FOP: how to spot this rare genetic disease

Helen Bedford-Gay reveals how fibrodysplasia ossificans progressiva (FOP) affected her family and explains how sonographers can help

MY PREGNANCY was uneventful until, at seven months, I had some spotting and was sent for a late ultrasound. Baby was moving, growing and doing well, except our sonographer could not see the big toes – the baby was probably curling them up.

When Oliver, was born, he was perfect, except for his big toes, which were shortened and tucked under, just like the sonographer had said.

After three months, an unusual swelling appeared on the back of Oliver's head. The paediatrician said it was not cancerous but did know the cause. At nine months, Oliver had neurosurgery to remove the swelling.

We had been attending appointments with a paediatric orthopaedic specialist to find ways to 'fix' Oliver's toes. It was clearly visible in the X-rays that he had a bone missing in both of his great toes, but this was not seen as a cause for further exploration.

When we attended a follow-up appointment for Oliver's neurosurgery in 2009, the specialist told us the three words that would ultimately change our lives: fibrodysplasia ossificans progressiva (FOP). The consultant did not seem unduly concerned. He wrote the three words on the top of a piece of paper and tore them off, explaining that as long as Oliver did not play rugby, he would be pretty much OK.

Later that day, we searched FOP on the internet and our world fell apart. We read that FOP was an ultra-rare genetic condition affecting just one in two million people. Muscles, ligaments and soft tissue would turn to bone due to a fault in the ACVR1 gene, restricting the affected person's movement. Over time, the person would become imprisoned in a second skeleton. Life expectancy was around 45 years.

We have been lucky. Oliver has only had one flare-up that we are aware of. When he was two years old, he awoke with a swelling under his neck. After a couple of weeks on steroids, his swelling subsided. An ultrasound scan showed a trace of new bone growth in his jaw muscle, but nothing that would restrict his movement.

Fast forward to 2021 and we are the founders and trustees of FOP Friends, and next year will be our 10th anniversary as a registered charity, recognised nationally and internationally. We have connected the UK's FOP community so people can find the information, friendship and support they need, and we raise funds to support research projects at the University of Oxford and around the world.

What is FOP?

Fibrodysplasia ossificans progressiva is the only disease where one organ (soft tissue) is changed to another (bone). There are around 900 known cases worldwide, 70 in the UK. Most cases are new, although where one parent carries the faulty ACVR1 gene, there is a 50% likelihood of passing it on. FOP is characterised by the congenital malformation of the great toes and progressive heterotopic ossification. People with FOP can also have shortened thumbs.

During the first decade of life, children with FOP experience unexplained swellings, which are painful, and are often misdiagnosed as cancer. When the swellings regress, new bone growth has usually occurred. Where this occurs over a joint or within a muscle, movement is lost.

FOP is triggered by trauma such as a knock, bump, fall, an immunisation, virus or surgery. However, flare-ups can also appear spontaneously without warning. Over time, FOP effectively imprisons the individual in a second skeleton. Attempts to remove the new bone can aggravate the FOP and trigger further and more painful growth.

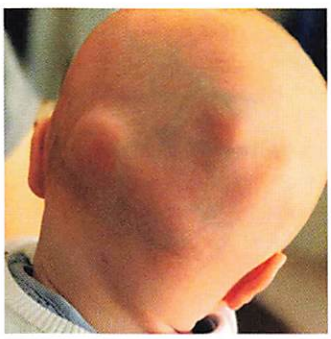
FOP is a variable disease, which means there is no timescale for the progression. There is no known treatment or cure. There is some evidence that a short course of high-dose steroids can help to reduce inflammation, but this is not recommended for all flare-ups and can only be used for a limited time.

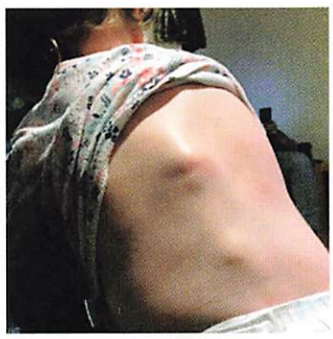
Care for children with FOP is precautionary and preventive. Individuals must avoid soft-tissue injuries, removal of heterotopic bone, contact sports, muscle fatigue and non-essential surgeries.

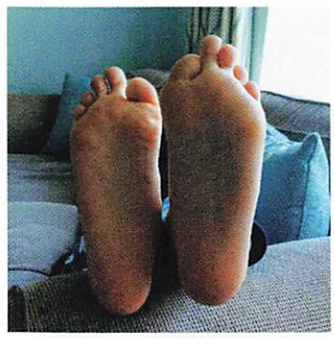
What can a radiographer or sonographer do?

If you see an unusual hallux valgus appearance, malformed big toes and/or unexplained swellings in a fetus or young child, consider FOP as a potential diagnosis. Where FOP is suspected, advice should be sought immediately before any further interventions are carried out. Under no circumstances should a lump be biopsied.

For the latest medical guidance on FOP, including Covid-19 guidance, visit www.fopfriends.com







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