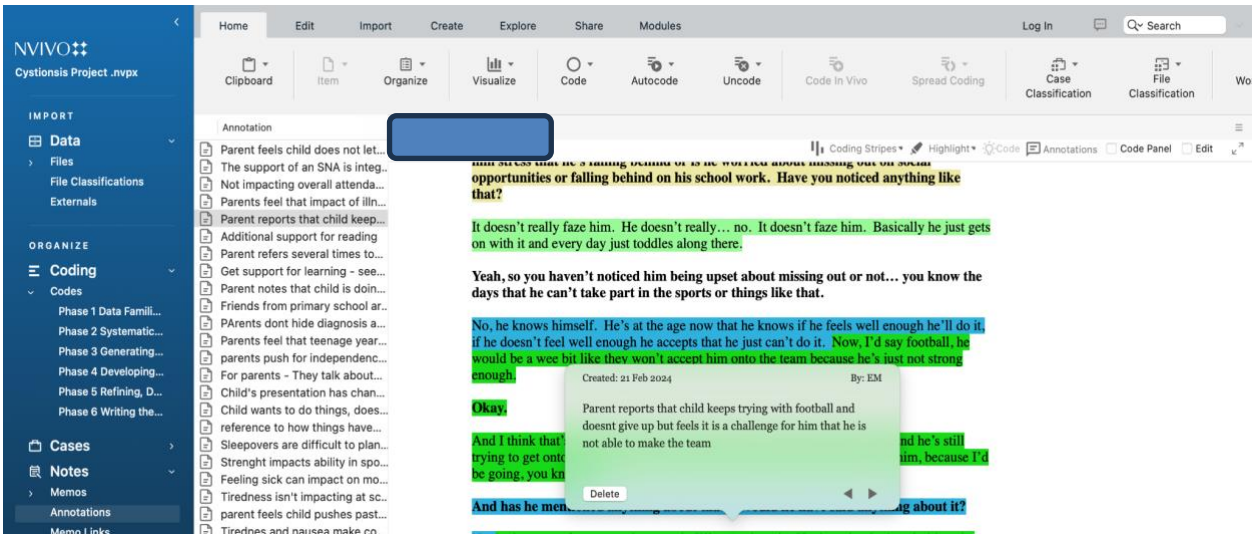
- Is there anything else that you would like to add in relation to the pupil's experiences in school?

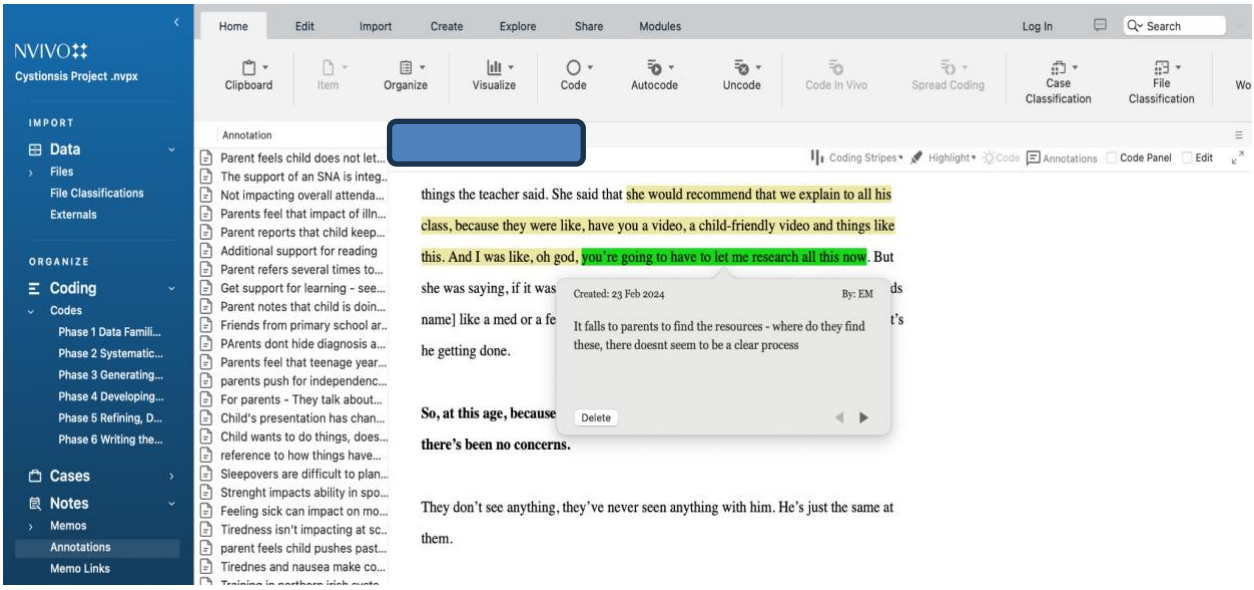
- Do you have any questions?

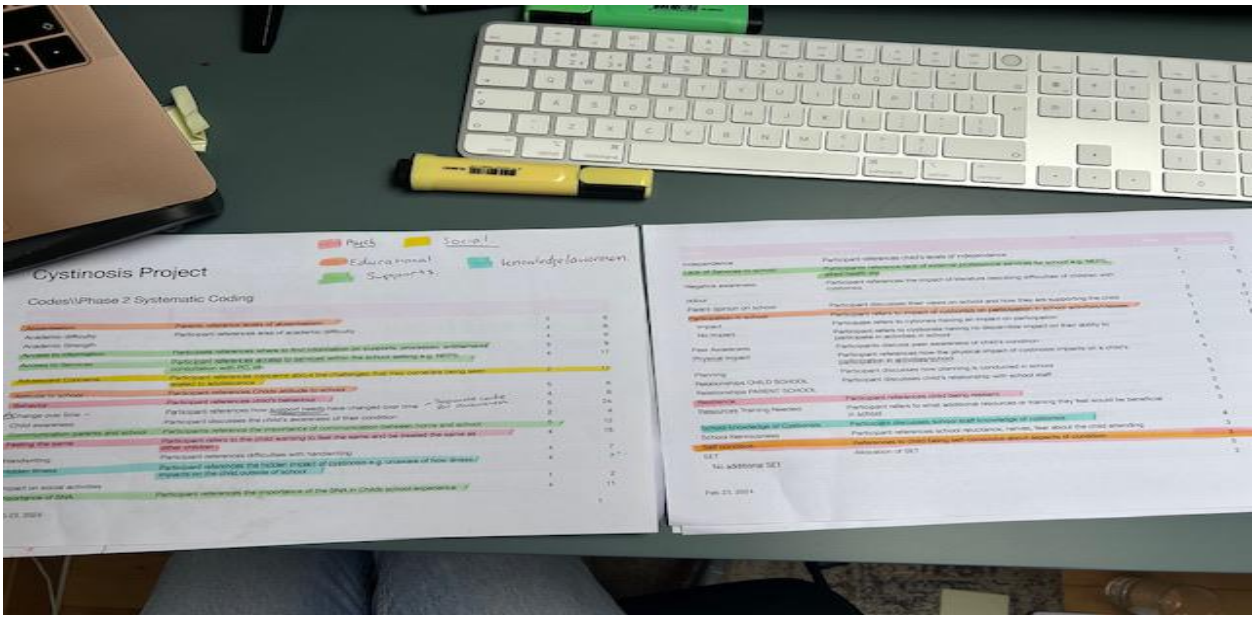
Interview Closure

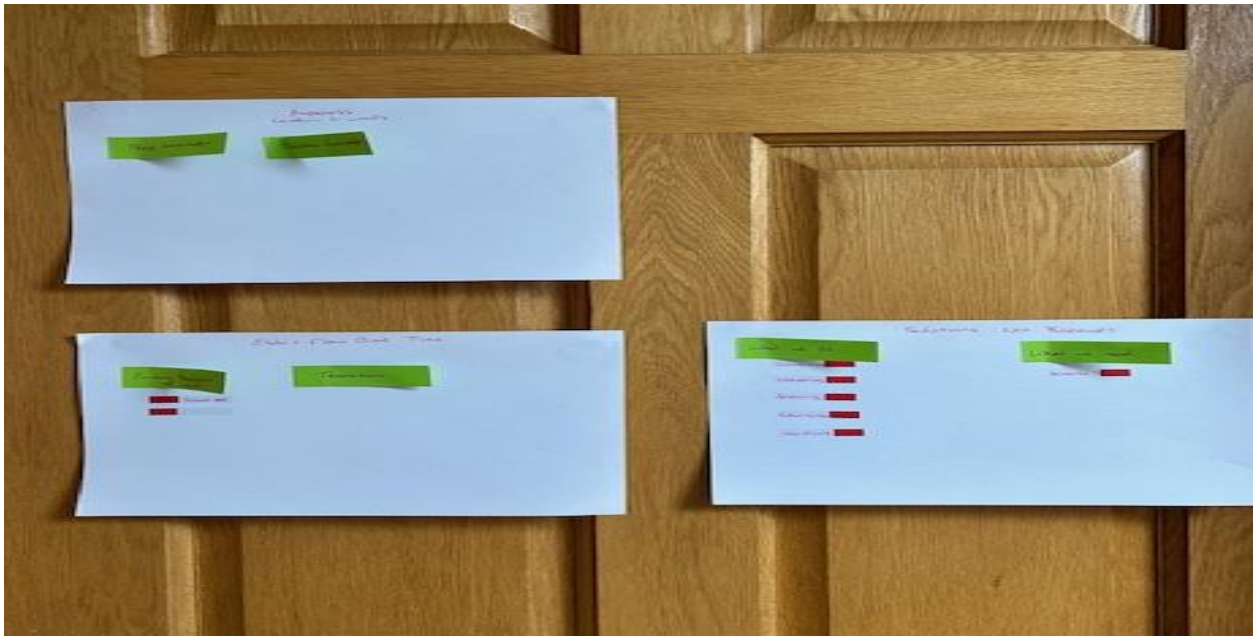
Thank participants for taking part. Explain that as some participants may experience stress or upset speaking about the wellbeing of the child, a follow up letter will be provided immediately following the interview containing contact information for support organisations. This letter also contains the contact details of the interviewer and research supervisor should they have any questions or feedback regarding the research.

Appendix H: Samples from Reflexive Thematic Analysis Process






Cystinosis Project
Codes\Phase 2 Systematic Coding



Appendix I: Ethical Approval New Researcher

UCD CONNECT

Elaine Mellotte <elaine.mellotte@ucdconnect.ie>

HS-21-61-Senior Amend, Extend & New Researchers Request APPROVAL

3 messages

hrec@ucd.ie <hrec@ucd.ie> 1 March 2023 at 11:44
To: Joyce Senior <joyce.senior@ucd.ie>
Cc: Agnes Fitzgerald <agnes.fitzgerald@ucdconnect.ie>, Elaine Mellotte <elaine.mellotte@ucdconnect.ie>

Dear Joyce

Thank you for your request to amend and add two new researchers (27/02/23) which has been **granted approval.** This is for the work and time period specified in your submission and is subject to the conditions below:

- The time period of the ethical approval for this study is valid until **June 1st, 2024;**
- Requests to amend and extend are limited to five requests within two years of the first approval date.
- The details of the approvals for **HS-21-61-Senior** date are as follows:

1. **Approval: 02/11/21**
2. **Amendment & New Researchers Approval: 01/03/23** *[new researchers: Agnes Fitzgerald Elaine Mellotte]*

- This approval is granted on condition that you ensure that, in compliance with the Data Protection Acts 1988 and 2003, all data will be managed in accordance with your application and that you will confirm this in your End of Study Report (HR6);
- You may require copies of submitted documentation relating to this approved application and therefore we advise that you retain copies for your own records;
- Please note that the granting of this ethical approval is premised on the assumption that the research will be carried out within the limits of the law;
- Please also note that approved applications and any subsequent amendments are subject to a Research Ethics Compliance Review.

If you have any queries regarding the above please contact the Office of Research Ethics and please quote your reference in all correspondence.

Regards

[redacted]