Horses, zebras, or simply hoofbeats? The responsibility of healthcare professionals towards young people with rare conditions

Rare conditions

As medical students, we are taught a simple rule of thumb from the beginning of our foray into medicine. When you hear hoofbeats, think horses, not zebras. This saying emphasises the importance of considering common pathologies before rare conditions. We are advised that rare conditions are 'low yield' and 'probably won't come up'. However, is it ever justified to ignore potentially life-saving knowledge simply for efficiency? It is the nature of medicine to put the patient at the heart of everything, with the ultimate goal being their well-being. If knowledge of rare conditions can benefit even one patient, it is important to recognise that hoofbeats are hoofbeats, whether they are from horses or zebras. Healthcare professionals (HCPs) must do all they can to catch the patients with the condition that 'probably won't come up'.

A rare condition affects fewer than 1/2000 people [1], yet approximately 300 million people globally are thought to be living with a rare disease [2]. Furthermore, 70% of rare diseases affect children [3], with 30% of these dying before the age of 5 [2]. While these conditions may be individually rare, they are collectively common, and it is the responsibility of HCPs to create the most positive experience possible for the young people living with them. Young people with rare conditions fight an uphill battle every day just to live the life that so many of us take for granted. I had the privilege of speaking with X, a 9-year-old girl living with cystinosis, to gain an insight into her condition from her and her family.

Cystinosis- A case study

Cystinosis is a lysosomal storage disorder [4] found in fewer than 1 in 100,000 people [5]. Mutation of the CTNS gene results in the ineffectual transport of the amino acid cystine out of the lysosome [6] and the eventual accumulation of cystine crystals in various organs. Cystinosis is a genetic disorder inherited with an autosomal recessive pattern [5] and can present in many ways. X lives with nephropathic cystinosis, the most common and severe form of the disease, which tends to manifest in infancy and, untreated, can cause renal failure by 10 years of age [7]. Indeed, X's mother remarked that they were somewhat lucky to receive a diagnosis shortly after X's first birthday, which led to the implementation of a treatment regimen which ensures that X continues to grow as an inspiration to us all, a true embodiment of resilience.

X had an uncomplicated delivery alongside her twin, M, who is not affected by cystinosis. The first symptom that X's parents noticed was how much X was drinking, even attempting to suck water out of the sponge during baths. She was referred to paediatrics following this, at 11 months old, due to a finding of proteinuria. Subsequently, around the time of her first birthday, X had consecutive hospital

admissions as a result of hives over her body. The emergency department was prepared to send her home until one doctor asked a question which X's mother describes as having changed their family's life course: 'Is there anything else that you can think of?'. Upon being asked this, she recalled the proteinuria. One of the major manifestations of nephropathic cystinosis is Fanconi syndrome, a form of proximal tubulopathy which can cause severe electrolyte imbalances, polydipsia [7], and tubular proteinuria [8]. The symptoms X had been exhibiting pointed towards Fanconi syndrome, which can be the first symptom of cystinosis [7], and the recognition of this shortly led to her diagnosis. This was a condition that X's mother had come across in her research to understand the distressing symptoms her child had been exhibiting. However, this had been dismissed by her family due to its rarity; 'it can't be', they thought. Nevertheless, in this case, the 'low yield' information was crucial for X's diagnosis.

Impact of rare conditions on young people

Young people with rare conditions inevitably take a slightly different life course from what many might consider a standard childhood. Instead, these children tend to develop their sense of self as they find a new normal [3]. A common theme in the literature investigating young people's experience of rare diseases revolves around the psychosocial challenges that they face. They have to bear the brunt of social stigmatisation [9] and cope with isolation and lower self-esteem [10]. Thus, it is critical to explore the impact of rare conditions on young people, focusing on how they might affect their identity and social life. It was these topics that I hoped to explore further when talking to X and her family.

While treatment for cystinosis is not currently curative, the most specific treatment is cysteamine, which reduces lysosomal cystine accumulation by binding to cystine and allowing its efflux from the lysosome via system C transporters [11]. Cysteamine administration is usually accompanied by symptom management therapies, such as electrolytes, to counter the effects of Fanconi syndrome [11] and growth hormone injections to combat the growth retardation often seen in young children with nephropathic cystinosis [12]. Until the age of 5, X was on several oral medications, but, after developing hand, foot, and mouth disease, her tolerance for oral administration plummeted, which led to the insertion of a nasogastric (NG) tube. X's experience with an NG tube was poor. Alongside the difficulties that her family faced in using it to administer her medication, the tube had knock-on effects on her social life, making her feel noticeably different to her peers at school. Cystinosis continued to affect X's social life during her first school trip away from home. With no staff on the trip trained to administer her medication, X's mother had to drop in, leaving X on unequal footing with her peers, who were all experiencing their first trips away from their parents. Furthermore, a characteristic side effect of cysteamine treatment is a marked odour caused by the formation of sulfur compounds [13], something which will only have a more profound social impact as she grows.

An alternative perspective

It is perhaps natural to assume that children with rare diseases might struggle with their identity. However, the resilience of young people must not be underestimated. In fact, many young people do not see their rare condition as overtaking their identity [3]. This may be especially salient in X's case. Upon talking to her, I was immediately struck by her upbeat and smiling nature, extremely cognizant of her condition and yet determined not to see it as a burden. Apart from this being simply in X's nature, some of the credit must be attributable to her family. X's sister, M, has made concerted efforts to protect and help X. Moreover, their mother and grandfather have been actively involved with awareness campaigns around cystinosis. The culmination of this is X, who sees cystinosis not as her burden but as her superpower. X's uniqueness gives her the strength to deal with the myriad complications that most of us cannot imagine enduring at such a tender age. HCPs must take it upon themselves to cultivate a positive and enduring atmosphere for young people with rare conditions.

Responsibility of healthcare professionals

Given the milieu of care that X has experienced, she has had numerous, varied encounters with healthcare. X's mother broadly categorises them in two ways. The first entails HCPs who focus very much on the medical process, leaving little room for the input of X's family. In stark contrast, the second involves professionals who asked a critical question: 'What can we do to help?'. It is easy to forget that while HCPs may be able to provide medical care, there is a chasm between those who treat a disease and those who live with it. The physician-patient interaction often does not fit traditional assumptions when dealing with rare conditions [14]. Deferring to a patient's or family's specialist knowledge is not a weakness but an acknowledgement of their lived experiences. Furthermore, there must be a greater emphasis placed on support groups for families with rare conditions. Caregiver-caregiver support is paramount towards enabling individuals to thrive, reducing caregiver burnout and allowing families to lead meaningful lives [15].

Conclusion

Young people with rare conditions are undoubtedly impacted in a variety of ways. However, they develop their own relationship with their condition, creating a new normal for them. HCPs have a responsibility to make sure that young people's relationship with healthcare is as positive as possible while also supporting caregivers, which ultimately aids the patient further. Much of this begins during education; rare conditions may be rare individually, but they are collectively common and must never be dismissed as 'low yield' simply because of their improbability. There must also be a willingness to buck the traditional physician-patient roles and ensure that treatment is always patient-centric. This experience gave me much insight into how I plan on approaching my continuing journey into medicine. When I hear hoofbeats, I will think neither of horses nor zebras. I will think of hoofbeats. I will think of the person behind the patient.

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