Great Ormond Street Hospital for Children NHS Foundation Trust: Information for Families

Saethre-Chotzen syndrome

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

What is Saethre-Chotzen syndrome?

Saethre-Chotzen syndrome is a type of complex craniosynostosis named after the two doctors who described it in the mid-20th century.

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'.

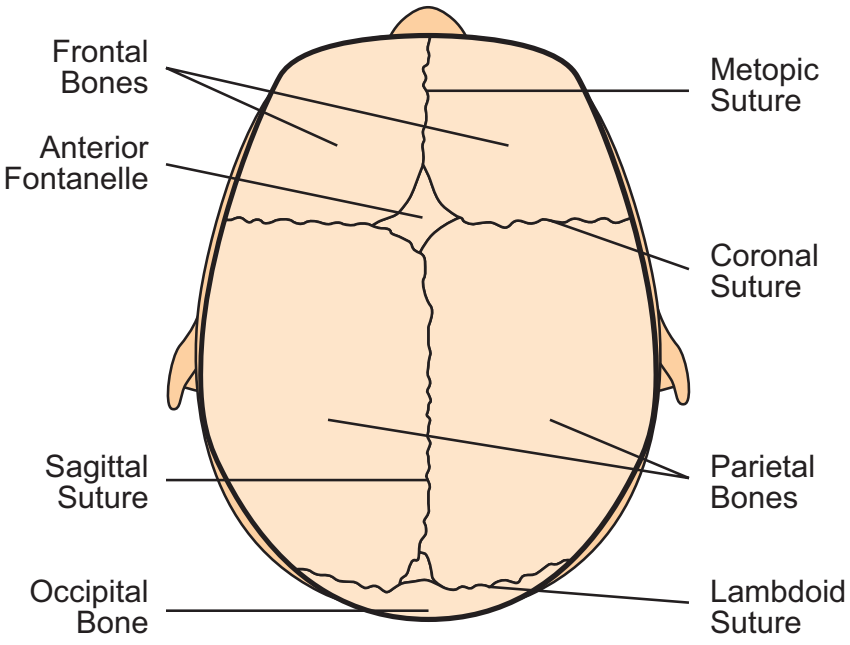
As we grow older, the sutures gradually fuse (stick) together, usually after all head growth has finished. When a child has craniosynostosis, the sutures fuse before birth. It can affect one suture or several.

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 Saethre-Chotzen syndrome, either one or both of the coronal sutures fuse before birth, leading to the skull being short from front to back but wide from side to side. If the coronal suture on only one side is fused, the skull will have a lop-sided appearance. As well as the skull, the hands and feet can also be affected, although rarely to such a degree that surgical treatment is needed. Another common feature is a drooping of one or both upper eyelids (ptosis).

Normal Skull of the Newborn



What causes Saethre-Chotzen syndrome?

Saethre-Chotzen syndrome is a genetic condition, caused by a mutation (change) on a specific gene. Research has identified the affected gene as the TWIST1 gene, which contains instructions for making a protein needed for bone and muscle development in the head and face. If the TWIST1 gene contains a mutation, this protein is not released leading to delayed development of cells in the skull.

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 his or her children will inherit the condition.

What are the signs and symptoms of Saethre-Chotzen syndrome?

The symptoms of Saethre-Chotzen syndrome are extremely variable from person to person – even if several members of the same family have the condition, the degree to which they are affected can range from mild to severe.

Children with Saethre-Chotzen syndrome have a characteristic appearance due to the problems with the skull plates fusing. They tend to have a high forehead with a low hairline at the front, widely spaced eyes often with droopy eyelids and a broad and prominent bridge of the nose. Some children may have a degree of facial asymmetry – that is, each side of the face is affected to a different degree. Others may have a cleft palate (a hole in the roof of the mouth) or low set ears. Intelligence is usually normal.

The hands and feet may be affected in Saethre-Chotzen syndrome, with the second and third finger being webbed towards the base of the fingers. The big toe may be broader than usual, sometimes with a duplicate bone at the nail end.

How is Saethre-Chotzen syndrome diagnosed?

As children with Saethre-Chotzen 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. Genetic testing of a blood sample may be suggested to confirm that the TWIST1 gene contains the mutation.

How is Saethre-Chotzen syndrome treated?

As Saethre-Chotzen 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.

Skull re-shaping surgery, when necessary, usually 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. Raised pressure inside the head is rare and most operations are needed for cosmetic reasons.

Surgery to treat the hand and foot problems is rarely needed as the webbing is quite mild and does not usually affect function. The broad big toe tends not to have any effect on walking. The droopy eyelids can be treated later in childhood if they are affecting vision, particularly when looking upwards, but this is a minor plastic surgery procedure.

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

What happens next?

The outlook for children with Saethre-Chotzen syndrome 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.

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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