Craniofacial microsomia

This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of craniofacial microsomia (also known as hemifacial microsomia Goldenhar syndrome) and where to get help.

What is craniofacial microsomia?

Craniofacial microsomia is a condition where one or both sides of the face (facial) is underdeveloped (microsomia). It is very variable and can affect some or all of the ear, eye, skull, cheek, jaw facial movement, soft tissue and teeth on one or both sides of the head.

What causes craniofacial microsomia?

In some cases, it is believed that craniofacial microsomia is a genetic condition caused by a mutation (change) on a specific gene, although all of the the genes are yet to be identified. In very rare cases, craniofacial microsomia can be passed from parent to child but in most cases develops sporadically (out of the blue).

What are the signs and symptoms of craniofacial microsomia?

The symptoms of craniofacial microsomia are extremely variable and can affect various areas of the skull and face on the affected side.

The jaw – both upper and lower jaws – can be underdeveloped which may causes problems with breathing, feeding and speech. Rarely, there may also be a cleft palate and problems with tooth development. The mouth may be wider than normal and missing some of the muscles surrounding it, leaving it weak and slanted downwards on the affected side. If the skull bones are affected, the forehead and cheek on one side may appear flattened and the eye socket may be smaller than usual or displaced. Occasionally, the eye on the affected side may also be smaller or absent.

The ear on the affected side may be an abnormal shape, smaller than usual or absent. The ear canal may also be absent causing and there can be associated total hearing loss. There are also sometimes skin tags in front of the ear although these do not interfere with hearing.

Sometimes other areas of the body can be affected, but children are given a full check-up and diagnostic tests to confirm or rule out any other problems.

How is craniofacial microsomia diagnosed?

Children with craniofacial microsomia tend to have a characteristic appearance so no specific diagnostic tests are needed. However, in mild cases, imaging scans such as x-ray and CT scans may be needed to 'measure' the amount of difference in each side of the face. As the gene and/or chromosome change causing craniofacial microsomia has not yet been identified, genetic testing is rarely suggested.

How is craniofacial microsomia treated?

As craniofacial microsomia can affect various areas of the skull and 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, maxillofacial surgeons, plastic surgeons, ear, nose and throat surgeons, audiologists, ophthalmologists, speech and language therapists, psychologists, dentists and orthodontists, with other specialists brought in as needed.

Early treatment may be needed if the underdeveloped jaw is causing problems with breathing or feeding. Airway support such as a nasal prong may be suggested – only rarely are the breathing problems so severe that a tracheostomy (artificial opening into the windpipe) is needed.

If the size of the eye socket is likely to cause vision problems, this may be re-shaped so that the eye can sit in the correct position and the eyelids close to protect it. If the eye is absent, a prosthesis (false eye) will be made to match your child's other eye.

At primary school age, ear reconstruction may be suggested, which involves a staged operation to create a 'framework' for the new ear from rib cartilage and insert it under the skin on the side of the head before it is elevated some months later. Alternatively, children may prefer to have a prosthetic (plastic) ear created which is fixed to a bar inserted into the skull bone. Both these options restore the appearance of the ear but cannot correct any hearing problems caused by an absent ear canal.

In childhood, surgery to correct the underdeveloped jaw may be suggested. This can be treated with using bone grafts from elsewhere on the body (usually the ribs) or by distraction, which involves an external fixator 'stretching' the bone until it is a more proportionate size. The position of the teeth will need correcting later in childhood, using orthodontic braces which may be removable or fixed for a period of months.

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

What happens next?

The outlook for children born with craniofacial microsomia is variable depending on the severity of their symptoms and the impact it has on bodily functions such as breathing, vision and hearing. Some children and families benefit from psychological input at various stages throughout childhood and adolescence. 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 or telephone them on 01454 850 557.

The Goldenhar Support Group offers support to anyone affected by Goldenhar syndrome, of which craniofacial microsomia is a feature. Visit their website at www.goldenhar.org.uk. If you would like to talk to a member of the group, email them via their website and they will call you back.

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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