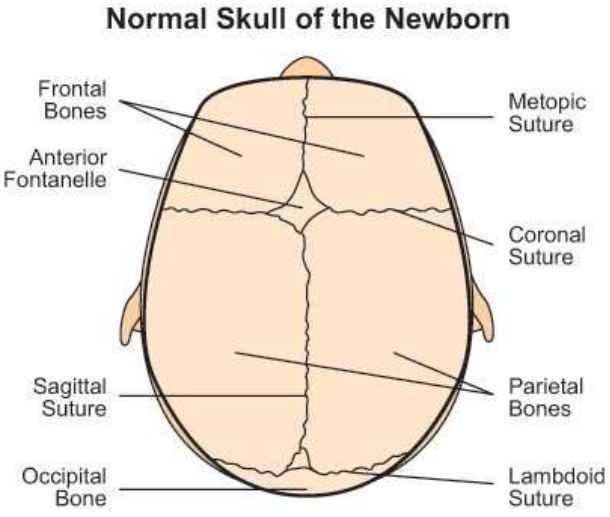
Muenke syndrome: information for families

Muenke syndrome (also known as FGFR3 associated craniosynostosis or P250arg mutation) is a type of complex craniosynostosis named after the doctor who first described it in the mid-1990s. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of Muenke syndrome.

As it is recently described, there are several other names for the condition: FGFR3 associated coronal synostosis which describes the gene and its usual effects and P250arg mutation which describes the location of the affected gene. In time, one name for the condition will become more frequently used than the others.

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 Muenke syndrome, one or both coronal sutures fuse before birth, leading to the skull being short from front to back either on one side or both but wide from side to side. Other sutures may occasionally be affected.

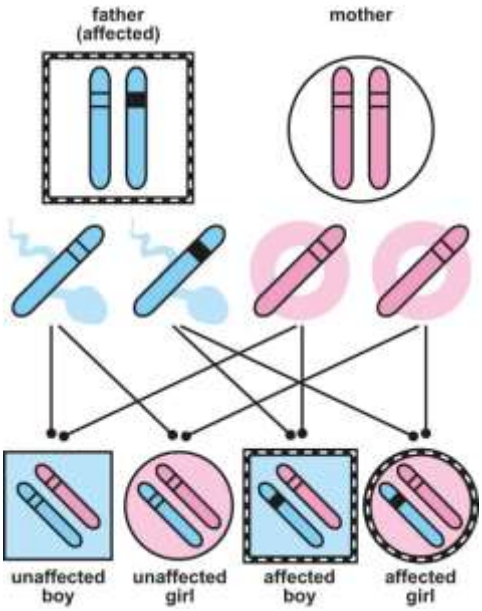
Children with Muenke syndrome often have sensorineural hearing loss. Some may have learning disabilities, although this tends to be mild rather than severe. Other parts of the body may be affected too, such as the hands and feet although this is not usually a problem.

What causes Muenke syndrome?

Muenke 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 3 (FGFR3) 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 their children will inherit the condition.



What are the symptoms of Muenke syndrome?

Muenke syndrome is a very variable condition with some people identified as having the mutated gene showing no signs at all.

Others may have a slightly enlarged skull (macrocephaly) but no fused sutures. In others, either one or both coronal sutures fuse early leading to a misshapen skull. Around 10 per cent of children have epilepsy. The fingers (and occasionally toes) may be shorter than usual, webbed (syndactyly) or bent, but rarely to such a degree that treatment is needed.

The majority of children have some sensorineural hearing loss – the hair cells inside the cochlea are damaged so although sound travels through the ear in the usual manner, the hair cells are not stimulated and so no nerve impulse is sent to the brain.

Some children with Muenke syndrome have learning disabilities or developmental delay, which tend to be mild rather than severe.

A small number of children have short stature, although their rate of growth and development is normal.

How is Muenke syndrome diagnosed?

As children with Muenke syndrome do not have a characteristic appearance unlike in other forms of craniosynostosis, diagnosis is usually made using a blood test to identify the specific gene mutation. As more is known about Muenke syndrome, it is becoming clear that children who may have been diagnosed with other craniosynstosis syndromes in the past, such as Pfeiffer, Crouzon and Saethre-Chotzen syndrome, may actually have Muenke syndrome. This misdiagnosis has no clinical effect on treatment.

How is Muenke syndrome treated?

As Muenke 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, audiologists (hearing specialists), dentists and orthodontists, geneticists and speech and language therapists with other specialists brought in as needed.

If required, 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 it to give a more normal skull shape. Raised pressure inside the head is rare and most operations are needed for cosmetic reasons.

There are various options for treating the sensorineural hearing loss depending on its severity. Some people can be fitted with hearing aids, which amplify (make louder) sounds until they can be picked up by the hair cells. However, this will not benefit people whose hair cells are

too damaged to pick up any sound and in these cases a cochlear implant may be suggested.

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

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

The outlook for children with Muenke syndrome is good with the vast majority growing up to lead a normal life, working and raising a family. Most children are of normal intelligence so usually do well at school, college and university. Hearing support early in life will help with speech and language development.

Children with Muenke syndrome will require long term monitoring, particularly during periods of growth in childhood and adolescence, but further craniofacial surgery is unusual. 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.