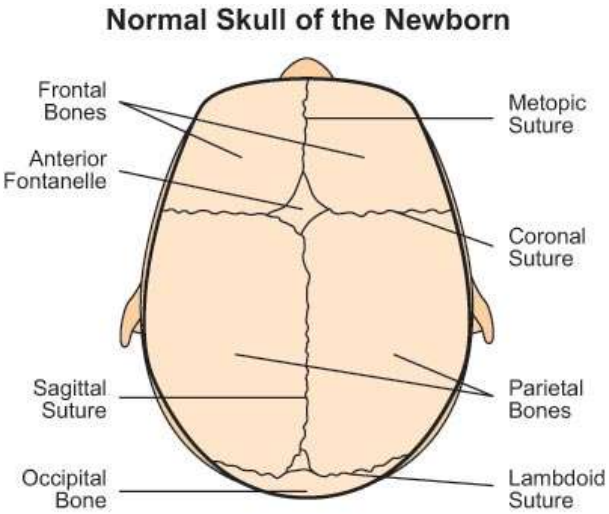
Carpenter syndrome: information for families

Carpenter syndrome is a type of craniosynostosis named after the doctor who first described the condition. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of Carpenter syndrome (also known as acrocephalopolysyndactyly type 2 or ACPS II).

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.

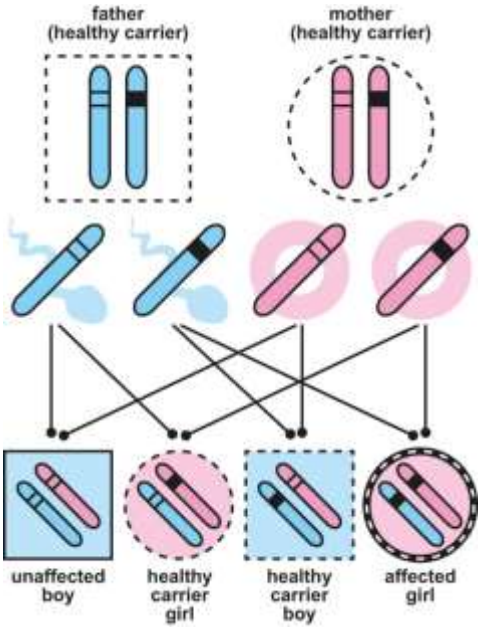
When more than one suture is affected, it is called 'complex craniosynostosis'. This may happen as part of a syndrome (collection of symptoms often seen together), and so may be referred to as 'syndromic' as well.

In Carpenter syndrome, one or more of the sutures can fuse too early, causing the skull to be misshapen. As well as the skull and face, the hands and feet are also affected in Carpenter syndrome with the fingers and toes joined or webbed (syndactyly) or duplicated (polydactyly).

What causes Carpenter syndrome?

Carpenter syndrome is a genetic condition, caused by a mutation (change) on a specific gene. Research has identified the affected genes as the RAB23 gene or MEGF8 gene. Both these genes affect 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 recessive manner – this means that a child only has to inherit the faulty gene from both parents to develop the condition.



What are the symptoms of Carpenter syndrome?

Children with Carpenter syndrome have a characteristic appearance due to the problems with the skull plates fusing. Their eyes are widely spaced and the bridge of their nose is flattened with wide nostrils. In addition, their eyes may be set lower than usual and may be an unusual shape.

The hands and feet may also be affected in Carpenter syndrome. Children may have short fingers (brachydactyly) that are joined (syndactyly). Their feet may also be webbed and some toes may be duplicated (polydactyly).

Most children with Carpenter syndrome have learning disabilities or developmental delay, which varies from mild to severe. The severity of learning disabilities does not seem to be linked to the severity of skull suture fusion.

Many children have heart problems, short stature and a tendency to being overweight. Children may have genital abnormalities such as undescended testicles in males.

How is Carpenter syndrome diagnosed?

As children with Carpenter 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.

How is Carpenter syndrome treated?

As Carpenter 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, ophthalmologists (eye specialists), ear, nose and throat (ENT) surgeons, 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 brain is raised. Children will be monitored regularly so that any problems are identified quickly so that treatment can be offered promptly.

In some 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. If the toes are fused as well, these may not require surgery as they rarely cause any functional problems.

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

What is the outlook for children and young people with Carpenter syndrome?

The outlook for children born with Carpenter syndrome is variable depending on the severity of their symptoms.

Children with Carpenter 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 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 offers 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.