I am number 17
Sally's experience

X-linked hypophosphatemia (XLH) is a rare bone condition which is caused by a deficiency of the mineral phosphate in the body. This leads to bone and muscle pain and weakness, stiffness, fatigue, and an increased risk of fracture. The symptoms and their severity can vary a lot across people with XLH, but the most common is a bowing of the legs and a difference in the way a person walks because of the pressure of the body on the legs. I was diagnosed at 18 months old when I started to walk, so I've lived with XLH as long as I can remember. Although this is an inherited condition, it is unusual in that it can also occur spontaneously which is what happened in my case.

As we had no family history of XLH, my diagnosis was a confusing and difficult time for my family to deal with in isolation and we were fortunate to get a referral to an expert paediatrician in Manchester where we live, and who supported us and helped me to get the correct treatment as I grew up. However, coming to terms with my diagnosis as I got older was particularly difficult and had an impact on my mental health as a teenager and in my early twenties.

I consider myself lucky that my symptoms are relatively mild, but XLH still affects almost every area of my life. I always have some level of pain and stiffness; however, it can be manageable. To keep the pain and stiffness at a reasonable level, I have to adjust the way that I sit at work, in restaurants, or on public transport.

I can't carry much weight, so I have to think carefully about what I need to take with me and how I organise my bag or luggage, and to plan ahead the level of activity that I'm doing and when I may need to rest. There is always something to be thinking about.

Due to the weakness in my bones, I'm not able to take part in any impact sports like running, so finding the right exercise has always been a struggle for me. Although now I really enjoy walking and Pilates to keep me in shape and to keep my strength up. XLH is progressive, so I need to be mindful of this and how it may get more challenging as I get older.

My condition is treated with a supplement which I take 4 times a day and is another thing which I need to plan around as it has to be taken in a drink, and timed around what and when I eat – and isn't particularly pleasant! I've also had multiple orthopaedic operations on my legs which have all involved taking extended time out of work or study, lengthy and complex recovery, and have also caused some nerve damage, which have limited my activities even further.

It's not always easy to know how to talk to people about XLH, especially as my condition isn't always apparent. Growing up I would only talk about it when I had to, but my life completely changed over 2 years ago, when I met other patients for the first time and realised that there was a community out there and a difference to be made. Since then, I've changed career to work for a rare disease patient support organisation, I volunteer with the UK and International groups for XLH patients and have started to develop my own advocacy for those living with XLH and other rare conditions. While finding a way to talk about such personal experiences in such a public way is still something I'm getting used to, working with the rare disease community is the most wonderful and rewarding thing I could do.

For me, society can have very set ideas of how a 'well' person should look and be, and how a 'sick' or 'disabled' person should look and be, and when you don't fit this expectation you can just be treated as 'weird'. As the symptoms of XLH can be unusual and aren't always consistent depending on the day or what I'm doing, I've found that I'm always on the edge of being different or fitting in. I particularly remember a few years ago being rudely challenged for using the priority seat on the bus and was questioned recently on why I needed to rest my feet on a cushion on the train, all by strangers. These events and witnessing the difference which can be made from opening the conversation about rare and hidden conditions, I want to be more open about my life with XLH. So, that we can raise awareness, understanding, and above all, acceptance.

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