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Many people with long term conditions are exposed to considerable uncertainty. For those living with rare conditions, this can be amplified due to the lack of information, services, and management.

Using a case study, explore how uncertainty can impact people living with rare disease and their families. Consider the psychological, emotional, and practical effects of effective and ineffective medical communication around uncertainty and health.

Background:

The United Kingdom and European Union define a rare disease, also referred to as orphan disease, as a condition which affects fewer than 1 in 2000 people.[1] Notably, the definition and prevalence of a rare disease differs in other countries such as the United States, where it is defined as affecting less than 200,000 people, making a conventional definition problematic.[1] Nevertheless, common elements remain inclusive such as the strong association between a rare disease and prolonged diagnosis, inappropriate management, and chronic debilitation.[1] In fact, the 2009 annual report by the Department of Health found roughly 40% of patients struggled to get a correct diagnosis for their rare disease.[2] Thus, unnecessary medical interventions were commonly reported, with 16% of participants reporting unnecessary surgery, and 33% reporting incorrect medical treatment.[2] Furthermore, Wakap et al. estimates there are currently over 6000 unique rare diseases globally, and 69.9% of conditions are exclusively paediatric onset.[3] Although an increasing number of children with rare diseases are surviving into adulthood due to advancement in treatment, the transition to expert adult services remains challenging.[2]

A prominent example is complex lymphatic anomalies (CLA), which are rare diseases of the lymphatic system. CLAs consist of 4 overlapping, congenital conditions: Gorhan Stout disease, generalised lymphatic anomaly (GLA), kaposiform lymphangiomatosis and central conduction lymphatic anomaly.[4] The clinical symptoms and the anatomical locations involved overlap between these conditions and diagnosis is often challenging and requires an interdisciplinary approach.[4,5]

This essay will be focusing specifically on GLA. Formerly known as lymphangiomatosis, GLA is a nonneoplastic condition characterised by abnormal proliferation of lymphatic channels resulting in multifocal lymphatic malformation and dilated, thin-walled lymphatic vessels.[5,6] Malformation is observed in both osseous and extraosseous tissue such as bones and thoracic and abdominal viscera; lesions in the spleen and liver are also commonly seen.[5,6] The presence of bone lymphatics is associated with pain and impaired mobility.[7] Although aetiology is poorly understood, Rodriquez-Laguna et al. has established the presence of PIK3CA variants in affected tissue samples.[8]

As mentioned above, diagnosis remains challenging and often relies on detailed patient history, specialised imaging techniques, biopsies, and histopathologic findings. Above all, however, a familiarity of the condition is critical to establish a clinical suspicion and timely diagnosis. This essay will explore the impact of uncertainty, diagnostic challenges and ineffective medical communication from a patient's and family's perspective. Crucially, it will highlight the need to improve services and information available to patients to reduce some of the psychosocial and emotional burden that is experienced.

Case study:

Alfie Milne was diagnosed with GLA at the age of 2 at Great Ormand Steet Hospital (GOSH), after a year of investigations. His mother, Tracy, initially noticed one of his legs was bigger than the other which prompted her to visit their General Practice, where he was referred to orthopaedics in the local hospital. Further symptoms such as pain, discomfort, bruising easily and discolouration of his legs arose. Alfie was subject to investigations such as regular bloods tests, ultrasounds, MRIs, and insertion of a Hickman line. Frequent hospital trips served as a significant burden on both Alfie and his families physical and emotional wellbeing. For instance, Alife's parents had to take time off work regularly; Alfie's older sibling was often left with friends and family and the travel from their home in Scotland to London was physically exhausting. Eventually, Alfie was referred to dermatology at GOSH, and after multidisciplinary involvement, a diagnosis was reached.

The diagnosis was a complete shock to Tracy, who states she was not prepared for such an outcome to arise from the initial symptom of a swollen leg. At the time, Tracy was only provided with a printout which had the name of the condition and 5 lines of information about GLA. She was told it was an orphan disease, with a wide spectrum of symptoms and complications, and the prognosis was poor with most patients surviving to the age of 5.

At the time, few specialist clinics were available, with a small number of patients at GOSH and St Thomas' Hospital. However, this was not the case for Alfie's local hospital, with clinicians never seeing or hearing about GLA prior to this. Therefore, there was very little information available at any of their local health services and communication to Alfie's family on the pathophysiology and prognosis were severely limited.

Likewise, literature online was limited, and Tracy recalls she could only find case studies based on severe cases of GLA and frightening to read about. Most literature was not in patient friendly terms and difficult to make sense of, contributing to the feeling of uncertainty for Alfie's future. This meant she shied away from independent research and further reading for a long time.

Furthermore, Tracy stated it was very isolating for Alfie, and the wide variability of an already rare disease meant it was hard to find a community to belong to. Alfie's walking deteriorated with time, and mobility became a significant issue. Although he was undergoing physiotherapy, Tracy recalls it was challenging as GLA had caused severe deformity of his lower limbs, and eventually a decision was made for him to be a full-time wheelchair user. She remembers attending a conference with Alfie about GLA, and Alfie wondering why he was the only one with a wheelchair. Similarly, at one point Alfie was managed with vincristine, a chemotherapy drug, and referred to a cancer support group. Again, Alfie and his family struggled to fit in as GLA is a different disease and felt like they almost shouldn't be there as they couldn't connect to other patients and families.

Also, after the diagnosis, Tracy recalls Alfie's older sibling struggling with being the 'healthy child', and the emotional distress of Alfie being unwell, with little information on his prognosis.

A major turning point for Tracy was many years after Alfie's diagnosis where she stumbled upon a patient organisation in America through Google. She reached out and was provided with all the information available at the time on GLA including names of specialist doctors, current literature and research, and another family in the UK with the condition. Tracy states gaining a better understanding of the condition, and meeting another family gave her newfound hope. Following this, Tracy founded the Alfie Milne Lymphangiomatosis Trust, which later became LGDA UK, and has raised over £325,000 between 2012 and July 2022. Patient friendly information is available on the LGDA website and provides details of local support groups, and patient stories.

Discussion:

This case highlights several difficulties which arise from inefficient medical communication and a lack of services. Notably, the emotional distress from the lack of information provided by healthcare professionals, specifically in patient-friendly terms. Patients may struggle to find information independently and determine which sources are reliable and accurate to their condition, fuelling their feeling of uncertainty and fear. Moreover, they might struggle to explain what the condition is and what the future holds to others such as family and friends. This uncertainty and limited understanding may also cause issues with employment, and health insurance. Similarly, services such as support groups and specialist centres remain limited, and it is unsurprising that patients feel isolated and alone. Frequent travelling to such services has a physical and financial burden due to continuous disruptions at work, as well as a strain on mental health due to time away from family and friends.

Currently, Health Education England (HEE) has developed a Genomics Education Programme (GEP) to increase awareness of rare diseases amongst healthcare professionals.[9] The aim is to help clinicians recognise rare diseases more readily; feel more confident in relaying information to patients and

answering patient questions. GEP is further supported by a patient advisory group which encompasses patient views and experiences to ensure resources are reflective and accurate.[9] It also ensures that individuals recognise that although their condition may be rare, they are not completely alone. In 2009 the Department of Health aimed to further increase resources available by supporting international collaboration and sharing of information, as well as strengthening medical research.[2] This remains as an aim under the 2022 plan by HEE. Lastly, national coordination of care is under progress to ensure services are of a constant standard and readily available regardless of where a patient lives.[2,9]

Conclusions:

It remains clear that uncertainty and poor medical communication can have a significant impact on the wellbeing of the patient and family members. Moving forward, enabling access to accurate information and effective communication remains crucial for improving patient care. An understanding into current research and future advancements provides a sense of control over their management, hope of a better future, and a sense of belonging; something that should not be taken away from anyone with ineffective medical communication.

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