Headlines
Craniofacial Support

APERT SYNDROME

An introductory guide for parents and carers

If your child has just received a diagnosis of Apert syndrome, you may be feeling a bit overwhelmed. There can be a lot of information to take in, particularly at a time when you may only just be getting used to having a new baby. This leaflet is intended to help you understand your child's condition and the treatment that you may be offered.

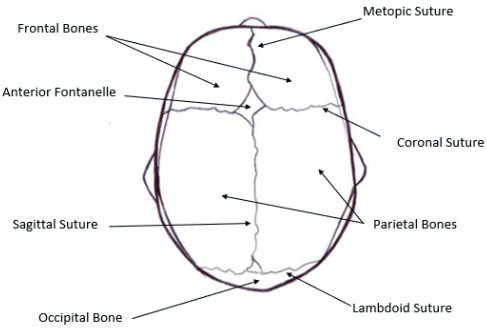
Apert syndrome is a very rare type of complex craniosynostosis named after the French doctor, Eugene Apert, who first described it in the early 20th century. He recognised a specific number of differences with children and realised there was a link with these differences. It is thought to occur in around 1 in 65,000 live births.

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 too early. 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) which is why it may it is also referred to as 'syndromic'.

Normal Skull of the Newborn



In Apert syndrome,several of the cranial sutures may be fused before birth, or none. Sometimes, some of the sutures may not be fused at birth, but begin to fuse as the child gets older.

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 differences with the skull and face, the condition also affects fingers and toes, which may be joined or webbed - a condition known as '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 and the craniofacial sutures – grow, divide and die.

In most cases of Apert syndrome, it occurs sporadically - that is, there is no family history of Apert syndrome. The gene mutation can be passed on from parent to child, but in many cases, develops out of the blue. If it is inherited from an affected parent, then there is a 50% risk of any children also being affected.

What are the symptoms of Apert syndrome?

Children born with Apert syndrome have a characteristic appearance because of the way the skull and facial bones have been formed. The fusion of the bones in the hands and feet also make the fingers and toes look different.

Failure of the midface bones to grow can affect breathing as the airway is narrow and a cleft palate may be present. A small number of children also have heart problems, which will require regular monitoring as they grow up.

Children with Apert syndrome are likely to experience some early learning delay. Some will catch up, but for others there may be longer term learning disabilities or developmental delay which can be mild, moderate or severe - each case will be different.

Treatment and surgery

Because Apert syndrome is such a rare condition, treatment is best delivered at a specialist centre where a multidisciplinary team approach can be taken.

There are 4 Designated Highly Specialised Craniofacial Units in England and 1 in Scotland - further details of these services and how to contact them are available on the Headlines website.

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.

Once you have been referred, your baby will undergo a number of investigative tests. These may include MRI and CT scans and a review by a clinical geneticist.

Treatment will be individualised according to how Apert syndrome has affected your baby: it may involve surgery on the skull early on in life, or the treatment of any breathing or eye concerns. All children will be monitored regularly to ensure that problems are identified quickly and treatment offered promptly.

In many cases, initial skull re-shaping surgery takes place within the first few months or years of life. This is to ensure that the pressure inside the skull stays within normal limits and 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. Fused toes may not require surgery, unless they are causing problems with mobility.

Orthodontic treatment using braces will usually 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 does the future hold for my child?

Children born with Apert syndrome in the UK today benefit from an array of recent advances in surgical techniques and treatments. The outlook for individual children depends on the severity of their symptoms and the impact the condition has on functions such as breathing, vision and hearing.

Children with Apert syndrome often have a degree of learning disabilities, although the severity of these varies. Although most will benefit from support in education and day to day life, many will go on to enjoy a degree of independence as they reach adulthood. They will also require long term monitoring, particularly during periods of growth in childhood and adolescence, but surgery tends to be completed by the time children reach their late teens to early twenties.

Some children and families benefit from psychological support at various stages throughout childhood and adolescence, and into adulthood.

Each of the specialist centres have designated psychologists to offer support and guidance.

Headlines produces a number of other leaflets covering different aspects of craniosynostosis and treatment. Please email helpline@headlines.org.uk for further information.

Headlines is the only national charity supporting those affected by craniosynostosis and rare craniofacial conditions. As well as providing information for parents, carers and families, we also offer a confidential helpline, a member magazine and regular newsletters, conference & information days, opportunities for members to meet, and an Annual Family Weekend.

Join us!

Membership is free.
Visit www.headlines.org.uk/membership.asp to join.

Find us on social media:


@headlinescraniofacialsupport


@HeadlinesCranio


@headlinescraniofacial


www.headlines.org.uk

Headlines is a registered charity in England & Wales (no. 1058461) and Scotland (no. SCO50262)

Headlines
Craniofacial Support