

Noonan syndrome

Noonan syndrome is a genetic condition that usually includes heart abnormalities and characteristic facial features. About one-third of affected children have mild intellectual disability. Noonan syndrome varies from relatively mild to severe. In some cases, the physical characteristics are so subtle that the child doesn't look noticeably different.

Estimates vary, but it is thought that about one in 1,000 to one in 5,000 children have Noonan syndrome, with males and females equally affected. Genetic changes (mutations) on several genes are known to cause Noonan syndrome, but the diagnosis is usually established on the basis of clinical features. There is no cure, but medical management of possible complications is important.

A range of abnormalities

Parts of the body typically affected by Noonan syndrome include:

- Heart
- Facial features
- Musculoskeletal system
- Kidneys, spleen and liver.

Heart abnormalities

About 65 per cent of children with Noonan syndrome have heart abnormalities. The most common include:

- **Pulmonary valve stenosis** – the pulmonary valve allows blood with reduced oxygen content (deoxygenated) to leave the heart and go to the lungs. Pulmonary valve stenosis occurs when the pulmonary valve is narrower than normal.
- **Hypertrophic cardiomyopathy** – thickening of the heart muscles.
- **Atrial septal defect** – an abnormal hole between the two upper chambers of the heart (atria).
- **Ventricular septal defect** – an abnormal hole between the two lower chambers of the heart (ventricles).

Characteristic facial features

The severity of the Noonan syndrome 'look' varies from one child to the next and can change with age. Characteristics may include:

- Heavy or hooded eyelids that may interfere with vision (ptosis)
- Downward sloping eyes with an extra fold of skin at each inner corner
- Widely spaced eyes (hypertelorism) with a flattened bridge of the nose
- Brilliant blue or blue-green eyes
- Strongly arched (diamond-shaped) eyebrows
- Low-set ears, tilted back so that the lobes point forward more than usual
- Low hairline at the neck
- Coarse and curly hair
- Short neck with extra skin folds or 'webbing' at the back of the neck – this makes the neck look wider when viewed from behind.

Musculoskeletal system abnormalities

Some possible abnormalities of the musculoskeletal system include:

- Short stature. Although birth weight is usually average, children with Noonan syndrome are frequently shorter than expected for their age. As adults, people with Noonan syndrome are often short - men average 165cm (5'5") and women 152cm (5') in height
- Low muscle tone and impaired coordination
- Deformity of the breastbone (sternum)
- Curvature of the spine (scoliosis).

Intellect is not always affected

It was once commonly believed that all children with Noonan syndrome were intellectually disabled. This is actually true for only about one third of affected children. Intelligence quotient (IQ) scores generally range from 50 to 119 (100 being average). Delays in language development are the most common problem experienced by children with Noonan syndrome, with one in four affected.

Associated problems in Noonan syndrome

Depending on the severity, problems may include:

- Intellectual disability, in about one third of cases
- Eye problems, including squint and short-sightedness
- Babies may have feeding problems and may vomit easily
- Hearing problems, including a susceptibility to glue ear
- Delay in reaching developmental milestones, such as sitting unaided and walking
- Easily bruised skin and susceptibility to nose bleeds
- Reduced resistance to infection
- Dental problems, including missing teeth
- Increased risk of seizures
- Undescended testicles
- Decreased fertility in males who have had undescended testicles
- Delayed puberty.

An inherited disorder

Noonan syndrome is a genetic condition caused by a change in one of several genes. It was once believed that most cases of Noonan syndrome were sporadic, which means the child's gene spontaneously changed. However, researchers now suspect that Noonan syndrome is inherited in up to 75 per cent of cases.

The pattern of inheritance is autosomal dominant, which means that the gene (and hence the condition) can be transmitted from parent to child. Each child of a parent who carries the gene has a 50 per cent chance of inheriting the gene and developing the condition.

Diagnosis of Noonan syndrome

Diagnosis is made by noting the presence of typical Noonan syndrome features and ruling out other possible causes, such as fetal alcohol spectrum disorder, neurofibromatosis type 1 (also known as Von Recklinghausen's disease, or NF1) and Turner syndrome. Gene testing is available and can be discussed with a geneticist.

Treatment for Noonan syndrome

There is no cure for Noonan syndrome. Treatment aims to ease some of the associated problems and may include one or more options:

- If the heart abnormality is mild, it may be enough to carefully monitor the child on an outpatient basis.
- Severe heart abnormalities are corrected surgically.
- If feeding is a problem, the baby may be fed by nasogastric tube (a tube threaded into the nose, down the oesophagus and into the stomach).

- Prescription glasses can correct short-sightedness.
- Regular injections of growth hormone can help the child to achieve their intended height.
- Undescended testicles can be corrected surgically (orchidopexy).
- If the child experiences seizures, anticonvulsant medication may be prescribed.
- Speech therapy may be helpful.
- Special education is necessary for about 10 per cent of affected children.
- Behaviour management may be appropriate.
- Treatment to manage mild blood-clotting problems may be necessary.
- Dental and orthodontic treatment may be advised.
- Glue ear can be managed with an operation to insert drainage tubes (grommets).

Where to get help

- Your doctor
- Genetic Health Services Victoria, Royal Children's Hospital Tel. (03) 8341 6200
- Genetic Support Network of Victoria Tel. (03) 8341 6315

Things to remember

- Noonan syndrome is a genetic condition that typically includes heart abnormalities and characteristic facial features.
- About one-third of affected children have mild intellectual disability.
- Noonan syndrome may be inherited in up to 75 per cent of cases.

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Genetic Health Services Victoria

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