Nager syndrome

This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of Nager syndrome and where to get help.

What is Nager syndrome?

Nager syndrome is a congenital (present at birth) condition affecting the bones and tissues in the face. It also affects the arms and hands, and occasionally the legs and feet too. Early in pregnancy, the cheekbones, eye sockets and jaw may not develop properly. This causes a characteristic appearance with downward sloping eyes and a small jaw. Nager syndrome is similar to Treacher-Collins syndrome, with additional problems with the arms and hands and often associated with cleft palate.

What causes Nager syndrome?

Doctors suspect that Nager syndrome may be a genetic condition, caused by a mutation (change) on a specific gene. The specific gene(s) causing Nager syndrome have not yet been identified.

The gene(s) suspected of involvement in Nager syndrome alter the development of the first and second pharyngeal arches early in pregnancy. The pharyngeal arches are structures either side of the head and neck that develop into the structures of the face and neck. In particular, the first and second pharyngeal arches form into the nerves and muscles needed for showing facial expressions and chewing, the outer ear and the structures within the middle ear.

Typical of syndromes there are associated problems with arm and hand development.

What are the signs and symptoms of Nager syndrome?

Children with Nager syndrome have a characteristic appearance due to the problems with their cheekbones, jaw and eye sockets forming. If the jaw is very small and/or underdeveloped, breathing difficulties may become apparent soon after birth. The jaw problems can also cause feeding difficulties. The ears are often affected in children with Nager syndrome either being absent or very small (microtia). If the internal structures of the ear are also underdeveloped this can lead to hearing impairment.

The arms and hands are also affected in Nager syndrome. Often, the elbow joint is stiff so bending the arm is difficult. The radius bone in the arm may be missing or underdeveloped causing the hand to be bent inwards towards the body (radially deviated). This can affect all the structures on the radial side of the upper limb, including the bones and soft tissues (muscles, tendons, joints, nerves and blood vessels).

Children with Nager syndrome will also have problems with their hands. Most commonly, the thumb of both hands is underdeveloped or absent, leading to problems with grip and fine movement. The other fingers may be abnormally curved, stiff and sometimes webbed (syndactyly). In some children, their legs and feet are similarly affected.

Rarely, other structures in the body are also affected.

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How is Nager syndrome diagnosed?

As children with Nager 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. Imaging may also be needed to examine the internal structures of the ear alongside hearing tests to diagnose hearing loss.

How is Nager syndrome treated?

As Nager syndrome can affect various areas of the head 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, ear, nose and throat (ENT) surgeons, hand surgeons, plastic surgeons, audiologists (hearing specialists), dentists and orthodontists, geneticists and speech and language therapists with other specialists brought in as needed.

Initially, stabilising a child's breathing problems will require treatment. For some children, these are so severe that they need a tracheostomy – artificial opening into the windpipe – to allow them to breathe. Other children may only need breathing support at night. Feeding problems may be helped with enteral feeding – a feeding tube or gastrostomy directly into the stomach bypassing the mouth and throat.

If the thumb is present but weak, surgery can be undertaken to stabilise any unstable joints and strengthen the thumb by taking a tendon or a muscle from elsewhere in the hand. If the thumb is absent or unstable, the index finger on that hand may be moved to the thumb position (pollicisation). Any finger webbing, if causing a problem with function, may be corrected in a series of operations during childhood.

Later in childhood, the underdeveloped jaw may require treatment – often with bone grafts and jaw distraction. This is a long term treatment but has good results for the majority of children. Ear reconstruction can also be carried out in later childhood, either creating a new ear modelled on the other ear or the parents' ears if both are absent. Ear reconstruction only improves the appearance of the ear, it will not improve function. Children with hearing impairment will need support in terms of hearing aids or cochlear implant as well as speech and language therapy.

As the bone continue to grow during childhood and adolescence, further surgery may be needed to reconstruct face shape.

What happens next?

The outlook for children born with Nager syndrome is variable depending on the severity of their symptoms and the impact it has on bodily functions such as breathing and hearing. 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.

REACH (The Association for Children with Hand or Arm Deficiency) can offer information and support. Call their helpline on 0845 1306 225 or visit their website at www.reach.org.uk

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.

Read Fiona's story on the GOSH website at – www.gosh.nhs.uk/medical-conditions/search-for-medical-conditions/nager-syndrome/nager-syndrome-real-stories

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