Helplessness

Helplessness – a horrible and arguably the worst state one can find themselves in. Unfortunately, it is also state, which to me, partly described the situations a patient and his family found themselves, when facing a rare disease.

JC is a 10 year old boy, who has been officially diagnosed with ring(20) chromosomal defect for the past 6 years. From hearing his story to finally being diagnosed, my insight into the reality of being in the "dark", when facing an unknown health condition, became, ironically, clearer. Unfortunately, occurrences of all rare diseases combined are paradoxically not as rare as I thought, affecting 1 in 17 people; this inevitably means most clinicians, including future ones such as myself, will have to deal with such conditions.

Phase 1: Pre-diagnosis

JC had a normal birth, and before the manifestations of the symptoms, his mother described him as a "lovely and quiet child". The first changes noticed where when JC was around 18 months old; as soon as he went to sleep, he would start screaming and hardly sleep throughout the night. This left the parents very concerned, but healthcare professionals at the time dismissed these as night terrors.

Later on, the problems at night faced by JC were not as bad for some time, but JC's mother mentioned how they noticed in April 2013, that he would "freeze as if someone pressed the stop button on him" for a few seconds at. These episodes gradually became more frequent and longer in duration. Also, the symptoms at night recurred, but this time the mother described them as occurring throughout the night – "It was like in the Exorcist! His arms stretched straight out, eyes were rolling in and out, and breathing was very fast and tense." However, even after being referred to specialist paediatrician and neurologists from the GP, there was no confirmation as to possible cause. The mother of JC, at this, expressed disappointment as the professionals were of no help and she felt pushed away.

Sadly, it is quite a common feeling for patients at times to feel "misunderstood and pushed away", as described by JC's mother. As a medical student, I do understand that it is easy to become so focused onto the biological aspects in a consultation, that one can easily miss out the psychological and social issues. As healthcare professionals, the primary aim is to improve the quality of life of the patients, which does not only consists of treating via medical means, but also dealing with the psychosocial implications. The concept describing this method of approach is known as the Biopsychosocial Model, which to me, essentially means viewing each patient as an individual fellow human being instead of simply a body not functioning properly. The mother described how there also was a complete change in JC's behaviour – he became aggressive and chaotic, having broken about 5 TV's which did not fit his usual self at all. Witnessing such erratic changes in a child is a mortifying experiences for any parent, and this can possibly lead to mental health issues; this demonstrates the use of the Biopsychosocial Model to identify these kind of issues in the life of the patients and their carers, allowing them to receive some kind of support,

Eventually, JC's continuous episodes at night, and the lack of support received, made the parents resort to A&E. This is where the doctors actually first time witnessed one of JC's episodes. "It was weird, because it was only having seen one of JC's episodes, that all the MRIs, CT scans and EEGs took place, but this was not the case, when we simply described the exact same thing as they witnessed.", explained JC's mother. A very important lesson can be learnt here- trust the patient. Trust is a key component in a doctor-patient relationship; however, I realise upon reflection, there is

very little emphasis on trust towards patients in comparison to towards the doctor. The history is an essential component of any consultation; thus taking a trusting stance when communicating with patients, will help to elicit the key details to make the correct diagnosis.

Phase 2: Diagnosis

After the incident in A&E, JC was put under the care of a specialist paediatrician. Only after months of investigation, did the consultant suspect something rare as the diagnosis. It was in December 2013, once the correct genetic testing had been done, that it was confirmed that JC actually had Ring(20).

Finally, the cause of all the problems JC and his family were persevering though for what seemed like a lifetime, had been finally revealed. Nevertheless, "shocking" was the word used to describe their feelings at the time. After all the time, to understand that it was a mere genetic defect, which was ultra-rare, JC's parents realised, even though a light was shed upon the condition of JC, they were still helpless as there is no cure or treatment guidelines.

One aspect of the JC's diagnosis I would like to point out is the duration for which the consultant was working with the family to help JC. Even though, all standard investigations came back as negative at first, the consultant in this case did not give up for months. It is common to see patients with rare disease having to wait a long time before their conditions are finally diagnosed; during those times, it is of utmost importance, that as doctors we continuously support these patients and make sure that they know we are trying our best.

As I have mentioned before, "helplessness" is a common theme in rare diseases, as often clinician are unable to deduce the diagnosis for rare disease for a long time, as was the case for JC. Hence, as doctors, it will make a great difference if we share the feeling of being in the dark, since for such a patient, the very knowledge that they are being looked after and supported, can greatly reduce this feeling of helplessness. This was the case for JC and his family, who praised the consultant for her efforts in finding the cause.

It is clear that awareness of conditions and what each family goes through plays critical role in the diagnosis such rare disease. This is shown by the world map plotted by Ring20 Research group, which demonstrates the distribution of most of the identified Ring20 cases around the world. Although, in terms of the statistics of the condition occurring there is meant to be an even distribution across the countries, it is clear some areas are more clustered, ironically those with major children hospitals with the specialists, who are more likely to be aware of Ring(20) (as shown in figure 1).

Figure 1 - Map of USA, demonstrating cluster towards the east coast

In order to improve the awareness, I propose the use of social media, in the form of podcasts. As a medical student, I understand that to expect us to learn all rare conditions known is an impossible feat. Nonetheless, the aim of the podcasts will simply be to gradually increase the awareness of rare condition. Each podcast would be focused on one rare disease and will include an interview with a patient with the particular condition, discussing their experiences. Through this, not only will there be an increased awareness of the various rare conditions, but also increase understanding of the social and psychological implications of having a rare disease. Thus, not only will the likelihood of

diagnosing rare conditions increase, but also by listening to the perspective of patients, will allow doctors to exercise empathy to a significantly greater degree.

Additionally, the theme of “helplessness” can also apply to the doctors caring for patients with rare diseases – for example, Ring(20) has no management guidelines currently, which means doctors will have to resort trial and error; these are rarely effective. Although, clinical guidelines are in the process of being synthesised, it is important to also highlight the existence of such guidelines once finalised – this could be another aspect of the podcast.

Phase 3: 6 years after diagnosis

To this day, JC still has daily absence seizures and the nightly episodes, with added difficulty in coping with certain aspects in life. The mother of JC did complain of a lack of support for JC in school as due to the ring(20) there is some degree of cognitive impairment – she was afraid that JC may be “kid in class, left by himself”. It was sad to hear, especially when she compared her child to children with autism, and how much more support they receive, whilst her child, whose condition is not so well known, is not in the reception of that kind of support. Thus, with the use of podcasts, the aim is to both increase awareness in the medical community and also the general population.

“Helplessness” is sadly a recurring theme in the healthcare of rare diseases, for both patients and doctors. In this essay, I outlined the lessons I have learnt, to tackle this major issue.