

“What’s the Point?” An Insight into Mental Health in Rare Diseases

“Living with a rare disease is a life-long learning experience which invariably leads to challenges with mental health alongside physical symptoms. Use a case study to demonstrate how future doctors can learn from patients to improve the management of these complex conditions”

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Introduction

Dealing with a rare disease can be a huge challenge that also provides an idealistic environment for mental health issues to develop in. A survey led by Rare Disease UK reported that 95% of the respondents felt worried or anxious, 90% have felt low and 88% have felt emotionally exhausted (1). This considerably high emotional impact highlights the need to discuss different methods in which clinicians can help improve their patients' quality of life.

I was fortunate enough to speak to S, an adolescent female who suffers with Wolfram Syndrome, a rare inherited disease, about her experiences with the disease and her mental wellbeing. Having insight into one patient's entire journey, from diagnosis to treatment, can help give perspective to the delicate issues faced by many other patients with rare diseases; the lessons that can be learnt are valuable to share and reflect on.

Background: Wolfram Syndrome

Wolfram syndrome (WS) is an inherited autosomal recessive disease that impacts up to 30,000 patients worldwide (2). It is quite a unique disease because it affects such a wide range of organ systems, and every patient has a different set of symptoms. This can make WS difficult to diagnose; most patients are diagnosed into their adolescence as their symptoms become more progressive, much like many other rare diseases.

WS is also known as DIDMOAD. This is an acronym for the 4 main symptoms that are typically characteristic of the disease:

- Diabetes Insipidus
- Diabetes Mellitus
- Optic Atrophy
- (Sensorineural) Deafness

Although current treatment is symptomatic based, the TREATWolfram research trial is looking into the use of sodium valproate to slow down the brain degeneration and potentially stop the disease (3).

The Power of Listening

For S, WS began with an early diagnosis of type 1 diabetes mellitus when she was 4. At such a young age, she was faced with the battle of various injection regimes, which also took a toll on her parents who tried their best to support her. At aged 6, S then began to notice her eyesight worsen. After visiting the opticians on several occasions, she was given glasses and was also allowed to sit at the front of her classroom. This was an exciting moment for her; she was relieved to finally have found a solution to this issue that was affecting her everyday life. However, she was left disappointed because, despite all her efforts, she still couldn't see clearly. When she told the optician and her teacher this, they both concluded that she was being pressured into saying this at school due to bullying, and that S could in fact see. This made her feel very helpless and lost, as she couldn't understand why no-one valued her concern or believed her.

Being diagnosed with varied health problems and not knowing how (or even, if) they link can be frustrating for patients. This is an understandable cause to feel defeated or insecure. Although S didn't get the support she needed at the time, it's an important reminder for clinicians to be open minded when taking a patient medical history. Every patient has been through a different (possibly difficult) journey leading up to their consultation with the clinician, so being hasty to come to a conclusion or being dismissive can really affect the patient's confidence and willingness to co-operate. At that point S wasn't suffering from any mental health issues, but experiences like these can trigger the feeling of invalidation which can build up over time and lead to anxiety or depression in the future (4).

The Power of Dialogue

Unfortunately, poor eyesight and being diabetic was just the start of S's health issues. By the age of 11, S was suffering with intense bladder and bowel issues. These were the worst symptoms she experienced as part of WS, because of how severely they affected her quality of life. For example, she would wake up every 30 minutes during the night to relieve her bladder. This meant she had difficulty concentrating in school and always felt tired due to her lack of sleep. As well as this, her bowel problems meant that sometimes she couldn't even make it to school before needing to turn back and go home. The social isolation that was caused due to her symptoms meant that S couldn't leave her house freely without the fear of being too far from a bathroom, potentially leading to an embarrassing incident. This idea is exacerbated by the fact that her peers might not be understanding of her condition, due to the

lack of awareness. This could perhaps lead to insensitive comments by those around her, leaving S with a level of trauma that is indelibly with her for life.

Fortunately, S was encouraged by her doctors to attend the annual WS Conference, where various healthcare professionals, other patients and their families meet to share their experiences and updates on new research. At the conference, S was able to discuss her bladder issue with another patient, who shared a similar experience to her. He recommended a drug called Desmopressin, which dramatically helped him improve his bladder issue. S was then able to discuss this with her doctor, and since that conversation a few years ago, S is still very grateful to that patient who shared his advice, as she is now able to sleep comfortably and rarely wakes up during the night because of the medication he suggested. It's uplifting to hear how impactful these discussions can be for patients; not only did this simple suggestion improve S's quality of life, it also made her optimistic about her health and made her feel more in control of her body. S and the other patient who made the suggestion both continue to regularly attend the conference to share their experiences and help empower others.

The Power of Support

Having spoken to S in depth about the various issues she faced whilst growing up, it was clear that she heavily relied on the support from her parents to help her through some of her tougher days. She especially credits her mother who helped her, both physically and emotionally, through her diagnosis. Although there is no cure to the disease, S and her mother are both motivated to learn more about new treatments and the current research that is going on with WS. Doing this is so important, because it keeps the patient feeling like they are included in the conversation, and having a family member who can provide support whilst doing this can help make it easier and less stressful.

S described one situation where she was much younger and had an embarrassing bowel incident at school; she isolated herself from everyone by staying in her room and refused to go back to school. "What's the point?", she told her mother, as she tried to convince her to get out of bed. S explained how her mother encouraged her that although these kind of incidents are out of her control, they shouldn't impact her day or how she views herself; her health issues shouldn't define who she is. Having supporting parents and a secure home life are basic protective factors in safeguarding the mental well-being of a person, especially if this is familiarised from childhood (5). While this isn't something clinicians can actively

learn from, the value of a support system is an important point for the healthcare team to be aware of and monitor.

Moving Forward

It is quite overwhelming to try and summarise all the different ways healthcare professionals can improve the clinical experience of patients. But, a good starting point is to evaluate the clinical environment in which the patient visits. I think that creating a comfortable clinical environment can sometimes be more important than feeding the patient loads of information. Although the patient might have a brilliant rapport with the psychologist, it's essential that they feel they are able to have the same conversations with the clinician as well, if necessary. By making a proactive effort to check up on your patients' mental wellbeing during every appointment is a great way to start this conversation. It can even be as simple as asking them how their mood has been lately, or what they've been thinking. Not every consultation should be prescriptive; patients, especially those with rare lifelong diseases, should feel they can talk to *any* member involved in their healthcare.

Conclusion

Promoting better mental wellbeing isn't restricted to just those with rare diseases; it is a challenge of the underfunded health services in the UK that is being tackled by everyone suffering. Those patients diagnosed with rare diseases are more vulnerable to feeling lost and alone. It is our jobs as healthcare professionals to make sure that they feel as involved as possible, for example, in up to date research or meeting other patients with the same rare disease. It might not be in every patient's interest to do so, but knowing they have the option to be more involved can be just as reassuring.

References

1. Living with a rare condition: the effect on mental health (2018) - Rare Disease UK [Internet]. Rare Disease UK. 2019 [cited 18 November 2019]. Available from: <https://www.raredisease.org.uk/our-work/living-with-a-rare-condition-the-effect-on-mental-health-2018/>
2. Wolfram Syndrome Explained - The Snow Foundation [Internet]. The Snow Foundation. 2019 [cited 18 November 2019]. Available from: <http://thesnowfoundation.org/wolfram-syndrome/>
3. Clinical Trial & Updates – Wolfram Syndrome UK [Internet]. Demo.wolframsyndrome.co.uk. 2019 [cited 18 November 2019]. Available from: <http://wolframsyndrome.co.uk/clinical-trial-updates/>
4. Causes of anxiety | Mind, the mental health charity - help for mental health problems [Internet]. Mind.org.uk. 2019 [cited 18 November 2019]. Available from: <https://www.mind.org.uk/information-support/types-of-mental-health-problems/anxiety-and-panic-attacks/causes-of-anxiety/#a>
5. World Health Organisation. Risks to Mental Health: An overview of vulnerabilities and risk factors. 2012.[cited 18 November 2019]. Available from: http://www.who.int/mental_health/mhgap/risks_to_mental_health_EN_27_08_12.pdf