"A Day in the Life: the experiences of disjointed care"

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8:00 am

Ward 36

You're here again, the bustling ward. The high-pitched screech of your pager is echoing. The bays of patients are at full capacity today, and you worry you won't be able to see everyone before your clinic this afternoon. You see patient after patient. You remember none of their names.

You hope to yourself that you will finish in time for your home-cooked dinner tonight.

08:00 am

Hotel room

You're here again, a silent hotel room. It is finally time to get out of bed, the night's sleep was interrupted by intrusive thoughts. A day full of appointments lies ahead, seeing specialist after specialist. How are you not used to this by now? The well-rehearsed explanation of Wolfram Syndrome plays over and over in your mind, remembering your last appointment is with a new doctor.

You hope to yourself that they will see you as more than just your condition.

12:00pm

Doctor's Mess

You step into the doctor's mess and as the heavy fire door closes behind you, you get a moment of complete bliss. You trudge over to the fridge and collapse into one of the large comfy sofas. Here, you reflect on the patients you've seen this morning and although you're sure you have provided them with the best clinical care, you feel guilty that you don't have the time to get to know them.

As you finish the last bite of your sandwich, you walk to the computer and pull up the list of patients you have to see in your afternoon renal clinic. You read through some referrals and start to gather ideas. Skimming through the notes, your mind bounces from disease to disease: these are all textbook diseases, knowledge that you have known since your undergraduate training. But then, your confidence cracks, Wolfram Syndrome (WS). What is

that? Panic sets in. Should I know what this is? Typing words into Goggle you get a brief summary:

Wolfram syndrome is a rare autosomal recessive disease that encompasses four common features (Diabetes insipidus, Diabetes mellitus, Optic atrophy and Sensorineural deafness)[1].

Although you want to read more, the first patient in your clinic arrives and you close the tabs. There's never enough time. Medical school does not prepare you for the real-life interaction with patient with rare diseases, nor do you as a student, have time to learn everything. Immense guilt encompasses you once more, feeling your lack of knowledge may neglect the quality of care you can provide for this patient.

12:00pm

Café

Three out of your four appointments are over, you head to the café where you collapse into the chair. You feel overwhelmed, this is a tiring process both physically and mentally.
The phone rings, on the other end of the line is a close friend checking to see how the day is going. Having WS herself, she is one of the only people that you can express your deepest worries to. You tell her that you are scared that your sight is worsening, she doesn't jump to the dismissal and the positive support; instead she listens and confides that she is petrified of the same thing. You both joke about how your boyfriends will get better at jump scaring you. Your dark humour sometimes seems obscure to others but to you it's the medicine you need.

As the time for your next appointment approaches, anxiety seeps in. Previously, doctors have not believed your symptoms to be real, they said that your extreme thirst is a habit and not a diagnosis of diabetes insipidus. On the contrary, some doctors have related any symptoms you have to your WS diagnosis and even went on to miss key signs of sepsis. The two extremes of your care worry you deeply.

Your next appointment is in an unfamiliar area of the hospital. Your uneasiness returns, finding your way around the hospital, can often be challenging for someone with a visual impairment. Why is there not better support during these appointments?

You are taken to the consultation room. Good news? Bad news? More diagnoses to add to the list? Will they even listen to me?

“Hi I am Dr. Brown…”

The doctor explains to you the reason for the appointment and she is honest about having no knowledge of your syndrome – you appreciate that. Their honesty means you can build a foundation together in order to understand how your symptoms may or may not relate to WS.

You think back to all the times doctors have changed your medication and diagnoses based on their prior opinions and knowledge without considering yours. Thinking back to the start of your journey with WS, you were thrown from specialist to specialist for months at the young age of 12; at the time you were so innocently oblivious to what was happening. Uncertainty about your health not only lay with you and your family but also the doctors looking after you. Medicine encompasses the art of problem solving and piecing together the unknown to come to a general consensus. How are you to solve a puzzle where the end goal is unknown? The doctors never had enough time to research your symptoms and thus your diagnosis was driven by the research of your parents – you worry about those that don’t have the support you did.

As the years went on you were told not to Google information about your condition, but you did, of course, and you found yourself worried about the future. No one really talked to you about how to keep yourself mentally healthy, and as such you have experienced depression and anxiety.

What matters the most to you is that healthcare professionals actively seek to improve their knowledge on rare diseases. It is okay not to know everything about every disease. Albeit this is hard for doctors who always strive to obtain the most knowledge. It is okay to show enthusiasm about meeting a person with a rare disease but acknowledging that they are much more than their physiology. Finally, it is okay to ask the hard questions about what a patient is concerned about. This opens an opportunity to build valuable rapport so that you can learn from a patient and help develop excellent patient care. It is important to you that doctors from many specialities work together, to provide you with the best care and communicate with each other to educate other.

A rare disease is with you 24 hours a day and not just in the consultation room.

Final Thoughts

Wolfram syndrome is a rare neurodegenerative disease that affects 1 in 770,000 individuals each year. It is an autosomal recessive condition, affecting the WFS1 gene on chromosome 4p16.1[1]. This genetic mutation inhibits the production of wolframin protein, that is present in all cells; predominantly in the pancreas, inner ear, heart and brain. Wolframin protein has an ambiguous physiological understanding, with the main principle being that it affects endoplasmic reticulum and mitochondrial function[1].

Although there is a stable foundation for the understanding of this rare disease and the ability to treat the complications of this disorder, the deficient awareness among healthcare professionals, can often lead to devastating affects among the target population.

Most commonly, the condition first presents with diabetes mellitus in early childhood. However, from my analyses of the case studies I have been fortunate to explore, the diagnoses from this point forward is poor and involves multiple consultations with a variety of specialists and a dispute amongst patient and carers with healthcare professionals.

The most imperative message from this case study is, that our patients are experts in their condition and often can guide us to a diagnosis. 1 in 17 people will experience a rare disease at some point in their lives, which indicates the likelihood of our interaction with such patient groups at some point in our career. Approximately 4 in every 10 rare disease patients have expressed that they have found it hard to get a correct diagnosis and also have raised issues on the coordination of care after a diagnosis[2].
It is important to approach the unknown with sensitivity, research and most importantly patient involvement. While we can use our previous education to treat complications of this rare disease, we must seek the guidance of colleagues, that are experts in the field, so that we make decisions in the best interest of the patient and not based on generalised treatment. Step outside the comfort of your regimented patient care because your judgement can often be clouded by your knowledge.

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References:

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