I am number 17
David's experience

I have Occipital Horn Syndrome. I call it OHS, it's ultra-rare and I'm the only person in the UK living with it. When I was younger, I was loosely diagnosed with Ehlers-Danlos syndrome (EDS) as a child at Great Ormond Street Hospital (GOSH). At the time they were more focused on treating symptoms rather than identifying the diagnosis, and I had tonnes of surgeries, from a year and a half old until I turned 18.

For me, the actual diagnosis didn't really mean anything. It wasn't until I was 24-26 when I had genetic testing that I received my diagnosis. Even now, I never really google stuff about my disease because I didn't see the value.

When I started having my genetic appointments, I didn't realise the knock-on effects my disease would have - on having children, passing it on to them. When you're 10 years old it's not in your thought process. It's only when you're older that's you realise it's an issue, including life expectancy.

If you've grown up with your disease from day one, you never know what it's like to be healthy. I've never lived a day without pain. The disease affects every aspect of my life. Musculoskeletal impact is a massive thing. When I was growing up, I didn't start puberty until I was 15, and had to be induced with hormones. Looking back now, this was something that was probably linked to my rare disease. Joint pain also has a big impact and from my early 20s has been more severe – anything from typing to using my phone is painful.

Then there is the internal aspect of my disease, which is the more dangerous side of things. I've been self-catheterising since I was 7, so that's 23 years now. I had my kidney removed 8 or 9 years ago, when I was at university as the function declined. I always have bladder or kidney infection and am resistant to pretty much all antibiotics. The older I get, the more of a problem this is. When the infection gets bad enough, I will ultimately have to go to hospital and be put on an IV drip. Now once I start getting infections, I'll have to get IV drips and dialysis every time, which makes me feel out of control.

I had a breakdown when I was 22 or 23, after university, and had to quit my job. I had a great job, that I always wanted to do, and I had to leave because my rare disease meant I wasn't able to do it. I had a mental health breakdown and at the time most of my friends were successful and I suppose I knew deep down that would never be me. I'll be able to achieve some of those milestones, but I'll be 10 years later because it's hard to work with a rare disease. I've had to change my job so many times.

With regards to people with rare diseases, it feels like everyone likes to talk about us, but no one really wants to help us. Lots of companies have this blue tick scheme which means they're disability friendly, I think it's a tick boxing, token exercise. When you have a rare disease, it affects you differently day-to-day. People with rare diseases get pushed aside out of work, into a rare disease bubble.

The media is another issue. If some outlets wrote an article about me, they would write an atrocious headline. So even if rare disease is in mainstream media, it's very rare to be a good article. The main problem in rare disease in literature is the hyperbolic literature and over the top language. There are definitely opportunities for rare disease patients to educate the media.

Career wise, GOSH has got me to where I am today. They were the steppingstone for me to realise I needed to speak out and I learnt the public speaking. They were also the steppingstone to working in rare disease. I then started doing some talks in Cambridge around rare disease and now I work with Rare Revolution, initially as a social media assistant and then moved to sales and business development, and also speaking at lots of events.

I am always aware of being ill. Before my breakdown I had the realisation that my illness is always there and not going away. It's going to get worse and worse, it's too late for gene therapy, and there's no knowledge about it. I take pain killers and they don't do anything. My life expectancy is 40-50, I'm not going to be 90 in a Zimmer frame.

People don’t understand how rare my disease actually is. Initially I found it funny, being the only one in the country with my rare disease, but now I’m more exposed to rare diseases, it makes me realise that no one will ever help me.

I see every medical department you can think of, 13 metabolic medicine people are in one room examining me. Lots of people don’t want medical students in the room, but in my case, it’s the only way more junior doctors are going to learn about my condition. If you want long term, chronic treatment you should be educating the younger generation.

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