



Cranio-fronto-nasal dysplasia

This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of cranio-fronto-nasal dysplasia (also known as cranio-fronto-nasal dysostosis) and where to get help.

What is cranio-fronto-nasal dysplasia?

Cranio-fronto-nasal dysplasia is a type of craniosynostosis. The name describes the parts of the skull and face affected.

The skull is made up of several 'plates' of bone which, when we are born, are not tightly joined together. The seams where the plates join are called 'sutures'.

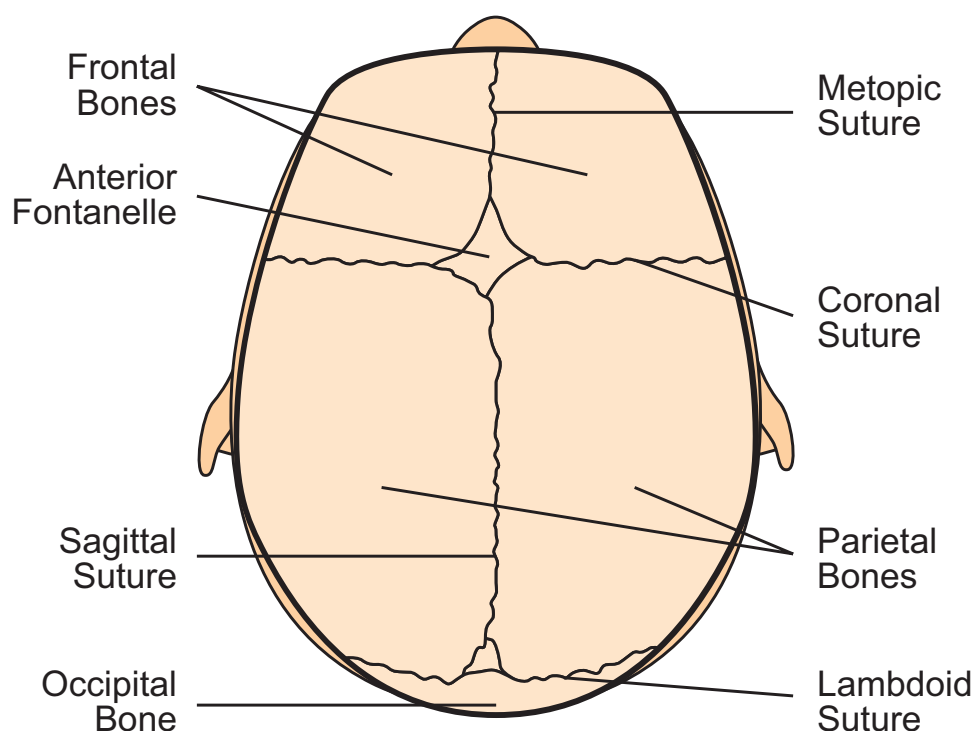
As we grow older, the sutures gradually fuse (stick) together, usually after all head growth has finished. When a child has craniosynostosis, the sutures fuse before birth.

It can affect one suture or several.

When more than one suture is affected, it is called 'complex craniosynostosis'. This may happen as part of a syndrome (collection of symptoms often seen together), and so may be referred to as 'syndromic' as well.

In cranio-fronto-nasal dysplasia, usually one or both of the coronal sutures fuses before birth, so the skull has a flattened appearance. The eyes tend to be widely spaced and there may be a bifid (split) nose.

Normal Skull of the Newborn





What causes cranio-fronto-nasal dysplasia?

Cranio-fronto-nasal dysplasia is a genetic condition, caused by a mutation (change) on a specific gene. Research has identified the affected gene as the EFNB1 or EphrinB1 gene on the X chromosome. X-linked disorders usually affect only boys but the opposite is true in cranio-fronto-nasal dysplasia, where it occurs in both sexes and girls are more severely affected than boys. The information sheet on cranio-fronto-nasal dysplasia from Headlines (details at the end) explains how and why this happens.

What are the signs and symptoms of cranio-fronto-nasal dysplasia?

Girls are more severely affected than boys and the main symptoms are early fusing of one coronal suture and widely spaced eyes which may develop a squint. Girls also tend to have distinctive hair and may have mild changes to their hands and feet with curved fingers and mild webbing (syndactyly). Their shoulders slope downwards and their neck may also appear webbed. Less commonly, girls may be born with a condition called diaphragmatic hernia, which will need treatment soon after birth. They may also have mild learning disabilities or developmental delay, a cleft lip and duplicated thumbs and big toes.

Boys with cranio-fronto-nasal dysplasia may only have widely spaced eyes and no other symptoms.

How is cranio-fronto-nasal dysplasia diagnosed?

As girls with cranio-fronto-nasal dysplasia have a characteristic appearance, no specific diagnostic tests are needed. Imaging scans, such as x-ray, CT or MRI may be suggested to monitor bone growth before, during and after treatment.

Boys may not be diagnosed with cranio-fronto-nasal dysplasia as the widely spaced eyes rarely cause problems. Diagnosis may only be suggested if a girl is born with cranio-fronto-nasal dysplasia in the family.

How is cranio-fronto-nasal dysplasia treated?

Boys rarely need any treatment for cranio-fronto-nasal dysplasia.

As cranio-fronto-nasal dysplasia in girls 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, neuro (brain) surgeons, ear, nose and throat (ENT) surgeons, ophthalmologists (eye specialists), audiologists (hearing specialists), dentists and orthodontists, geneticists, psychologists and speech and language therapists with other specialists brought in as needed.

In many cases, initial skull re-shaping surgery takes place within the first few years of life. This will involve cutting through the fused suture in the skull and re-shaping it to give a more normal skull shape. The bifid nose may also be corrected at the same time – this involves splitting the skull down the middle and reshaping the centre portion so that the nose and other midline structures are more even.

There are several methods of treating a squint if one is present. The aim of all methods of treatment is to align the eyes so that they look normal and work properly. Non-surgical methods like glasses or patches are tried first.



Only if these methods do not correct the squint is an operation considered.

Diaphragmatic hernias are repaired in an operation under general anaesthetic. If the thumbs or big toes are duplicated, the additional digit will be removed in an operation.

What happens next?

The outlook for children with cranio-fronto-nasal dysplasia is good with the vast majority growing up to lead a normal life, working and raising a family. Children are of normal intelligence so usually do well at school, college and university. They will require long term monitoring, particularly during period of growth in childhood and adolescence, but surgery tends to be completed by the time the child is in their mid-twenties. 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.