Living with a rare condition can have a significant impact on childhood, adolescence, and the formative stages of adulthood. During these years, interactions with individual healthcare professionals can have a significant impact on a young person's life and relationship with the condition.
Using case studies, explore the impact of a rare condition on young people. You may explore issues such as identity, social life, education, and relationships with healthcare. Reflecting on both the case studies and your own experiences, how can individual healthcare professionals create a positive healthcare experience for children and young people?

The Impact of Living with Cystinosis on Young People: A Case Study Approach

Rare Diseases

The European Union and the UK define a rare disease as one with a prevalence of less than 5 per 10,000 people [2]. Although individually uncommon, collectively 3.5 million people in the UK live with a rare disease, with a shocking 1 in 17 affected during their lifetime [1]. Of the 7000 identified conditions, an alarming 69.9% are pediatric onset yet only 5% have an authorised treatment [2]. Tragically, this leads to 3 in 10 children passing away before reaching their 5th birthday [1].

Rare diseases are characterised by limited understanding and research, often resulting in misdiagnoses, delayed treatment, and fragmented healthcare services. Children living with rare conditions face significant challenges in their education, mental health, identity, and social lives. The burden on these individuals and their families is extensive, highlighting the importance of understanding and addressing patient perspectives [1].

Cystinosis is a rare genetic disease that is diagnosed in early childhood. Estimates suggest that cystinosis affects between 1 in 100,000 to 1 in 200,000 live births. Each year, 2-3 children in the UK are diagnosed with this lifelong disorder. While the disease primarily strikes the kidneys, other organs such as the eyes, thyroid, and pancreas can also be involved. Despite being a systemic disease, it is one of the few rare conditions for which a treatment has been approved [4].

This essay explores the consequences of rare diseases on children through a case study of cystinosis and discusses the role of healthcare professionals in mitigating these effects.
Pseudonyms are applied throughout to maintain anonymity.

Cystinosis

Cystinosis is an autosomal recessive condition caused by mutations in the CTNS gene. It is distinguished by the accumulation of the amino acid cysteine which forms crystals in cellular lysosomes. These crystals disrupt the function of various organs in the body, predominantly the kidneys and eyes. There are 3 categories for this condition: infantile (nephropathic), juvenile (intermediate), and adult (ocular).
The infantile form accounts for 95% of all cases and is the most severe [5]. Clinical manifestations include short stature, dehydration, photophobia, recurrent vomiting, speech delays, muscle wasting, and congenital hypopigmentation. Furthermore, it is the most common cause of Fanconi's syndrome in children [3].
Diagnosis typically involves testing for elevated cysteine levels in the blood. However, molecular study for the CTNS gene or slit lamp examination to detect corneal crystals can also be performed [6].

Cysteamine is the accepted drug for this disorder. It delays the progression of end-stage renal failure by 6 to 10 years thereby postponing the need for kidney transplantation during childhood [3]. Large cohort studies have revealed that long-term oral cysteamine therapy decreases the incidence of diabetes mellitus and myopathy from 28% and 60% to 0% respectively. Additionally, it lowers the risk of hypothyroidism and improves survival outcomes [7].
This drug is associated with side effects such as nausea, sulfur-like body odour, abdominal pain, rickets, and increased gastric acid secretion. Unfortunately, additional medications have to be consumed to manage these effects.
As the disease progresses, renal transplantation becomes inevitable [5].

Case Study

Chloe, a baby girl, is brought into A&E by her mother Stacey who is concerned about an ambiguous rash. “Have you noticed anything else unusual?” the Doctor asks. Stacey mentions that her daughter appears excessively thirsty and has been drinking large amounts of water. A blood test is requested and an abnormal result triggers further investigations and discussions among the multidisciplinary team.

Two weeks later, a phone call for Chloe reveals the diagnosis of cystinosis.
Hearing such news can be profoundly devastating for families and ongoing support from healthcare professionals is essential throughout the patient’s life.

The term ‘Diagnostic Odessey’ describes the maze of uncertainty and misdiagnosis that the majority of individuals with rare diseases endure, until an exit in the form of a definitive diagnosis is found [10]. This period can be emotionally taxing and frustrating, with studies suggesting that 50% of patients are dissatisfied with the information provided during the investigative process [14]. Often, families and patients find themselves in disbelief, facing feelings of insecurity about their identity.
To alleviate this problem, a greater awareness of rare diseases is needed within healthcare professionals. Chloe’s family expressed their gratitude to a senior doctor in A&E whose prior knowledge of cystinosis spared them a prolonged wait.

Every 3 months, Chloe visits the hospital to ensure her condition can be closely monitored and treatment can be appropriately adjusted. It is vital to educate families about cystinosis and its management to empower them to make day-to-day decisions and execute treatment plans effectively. Healthcare professionals should take time to thoroughly explain these aspects.
For many families, specialist centres can be located far from their homes and require long journeys. Therefore, good communication between sites is crucial to deliver coordinated care [13].
Moreover, the involvement of nephrologists, ophthalmologists, and geneticists offers a holistic approach to treatment and should be continued throughout childhood [5]. This team method should not only improve outcomes by minimising delays but also consider the patient and family as integral members of the care process [9].

Cystinosis Foundation UK (CFUK) is a charity that raises awareness and provides a sense of belonging for affected families. Chloe’s grandfather and patient advocate from CFUK, describes how conferences help to bring stakeholders together and allow families to learn from each other. He shares ‘It can give hope seeing other children a lot older than Chloe and it is reassuring to hear from families with more experience’. GPs can attempt to direct rare disease patients to charities like these that can offer extra support and reassurance outside of healthcare.

Cystinosis has a harsh regime of medications such as cystagon, eye drops, calcium, potassium, growth hormone, thyroid hormones, vitamin D, PPIs, riboflavin, and others [5]. This list of drugs has to be administered on a strict schedule, which can be challenging to manage.
Chloe has a gastrostomy tube inserted into her stomach to facilitate with medication delivery. This prevents Stacey having to disturb her sleep at night, although the tube can affect Chloe's self-esteem and influence her clothing choice and participation in activities [11]. Chloe is very active and enjoys playing with her twin sister but due to her short stature and higher voice, she is often mistaken as being younger. This may come as an issue when forming friends, leading to loneliness and social withdrawal. Chloe's grandfather emphasises the importance of public education which can reduce the isolation and discrimination experienced by patients [15].

Recently, Chloe has started taking her Cystagon orally which simplifies the medication routine. She also applies fragrance to overcome some of the side effects such as bad odour and breath (halitosis). As children grow older, they start to feel more different from their healthy peers and wish to be actively involved in their own care [11]. For children like Chloe, physiotherapists for help with muscle weakness and speech and language therapists for assistance with swallowing difficulties can be very incredibly useful. This can be a milestone for children and a sense of control can positively impact their mental health.

Chloe's school life is affected because the teachers must give her medicines during school hours, she may need to go to the toilet more often, and may wear sunglasses due to her photophobia. These adjustments can constantly remind children of their differences and contribute to anxiety and depression. School trips can be tough for Chloe due to the difficulty of adhering to medication timings and the need for a trained carer.
On average, children with rare disease like Chloe have 45.4 primary care episodes compared to 27.8 for those without [11]. These frequent appointments disrupt Chloe's education and interrupt her activities such as gymnastics, dance, violin and school choir. To address this, check-ups outside of school timings should be scheduled and more walk-in clinics should be established.
The Born in Bradford cohort study found that children with rare diseases were twice as likely to be admitted to hospital with the mean length of stay being 5.3 days. Additionally, 50% of the sample fell below the education requirement for KS1 [8]. Although Chloe is slower at reading and numbers, she does not require extra lessons and can keep pace with her peers. In spite of research indicating that cystinosis may affect cognitive ability, short-term memory, and cortical atrophy, it is essential to digest that each case and individual is unique [12].

Conclusion

The prognosis has improved drastically since cystinosis was first identified. However, this achievement is accompanied with unawareness regarding the long-term effects of the disease and its medications [3]. Living with cystinosis remains challenging and issues surrounding identity, education, social life, well-being, and access to healthcare illustrate this point.
Increasing funding for specialist centres and further training on rare diseases for healthcare staff and doctors, especially paediatricians, would be beneficial. Lastly, efforts for new drug trials and research are necessary for advancing treatment options.
Although the prevalence of a rare disease is low, we can acknowledge through this essay that the impact at an individual level is extremely high.

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