

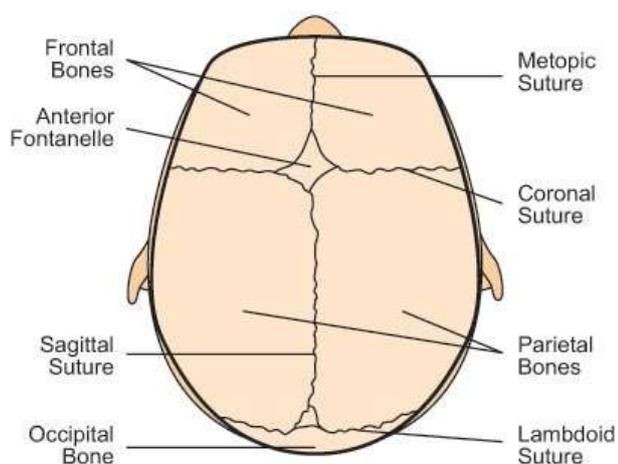


# Apert syndrome: information for families

Apert syndrome is a type of complex craniosynostosis named after the doctor who first described it in the early 20th century. As well as the skull and face, the hands and feet are also affected. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of Apert syndrome.

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'.

Normal Skull of the Newborn



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 Apert syndrome, both coronal sutures fuse before birth, leading to the skull being short from front to back but wide from side to side. Other sutures may also be affected either from birth or later.

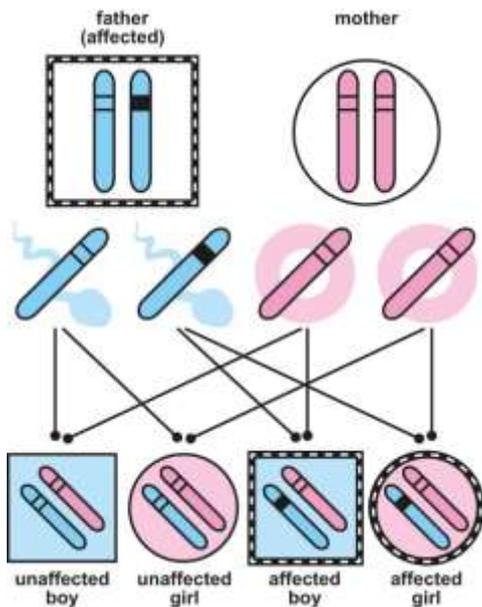
The facial bones are also affected, as the cheekbones and upper jaw do not grow in proportion to the rest of the skull. The bones around the eyes (orbits) are wider spaced and shallower than usual, causing the eyes to bulge outwards.

As well as the skull and face, the hands and feet are also affected in Apert syndrome with the fingers and toes joined or webbed (syndactyly).

## What causes Apert syndrome?

Apert syndrome is a genetic condition, caused by a mutation (change) on a specific gene. Research has identified the affected gene as the Fibroblast Growth Factor Receptor 2 (FGFR2) gene. This affects how certain cells in the body – including bone cells – grow, divide and die.

The gene mutation can be passed on from parent to child but in many cases develops sporadically (out of the blue). If it is inherited, it is passed on in an autosomal dominant manner – this means that if one parent is affected half of their children will inherit the condition.



## What are the symptoms of Apert syndrome?

Children with Apert syndrome have a characteristic appearance due to the problems with the skull plates fusing and midface bones not growing in proportion. If the skull plate fusion is severe, pressure can build up inside the brain (intracranial pressure) which will require urgent treatment.

There is a risk of developing hydrocephalus. Hydrocephalus occurs when the cerebrospinal fluid (CSF) is stopped from circulating or being re-absorbed. The CSF builds up within the ventricles (cavities) of the brain resulting in increased pressure on the brain.

Rarely, the bones of the spine in the neck area (cervical spine) can also be affected, causing a condition called Chiari malformation, where the base of the brain is squeezed.

Failure of the midface bones to grow can affect breathing as the airway is narrow. A small number of children also have heart problems, which will require regular life-long monitoring.

A cleft palate may be present. Vision may also be affected as the eyes are not protected by the orbits and eyelids. Most children with Apert

syndrome have learning disabilities or developmental delay, which varies from mild to severe. Syndactyly or fused fingers and toes is also a symptom of Apert syndrome.

## How is Apert syndrome diagnosed?

As children with Apert syndrome 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 and to check for hydrocephalus and raised intracranial pressure.

## How is Apert syndrome treated?

As Apert syndrome 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.

Depending on the severity of the skull fusion, treatment soon after birth may be needed if pressure inside the head is raised, breathing problems are severe or there is a risk of eye damage. Children will be monitored regularly so that any problems are identified quickly so that treatment can be offered promptly.

In many cases, initial skull re-shaping surgery takes place within the first few years of life. This will involve cutting through the fused sutures in the skull and re-shaping them to give a more normal skull shape.

Surgery to separate the fused fingers is carried out in several phases, often with separation of the index and middle finger carried out first, followed

by further operations to separate the other fingers. The fused toes may not require surgery unless they are causing problems with mobility.

Further surgery to improve the midface problems will usually be carried out in late childhood, most commonly using a rigid external distraction (RED) frame to gradually pull the affected bones forward over a period of weeks and months. The eye sockets will also be re-shaped so that the eyes sit more deeply inside the skull so that the eyelids can close fully to protect the eyes.

Hydrocephalus will need a shunt operation to divert the obstructed CSF to the abdomen. Orthodontic treatment using braces will be suggested to improve overcrowding and speech.

As the bone continues to grow during childhood and adolescence, more surgery may be needed to make further corrections to the skull shape and midface area.

## **What is the outlook for children and young people with Apert syndrome?**

The outlook for children born with Apert syndrome is variable depending on the severity of their symptoms and the impact it has on such functions such as breathing, vision and hearing.

Children with Apert syndrome often have learning disabilities, although the severity of these is variable. Most will benefit from support in education and day to day life although a degree of independence may be possible.

They 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 late teens to early 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](http://www.headlines.org.uk)

Changing Faces is another organisation that offers help and support to anyone living with a condition that affects their appearance. Visit their website at [www.changingfaces.org.uk](http://www.changingfaces.org.uk) or telephone their helpline on 0845 4500 275.