Discuss how experiences of a patient with a rare disease may differ from those of a patient with a common disease

“Have you ever paid or been paid for sex”
“No, what’s the going rate? Sorry, I’m joking, no”

I’m nervous, I always make inappropriate jokes when I’m nervous. And today I’m really nervous. I’m in a sexual health clinic in London and I’ve just asked for an HIV test. My boyfriend of one year is in the waiting room awaiting the call of his name and I can tell he wants the test to be positive. He wants an explanation for his symptoms and is desperate for any diagnosis.

I’m hoping it’s not HIV. I tell the nurse that we’ve only ever been sexually active with each other and he looks puzzled but assures me that there are other transmission methods and it’s always good to be safe.

‘It can’t be positive, it can’t be positive, it can’t be positive’ I keep murmuring under my breath.

It was negative.
When I returned to the waiting room, Doug shook his head, also negative- disappointment clear on his face.

It was in that moment that I got a taste of the desperation that comes with living with an undiagnosed rare disease. On the outside we looked just like any other couple walking down the street, but inside, Doug was fearful in a way he had never been. Weeks before he had been a physically fit, cycling enthusiast excelling at medical school and now he was spending hours of the day trawling the internet for diagnoses that matched his symptoms. We would rerun the events in the lead up to sudden onset of symptoms trying to find any clue to their cause.

It was a Saturday, and Doug had woken up with a pain in his forehead that radiated across to one side, a ringing in his ear and pixelated vision, like static on a television screen. He was, as one would expect, highly anxious and felt tired, so slept most of the day. Come the end of the day, the symptoms have not resolved and we headed into Accident and Emergency. He was found to have the usual neurological responses, was given water to drink, and after a few hours we were discharged. We headed to the GP on the Monday, and the doctor suggested that it was likely to be an inner ear infection, known as labyrinthitis, and that with rest, the symptoms should subside in a few weeks. There was no such improvement during this time and further sessions with the GP proved simply to agitate the doctor and increase Doug’s anxiety.

It became quickly apparent that strange combinations of qualitative symptoms, such as those experienced by Doug, were not going to make for an easy diagnosis. Though I am only 2 months into my first year of medical school, it is already clear that the old adage ‘common things are common’ has come to form the foundation of medicine.

Of course, the rationale behind this is to some degree understandable, it became apparent in meetings with the GP that an open mindedness and curiosity is vital in producing doctors who are capable of diagnosing rare diseases. Increasingly as appointments were made, it became clear that there was an overarching sense of frustration from the doctor as Doug attempted to explain his symptoms as clearly as possible. Indeed, before and after consultations with the GP Doug felt embarrassed and uncomfortable, like he was wasting the doctor's time and I can imagine this to be a common experience of someone with a rare disease. The life of a GP in central London is busy, and this was made quite clear to us in our consultations, the doctor came across irritable and disinterested which left us to our own research- hence the visit to the HIV clinic after reading of similar associated symptoms, which will go down as the single most surreal moment of my life to date.

Our online research did lead us to get involved with an online forum of people suffering from similar symptoms. Though the types of people affected varied, they all shared similarly disappointing experiences of doctors who disregarded their symptoms, preferring to explain them as a response to stress. These forums were an interesting place to compare and contrast symptoms, with people reporting changes with the time of day, lighting, diet, and mood; they offered a platform where people who had suffered for a long time could inform others who only recently had experienced symptoms, of what the future would likely look like. This online forum meant that there was a community of people who could both comfort and educate one another. It became apparent that the 'pixelated television like static' Doug had been trying to describe in his talks with the doctor was known as 'visual snow' to the network. Very little is known of the condition, indeed, though Doug presented with his first symptoms in February 2013, the first major trial into visual snow was only published in May this year by CJ Schankin in association with the American Headache Society.

This highlights further the differences between patients with a rare disease when compared to ones with a common disease- the support networks available to those with common debilitating diseases are often mainstream and well publicized. In the case of rare diseases, these services are sparse, to match the general lack of understanding of the diseases, treatment and low patient numbers. This has a negative knock on effect whereby the small number of patients affected struggle to find a medical professional contact interested in conducting research and so studies are often small scale, as in the case of the May 2014 study where only 120 patients were enrolled.

Further, my experience made clear the stigma associated with certain rare diseases; when Doug described his visual disturbance he was met with raised eyebrows and inundated with questions regarding illicit drug taking. Despite having never experimented with recreational drugs there was always an air of suspicion. Sadly, this stigma stretched outside of the doctor's office, the chronic fatigue and issues concentrating when reading Doug experienced were put down to apathy by some family and friends. Over time, he became progressively more reclusive and was prescribed anti-depressants, all the while having had no official diagnosis by a

medical professional. The GP was unsure which medical department to get in contact with and a combination of disinterest, lack of funding from the practice meant that the focus was on monitoring Doug's anxiety, as diagnosis and more importantly treatment of the symptoms became even less of a priority.

It was around this time that the online forums on visual snow yielded accounts of people who had tried including or eliminating certain food groups and reported an improvement in visual symptoms. With no medical guidance, Doug embarked on a strict, protein based diet with an emphasis on eliminating all sugars-a diagnosis of 'toxic mould syndrome' was discussed on the forum and suggestions that through sugar elimination one could 'starve' the mould in the brain. After 6 months trialing this diet Doug was 15kg lighter and reported no notable improvement in his 'visual snow' but rather a substantial reduction in energy levels. Resolved to likelihood that his sight would not return to normal, his diet returned to normal and even to this day, Doug has not received a diagnosis for his symptoms but after taking some time off university he has returned to medical school.

Through my experience of looking after someone with a rare disease and attending doctors appointments I have painted a negative picture of the experiences of rare disease patients. With this in mind, alongside Doug and some friends, we are in the process of setting up a Rare Disease Society at the university. Talks will be held by doctors in the field who dedicate their lives to spotting the nuances and symptom oddities that lead to the diagnosis of rare diseases. Such meetings remind medical students that whilst 'common things may be common' it is essential that we keep our minds open and work alongside patients, to improve the medical world of tomorrow.