



Great Ormond Street Hospital for Children NHS Foundation Trust

Midfacial cleft: information for families

A midfacial cleft is a congenital (present at birth) condition affecting the bones and soft tissues of the face and neck. The two halves of the skull fail to join during pregnancy, leaving a cleft or dip along the central portion of the face. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes symptoms and treatment of midfacial cleft.

Midfacial clefts can occur as part of a syndrome (collection of symptoms often seen together) or on its own.

What causes midfacial cleft?

Midfacial clefts occur early in pregnancy when the head and neck are forming. For reasons we do not yet know, the two halves do not join together as they should.

Doctors believe that in some cases, a midfacial cleft is caused by a mutation (change) on a specific gene. The gene mutation can be passed on from parent to child but in many cases develops sporadically (out of the blue). The genes causing a midfacial cleft have not yet been identified.

Another less common cause of midfacial cleft is trauma early in pregnancy, which interrupts the formation of the head and neck. In many cases, the cause is unknown (idiopathic).

What are the symptoms of midfacial cleft?

The main symptom is a cleft or dip in the central portion of the face that runs from the top of the skull to the chin. The cleft can affect all or part of this area of the face. It can cause a cleft lip and/or

palate affecting both sides of the mouth (bilateral).

The nose is also affected – in mild cases, the nose is wide with deep grooves either side. In more severe cases, the nose is split in two (bifid) down the centre. The eyes are also widely spaced (hypertelorism) due to the cleft.

The front of the forehead may also have a cleft, which sometimes leaves the surface of the brain exposed (encephalocele). Children with an encephalocele can have additional problems with their hormones (chemical messengers that turn on and off processes in the body). This may have an effect on growth and puberty. They may also have learning disabilities although these are often at the mild end of the spectrum.

In addition, there may be a degree of facial asymmetry, where one side of the face looks quite different to the other. The upper jaw may be underdeveloped and set further back than usual and the eyes may be uneven in size and position.

How is midfacial cleft diagnosed?

As children with a midfacial cleft have a characteristic appearance, no specific diagnostic tests are needed. Imaging scans, such as x-ray, CT or MRI may be suggested to monitor facial abnormalities before, during and after treatment.

A midfacial cleft can sometimes be diagnosed before birth using routine ultrasound scans. Otherwise, all are diagnosed soon after birth.

Genetic testing may be suggested to try to identify a gene mutation, which may be helpful in planning future pregnancies.

How is midfacial cleft treated?

As midfacial cleft can affect various areas of the face, 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.

The first stage of treatment is to correct the cleft lip and/or palate if this is present – this usually occurs within the first year. The midfacial cleft will need to be re-shaped and closed earlier, especially if the brain is exposed or at risk of damage due to the lack of skull protection.

In many cases, initial skull re-shaping surgery takes place within the first few years of life. The bifid nose may also be corrected at the same time – this involves splitting the skull down the middle and reshaping the centre portion so that the nose and other midline structures are more even or closer together.

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.

Any endocrine problems will need long term monitoring and additional treatment may be needed, for instance, to bring on puberty if this is delayed due to hormonal problems.

The eye sockets will be re-shaped so that the eyes are not so widely spaced. Bone grafts to fill any gaps left by the cleft, particularly the upper and lower jaw, is carried out in adolescence, when adult teeth have erupted. Orthodontic treatment using braces will be suggested to improve overcrowding and function.

Further surgery to improve the midface problems can be carried out at any time during childhood if they are causing functional problems. Surgery to improve your child's appearance rather than function will usually be carried out in adolescence.

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 a midfacial cleft?

The outlook for children with a midfacial cleft is good with few long term effects on bodily functions such as breathing, vision and hearing. Children with learning disabilities will benefit from support in education and day to day life although a degree of independence may be possible. The majority of children with no learning disabilities grow up to lead a normal life, working and integrating into society.