



Great Ormond Street Hospital for Children NHS Foundation Trust

Freeman-Sheldon syndrome: information for families

Freeman-Sheldon syndrome (previously known as Whistling Face syndrome) is a rare genetic condition that affects the mouth, face, hands and feet. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of Freeman Sheldon syndrome.

In Freeman-Sheldon syndrome, the muscle fibres do not form properly so development is interrupted.

What causes Freeman-Sheldon syndrome?

Freeman-Sheldon syndrome is a genetic condition, caused by a mutation (change) on a specific gene. Research has identified the affected gene as the MYH3 gene, which is responsible for the movement of substances between cells during development. It is a major component of muscle fibres so is involved in muscle development.

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 can be passed on in an autosomal dominant manner – this means that a child only has to inherit the faulty gene from one parent to develop the condition. It can also be passed on in an autosomal recessive manner – this means that a child has to inherit the faulty gene from both parents to develop the condition.

In some people, the gene mutation is never identified so the cause of the condition is not known.

What are the symptoms of Freeman-Sheldon syndrome?

The main feature of Freeman-Sheldon syndrome is an undersized mouth (microstomia) with pursed lips – this is why the condition was previously known as Whistling Face syndrome. The airway may also be narrowed, which can cause breathing difficulties soon after birth.

Children can also have a condition called malignant hyperthermia, which causes a severe and life threatening reaction to some anaesthetic medicines.

In addition, the middle portion of the face is flatter than usual, with a decreased amount of nose cartilage leading to a broad flat nose. The forehead may be prominent and the space between the base of the nose and upper lip may be quite deep.

The eyes can also be affected in children with Freeman-Sheldon syndrome, being widely spaced, sometimes with droopy eyelids and a squint. One or both feet may be affected by talipes (clubfoot) where the foot is turned inwards and upwards. Muscle contractures (shortened muscles) can also affect movement in the hands. Some children have feeding problems and mild learning disabilities.

How is Freeman-Sheldon syndrome diagnosed?

As children with Freeman-Sheldon 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 testing may also be suggested to identify the gene mutation and/or monitor future pregnancies.

How is Freeman-Sheldon syndrome treated?

As Freeman-Sheldon syndrome can affect various areas of the body, treatment is best delivered at a specialist centre where a multidisciplinary team approach can be taken and specialist anaesthesia is available. 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 skull bones may be re-shaped in childhood to reduce the prominence of the forehead. Orthopaedic surgery or bracing using the Ponseti technique will be needed to correct the talipes. Hand surgery and splintage is usually required to maximise the use of the hands, alongside physiotherapy and occupational therapy. Speech therapy may be needed to help with any feeding problems.

What is the outlook for children and young people with Freeman-Sheldon syndrome?

The outlook for children with Freeman-Sheldon syndrome is good once any initial breathing difficulties have been treated.

The results of talipes treatment are usually very good with few lasting effects, as are the results of hand surgery. The majority of children grow up to lead a normal life, working and raising a family.

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.

STEPS is the UK charity supporting people with lower limb abnormalities. Call their helpline on 01925 750 2719 or visit their website at www.steps-charity.org.uk