



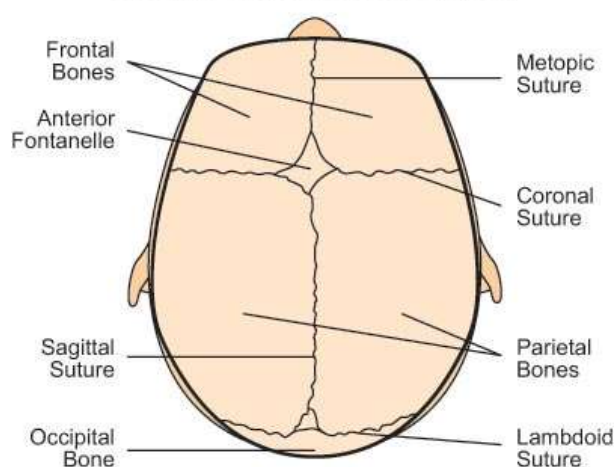
Great Ormond Street Hospital for Children NHS Foundation Trust

Antley-Bixler syndrome: information for families

Antley-Bixler syndrome is a type of complex craniosynostosis named after the doctors who first described it. As well as the skull, the arms may also be affected. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of Antley-Bixler 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.

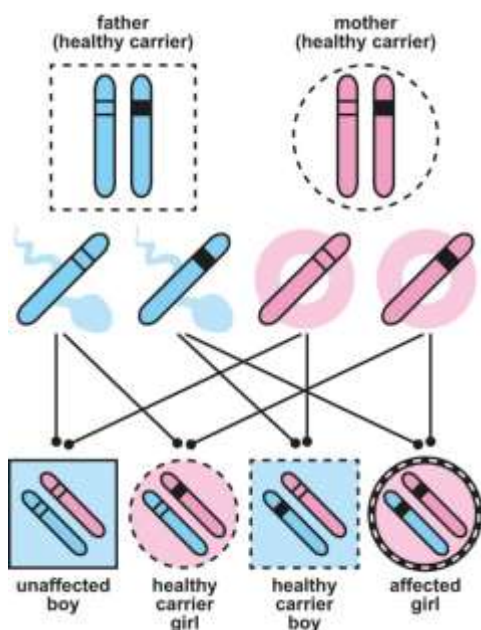
When more than one suture is affected, it is called 'complex craniosynostosis'. This may happen as part of a syndrome (collection of symptoms often seen together), and so may be referred to as 'syndromic' as well.

In Antley-Bixler syndrome, both coronal sutures fuse before birth, leading to the skull being short from front to back but wide from side to side. More rarely, the lambdoid and metopic sutures are also fused. The facial bones are also affected, as the cheekbones and upper jaw do not grow in proportion to the rest of the skull. As well as the skull and face, the upper arm bones are also fused so that the elbow joint has limited movement.

What causes Antley-Bixler syndrome?

Antley-Bixler syndrome is a genetic condition, caused by a mutation (change) on a specific gene. Research has identified two affected genes – one is the P450 oxidoreductase gene and the other is the Fibroblast Growth Factor Receptor 2 (FGFR2) gene. Both 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 Antley-Bixler syndrome?

Children with Antley-Bixler 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.

They also have a condition called choanal atresia – this is where one or both nasal passages are blocked by bone or tissue. Choanal atresia causes breathing difficulties, as babies do not breathe properly through their mouth for the first few months. This will require surgery soon after birth.

In addition, the elbow joint has restricted movement as the radius and/or ulna and humerus are fused. The other joints in the arm – shoulder and wrist – are not usually affected.

There may also be abnormalities affecting the internal organs, particularly those in the genito-urinary system. Some children with Antley-Bixler syndrome, particularly those with the P450 oxidoreductase gene mutation have problems with the creation and use of steroids within the body.

All the signs and symptoms of Antley-Bixler syndrome are extremely variable – where several family members are affected, they may have a range of symptoms.

How is Antley-Bixler syndrome diagnosed?

As children with Antley-Bixler 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 tests may be suggested to identify the gene mutation for planning future pregnancies.

How is Antley-Bixler syndrome treated?

As Antley-Bixler 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, hand 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 or breathing problems are severe. 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.

Further surgery to improve the midface problems may 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. Chiari malformation (if present) can be treated by widening the open at the base of the skull. Orthodontic treatment using braces will be suggested to improve overcrowding and speech.

Unless there are severe functional problems, the fused elbow joint is not usually corrected as most children find ways to manage daily tasks. Support and guidance from occupational therapists and physiotherapists will be helpful, as can adaptations and specialised equipment to help with specific tasks.

As the bone continue 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 Antley-Bixler syndrome?

The outlook for children born with Antley-Bixler syndrome is variable depending on the severity of their symptoms and the impact it has on bodily functions. 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.