

# The Journey of a Rare Disease Patient

Entry for the Find A Cure Student Voice Essay Prize: Question 2.

## Case 1

An unconscious 36-year old soldier presents to an American military hospital 70km away from his base. He is normally fit and well but has experienced diarrhoea and vomiting for the past week and has lost 7kg since his last medical check-up.

## Case 2

A patient presents to their GP with a fever, tachycardia and feeling of general malaise. They had been bitten by insect 2 days prior. They deteriorate quickly and are admitted to hospital. A diagnosis of sepsis is made.

## Case 3

A man with a recent diagnosis of Raynaud's disease suffers an injury whilst on holiday in Spain. He experiences a sore shoulder, tightness in his chest and difficulty breathing. He is rushed to hospital and requires analgesia and oxygen.

If these three cases strike fear into your heart, then you are not alone, and you are probably a fellow medical student. They are the classic openers to the multiple-choice exam questions that we are all so familiar with. You were probably running through potential problems and management plans as you read them, organising differential diagnoses by likelihood and thinking about which investigations you would like to order. But what if I told you that the patient in each of these cases was the same person, a 40-year old man called D. The question of diagnosis suddenly becomes a lot more difficult.

## D

I was put into contact with D as part of the Student Voice Prize Patient Pairing scheme. I knew that D was a patient with a rare disease and not much else. We exchanged a few pleasantries over email and set a date for a Teams meeting. Plagued with the fear of awkward silences, I drafted a long list of questions to ask in the meeting. It was very lucky for both of us that D decided to ask the first question:

*“So, how much do you know about rare diseases?”*

With a slight sense of shame, I admitted that I didn't know much. I had become victim to the trap that many medical students fall into, of seeing a low prevalence statistic for a particular disease and concluding that it was very unlikely that I would see anyone with that condition in the future, proceeding to focus my revision on the illnesses that I considered to be more common.

D proceeded to explain to me that while only 5 people per million had the same condition as him, 1 in 17 people will be affected by a rare disease at some point in their lifetime (Rare Disease UK, 2020).

My initial reactions to this fact were shock and then a sense of guilt. I entered Medical School to become a doctor, not just to pass exams. Was I letting down my future patients by not knowing more about rare diseases?

D's experience of healthcare as rare disease patient is not exactly typical but then again, he isn't a very typical individual. D spent his 20's and early 30s travelling around some of the most dangerous places in the world as an officer in the Royal Engineers. He enjoyed keeping fit, was rarely unwell and came 2<sup>nd</sup> or 3<sup>rd</sup> when his unit of 100 soldiers went for squadded runs. His health started to deteriorate in his mid 30s, he started falling behind in the runs and was losing a lot of weight. All of the medical emergencies described in the cases above occurred within the next few years. Each time D became acutely unwell, he was told by healthcare workers that his bloods seemed “funny”, but this was never investigated further.

D became increasingly frustrated, as he knew that he was not well, but continued to push himself in work and at home. It was D's civilian GP that eventually solved the mystery of his health problems. She looked at his 6-inch thick medical notes and said: "we're missing something here". She decided to carry out a short synACTHen test, which looks at how well the adrenal glands respond to hormonal stimulation. The test showed that D's adrenal response was inadequate, finally giving him a diagnosis.

## Addison's Disease

Addison's disease is a rare endocrine disorder in which the adrenal glands no longer produce enough adrenocortical hormones, including cortisol, aldosterone and androgens. Any process which results in destruction of the adrenal glands can result in primary adrenal insufficiency (Addison's Disease), but it is most commonly due to autoimmune destruction (Mitchell and Pearce, 2012). Addison's disease usually has a gradual onset of non-specific symptoms such as fatigue, generalized weakness, weight loss, nausea, vomiting, abdominal pain, skin hyperpigmentation, dizziness, tachycardia and postural hypertension.

In many cases, a diagnosis is only made when the patient has presented with an acute adrenal crisis (hypotension, hyponatraemia, hyperkalaemia and hypoglycaemia) precipitated by a stressful illness or triggering factors, such as infection, trauma, surgery, vomiting and diarrhoea. Upon reflection, D thinks that he experienced an adrenal crisis each time he became acutely unwell, although this was never confirmed.

Addison's disease is treatable with present-day medicines but delays in diagnosis have led to preventable deaths from adrenal crisis. D recalled that in the past three years he had heard word of three men under the age of thirty-five dying from adrenal crises in the UK. Two of them passed away before a diagnosis of Addison's disease was made. Indeed, researchers estimate more than 50% of patients presenting with adrenal crisis don't have a prior diagnosis of Addison's Disease (Elshimy et al. 2020).

Unfortunately, there are still a number of difficulties that exist for clinicians in making a rare disease diagnosis. These can include their ability to recognise the symptoms of a rare

disease, especially when those symptoms are non-specific such as those experienced by D. The training on rare disease in Medical School is often limited and medical students, like myself, may focus on areas of the course that they believe to be higher yield. In practice, developing knowledge specific to a particular rare condition is challenging, given the small number of people with the disease. Further support is required by healthcare professionals including education and training and investigation into the implementation of technological aids to diagnosis such as diagnostic decision support systems (DDSSs) (Ronicke et al. 2019).

After finally being diagnosed with Addison's Disease D was discharged from the military on medical grounds. The diagnosis that he had been searching for spelled the end of the career that he loved. He was left, understandably, feeling a bit lost and unsure of what the future would hold.

## After Diagnosis

Upon first speaking to D, I expressed an interest to learn more about Addison's Disease and he pointed me towards the website of the Addison's Self-Help Group. Initially I was sceptical about what I could learn from a support group website, but I couldn't have been more wrong. The site offers a myriad of useful information, aimed at clinicians, patients and their families (ADSHG, 2020).

Through conversation with D, it is clear that the Addison's Self-Help Group has played a massive role in supporting him to learn more about and manage his disease. Speaking to others on online forums and at in-person meetings has given him the chance to share experiences of Addison's with others and both receive and provide support. Reading articles on the treatment of Addison's Disease and prevention of Adrenal Crisis have allowed him to gain knowledge that backs up his lived experience and helps him to coordinate and manage his care.

A review by Walton et al. looked at what coordinated care meant in the context of rare conditions. They proposed the following definition:

*“Coordinated care involves working together across multiple components and processes of care to enable everyone involved in a patient’s care, to avoid duplication and achieve shared outcomes, throughout a person’s whole life, across all parts of the health and care system...”*  
(Walton et al. 2020).

Patient organisations such as the Addison’s Disease Self Help Group are key in coordination. Prior to speaking to D, I was of the misconception that patient organisations were (as the name suggests) for patients and were only of limited value to clinicians. Now I see that a group of patients working together is extremely powerful, as patient experience is central to the coordination of the diagnosis and management of rare disease.

Transitioning forward to the present day, D has been contributing to the organisation of the Addison’s Self-Help group and enjoys the feeling of “giving something back” to those that are newly diagnosed. Post diagnosis, he continued to work in defence in some of the most dangerous places in the world, carrying 3 months of medication at a time and making sure his colleagues knew what to do in an adrenal crisis. Speaking about his condition now, D says:

*“Some people talk of it [Addison’s Disease] as a disability. I don’t see it as that. It is a limitation you have to manage.”*

While this fundamentally illustrates how admirable D’s attitude towards his disease is, I think it also makes more widely applicable point. We, as the medical professionals of the future, have the ability to give rare disease patients like D the confidence to manage their conditions and change their own perceptions of illness. It is easy to fall into the trap of telling patients what not to do post diagnosis, especially in rare conditions where we may be less familiar with the disease. Instead I believe that it is important to work with patients, their families and other members of a multidisciplinary team to find solutions that allow them to live their life with as little interference from rare disease as possible.

## References

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