Fibrous dysplasia

This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of fibrous dysplasia and where to get help.

What is fibrous dysplasia?

Fibrous dysplasia is a congenital (present at birth) condition that affects bone growth and development. Instead of maturing into solid bone, affected bones stay at the immature fibrous stage so are weak and misshapen. Any bones can be affected – if only one bone is affected this is referred to as monostotic but if more than one is affected it is referred to as polyostotic.

What causes fibrous dysplasia?

Fibrous dysplasia is a genetic condition, caused by a mutation (change) on a specific gene. This mutation develops sporadically (out of the blue) very early in pregnancy so is not inherited from parents or passed on through the family. The gene affected has been identified as the GNAS gene which affects how certain cells in the body – including bone cells – grow, divide and die.

Fibrous dysplasia can occur on its own or as part of a syndrome (collection of symptoms often seen together) called McCune-Albright syndrome. Children with McCune-Albright syndrome also have problems with their endocrine (hormone) system.

What are the signs and symptoms of fibrous dysplasia?

The first sign of fibrous dysplasia is often a swelling in the jaw and gaps developing between the teeth. This may be uncomfortable and cause problems with eating. If the bones in the skull are affected, this can make them misshapen affecting the overlying tissue. Rarely, the abnormal bone growth can affect hearing if the ear canal narrows or vision if the optic nerve is compressed. If the long bones are affected, they will be weaker than usual so more likely to fracture. Over time, this may lead to unequal limb length.

The formation of weak fibrous bone tissue continues during childhood and adolescence until growth is complete. The body then slows producing fibrous tissue and the affected areas do not 'spread' to other parts of the body.

How is fibrous dysplasia diagnosed?

Fibrous dysplasia may also be diagnosed by chance following x-ray for a suspected fracture. Jaw pain and problems eating may also suggest a diagnosis of fibrous dysplasia. Doctors will take a clinical history of when and where the symptoms appeared and their severity, as well as a physical examination. Imaging scans, such as x-ray, CT or MRI may be suggested to monitor bone growth before, during and after treatment. Occasionally, a bone isotope scan may be needed to identify the affected areas of the body. A bone biopsy – small sample of bone tissue – may be taken for examination in a laboratory.

How is fibrous dysplasia treated?

As fibrous dysplasia can affect various areas of the body, treatment is best delivered at a specialist centre where a multidisciplinary team approach can be taken. The multidisciplinary team will usually comprise craniofacial (skull and face) surgeons, neurosurgeons, ear, nose and throat (ENT) surgeons, ophthalmologists (eye specialists), audiologists (hearing specialists), dentists and orthodontists, with other specialists brought in as needed.

Initially, the symptoms of the abnormal bone growth will be treated, often with medications to strengthen the bone. Fractures will be treated with a plaster cast.

Surgery to remove the fibrous tissue is difficult due to the thickness of the bone and is not always successful. If bone growth is affecting function, particularly vision, surgery may be carried out earlier in childhood. Any areas where a large area of fibrous bone has been removed will need to be replaced with bone grafts from elsewhere in the body. If the skull is affected, surgery to re-shape the skull may take place in several stages. Support to improve any hearing or visual impairments will also be needed.

As the bone continue to grow during childhood and adolescence, further surgery may be needed to make minor corrections to bone shape.

What happens next?

Once growth is complete in adolescence, no further fibrous bone tissue develops and affected areas do not 'spread'. The outlook for children with fibrous dysplasia is good but regular monitoring and follow up, particularly of vision will be needed throughout childhood and adolescence into adulthood. The majority of children grow up to lead a normal life, working and raising a family. Some children and families benefit from psychological input at various stages throughout childhood and adolescence.

Further information and support

Headlines – the Craniofacial Support Group – is the main support organisation in the UK for families of children and young people affected by a craniofacial disorder. Visit their website at www.headlines.org.uk.

Changing Faces is another organisation that will be able to offer help and support to anyone living with a condition that affects their appearance. Visit their website at www.changingfaces.org.uk or telephone their helpline on 0845 4500 275.

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