

Q2. Living with a rare disease is a life-long learning experience in which patients become experts in their own disease. Use a case study to demonstrate what future doctors can learn from patients with rare diseases.

4 things I learned from Heather, an individual with fibrous dysplasia

“I will never remember all of this”. I am certain every medical student has had this thought many a time due to the sheer volume of information we are plugged with. When it comes to rare diseases things get even harder; there are currently 6,000-8,000 known rare conditions and each condition affects less than 1 in 2,000 patients. This makes the likelihood of ever coming across many of these conditions extremely low. However, rare diseases collectively affect 1 in 17 people meaning that as future clinicians we need to consider how we can provide high quality care to patients with diseases we are not familiar with (1).

Fibrous dysplasia (FD) is one of those conditions. With an estimated prevalence of 1 in 15000-30000 individuals (2), it is unsurprising that I had never come across it. Even after researching it I don't believe I understood how debilitating it can be until I talked to Heather, an individual with FD, about her experience. It was saddening to hear how FD has severely disrupted her life and disheartening (or at times shocking) to hear about her experiences with healthcare. The chances of me or anyone else coming across a patient with FD may be low but there are many lessons in Heather's story that, although seemingly obvious, are important for all good clinicians to remember.

What is fibrous dysplasia?

FD is a genetic condition that causes fibrous lesions to develop on bone instead of normal mineralised bone development. This makes the bone weaker and thus more likely to become deformed or fracture. The presentation is highly variable as it can affect a single bone or multiple bones and the bones affected differs between patients. Current treatments involve surgery to repair the bone shape and treat or prevent fractures. Bisphosphonates, drugs that reduce loss of bone density in osteoporosis, can be given to strengthen bone and may reduce pain in some patients (3, 4).

Heather's story started at 10 years old after her dad took her to A&E as she had pain whenever she walked. An x-ray revealed a lesion believed at the time to be bone cancer, but after a biopsy and six weeks in hospital she was diagnosed with FD. Her left tibia, femur and hip has been affected by these lesions. Since then she has had 8 surgeries and many trips to hospital for scans and other complications. At 14 she was also diagnosed with Hashimoto's thyroiditis. Her condition has progressively deteriorated over time, to the point where she can no longer work and has difficulty engaging in normal social activities.

Lesson number 1: If your patient says they're in pain, believe them.

This may appear instinctive, however this is the point Heather was most keen to get across to me. Historically, descriptions of FD do not associate the condition with pain, although many publications now acknowledge it as a potential manifestation of the disease (5). Due to this she has previously been told that she 'shouldn't' be experiencing pain, with some going as far as telling her that the pain is not real. She believes that part of the reason people don't understand her pain is because she is not on crutches and looks 'normal'. A quick google search will bring up many stories where the patient felt that their pain was not taken seriously and the detrimental effect this had on the doctor-patient relationship is obvious. Refusing to understand a patient's pain in any scenario will leave them feeling frustrated and ignored, or doubting the doctor's level of care.

For Heather, it seemed that worsening pain was often written off as unimportant or possibly just an unexplained part of the condition. Most notably, her GP once turned her away for back pain after giving her anti-inflammatory drugs despite her insistence that the pain was not normal. She was eventually referred to a back surgeon separately through her rheumatologist where she was found to have a slipped disc. As shocking as I found it to think that her slipped disc was almost left untreated, it made me stop and think about the importance of ruling out any differential diagnoses rather than making assumptions.

It is estimated that 43.5% of people in the UK experience chronic pain, making it crucial that clinicians are receptive to patients claiming to be in pain so that steps can be taken towards attempting to manage it (6). In Heather's case, she is now being treated with painkillers (and occasionally bisphosphonates) for her hip and leg pain which, although not effective for curing the pain, do help. Furthermore, she undertook a pain rehabilitation course two years ago and found the pain psychologists there quite helpful. Unfortunately, she is still in pain every day which restricts her daily life.

Lesson number 2: Sometimes your patient may know more than you.

Many patients, especially those with rare diseases, will undertake a lot of research to understand their condition or find new or emerging treatments. Furthermore, support groups are an important source of knowledge and comfort for many individuals. Heather described how attending Fibrous Dysplasia UK meetings has allowed her and other people with FD to learn about treatments available or how to better manage their condition. This information mostly comes from talking to other patients about their experiences (and occasionally from doctors that come to give presentations). Over time they will become highly knowledgeable about their disease due to their own experiences of living with the condition and the information they have gained from other sources. These patients are often described as “expert patients”. As such it is important for doctors to be willing to learn from their patient and be open to collaborating with them.

Lesson number 3: Be flexible with your treatment plan.

For rare conditions, there is often no clinical pathway to follow as there may not be enough demand or evidence to create one. In these situations clinicians may have to do their own research to understand the disease and create the best treatment plan, or find a more suitable hospital patients can be referred to. In Heather’s case, she has found that doctors that know more about the disease are more likely to listen to her suggestions and take on board what she wants. After her original doctor left at 21, she felt her new doctor had a limited understanding of the condition and was too quick to want to operate. “It doesn’t just go away by doing a metal graft,” she told me. Each patient is individual, a good doctor has the ability consider all the options available and tailor their treatment.

Lesson number 4: The best way to understand a condition is to talk to a patient.

Prior to speaking to Heather, my research had led me to underestimate the effects of FD on an individual’s life. For example, the information on some websites suggest that whilst the condition is very disruptive for children and young adults due to the amount of corrective surgery required, it is less problematic for adults as the bones are no longer growing and lesions will not spread (7). This may be true for some, but due to the variable nature of the disease it is not necessarily the case and some patients may be wheelchair bound (8).

As with all diseases, patients can be affected in different ways but with rare diseases there may not have been enough patient studies to understand these differing manifestations. For

example, Heather described how she feels constantly tired which I had not come across in my initial research. However, upon undertaking further research on fatigue and FD, I discovered a recent paper that found that individuals with FD generally have lower level of energy (9). Despite being a common symptom, this information is clearly not widespread yet. As with all diseases, taking the time out to talk to individuals with the condition will help future doctors expand their understanding beyond that which can be gained through medical books.

Conclusion

Many things can go wrong with the human body and I am aware that, as a future doctors, we will unfortunately never know all 6000-8000 rare diseases. As more research and awareness campaigns go into rare diseases, it will hopefully become easier to access guidance for many conditions. However, in the case that we come across something we are unfamiliar with, it is still important to make the patients feel cared for, do our best to understand their condition and work to maximise their quality of life. Doctors can learn a lot from patients with rare conditions, from information about the condition itself to what can happen in healthcare to patients whose cases are not straightforward. In my case, Heather's story has reminded me of the number one skill I will need to be a good doctor: listening, and I mean really listening, to your patient.

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