CONDITION

Noonan syndrome

Noonan syndrome is a genetic condition that can affect growth, development, facial features, and the heart.

WHAT IT IS
Genetic condition affecting growth, facial features, heart, and development

BODY SYSTEM
Growth, heart and circulation, development, blood, and lymph system

MAIN SYMPTOMS
Distinct facial features, short stature, heart problems, developmental differences

WHAT CAUSES IT
Inherited or new changes in cell-signalling genes

HOW IT IS DIAGNOSED
Clinical assessment and genetic testing for a causative variant

MAIN TREATMENT
Monitoring and treatment of individual health problems

CAN IT BE CURED?
No, lifelong genetic changes cannot be removed

IS IT AN EMERGENCY?
Sometimes, with breathing trouble, fainting, or uncontrolled bleeding

LONG-TERM OUTLOOK
Many lead active lives with ongoing health monitoring

Overview

Noonan syndrome is a genetic condition present from birth. It affects how cells communicate during growth and development. Its effects differ widely, even among people in the same family.

Common features include distinctive facial characteristics, shorter height, and differences in the shape of the chest. Some people have a heart condition, learning difficulties, feeding problems, easy bruising, or delayed puberty. Others have few health problems and may not be diagnosed until adolescence or adulthood.

Noonan syndrome belongs to a group of conditions called RASopathies . These involve the RAS-MAPK pathway, a chain of signals that helps control how cells grow and divide.

The condition matters because some of its effects, particularly heart problems, bleeding difficulties, and developmental needs, require monitoring. Recognising the condition can also help families understand inherited risks and arrange suitable support.

Symptoms

The signs of Noonan syndrome may be noticeable before birth, during infancy, or later in life. No single feature is present in everyone.

Facial features may include widely spaced eyes, low-set ears, drooping upper eyelids, or a broad forehead. The neck may appear short or have extra folds of skin. These features often become less noticeable in adulthood.

Other possible features include:

The most common heart problems include pulmonary valve stenosis , in which the valve carrying blood from the heart to the lungs is narrowed, and hypertrophic cardiomyopathy , in which the heart muscle becomes unusually thick. Some people have other structural heart differences or an irregular heart rhythm.

Intelligence is often within the usual range. When learning difficulties occur, they are commonly mild, although some people need more substantial support. Attention, coordination, language, and social communication can also be affected.

- Shorter height or slower growth
- A broad or sunken chest with widely spaced nipples
- A heart condition present from birth
- Feeding problems and slow weight gain during infancy
- Low muscle tone or delayed movement skills
- Speech, language, or learning difficulties
- Easy bruising, frequent nosebleeds, or prolonged bleeding
- Vision problems, squint, or hearing loss
- Swelling caused by a build-up of lymph fluid
- Delayed puberty
- Testicles that have not moved into the scrotum

Causes and risk factors

Noonan syndrome is caused by a genetic change that disrupts cell signalling. Several genes can be involved, including `PTPN11`, `SOS1`, `RAF1`, `RIT1`, and `LZTR1`. The gene involved can influence which health problems are more likely, but it cannot predict a person's exact course.

Many people have a new genetic change that began in the egg or sperm and was not inherited from either parent. This is called a de novo change.

Most forms follow an autosomal dominant inheritance pattern. This means one altered copy of the gene can cause the condition. A parent with one of these forms has a 1 in 2 chance of passing the altered gene to each child. This chance is the same for every pregnancy.

Some changes in `LZTR1` can follow either dominant or recessive inheritance. In recessive inheritance, a child usually receives one altered copy from each parent. Genetic counselling can explain the pattern that applies to a particular family.

Nothing someone does during pregnancy causes Noonan syndrome.

Diagnosis

Diagnosis begins with a detailed medical and family history and a physical examination. The combination of growth pattern, facial features, chest shape, development, and heart findings may suggest Noonan syndrome.

Genetic testing can confirm the diagnosis when it identifies a causative change in a gene linked to the condition. Testing may use a panel that examines several RAS-MAPK pathway genes at once. A negative result does not always rule out Noonan syndrome because current testing may not detect every causative change.

Other tests assess how the condition is affecting the body. These may include:

Testing during pregnancy may be possible when a family-specific genetic change is known. Ultrasound findings such as extra fluid behind the fetal neck can raise suspicion, but they do not confirm Noonan syndrome because they also occur in other conditions.

Conditions such as Turner syndrome and other RASopathies can share some features. Genetic testing and chromosome testing may help distinguish between them.

- An echocardiogram, which uses ultrasound to examine the heart
- An electrocardiogram, which records the heart's electrical activity
- Blood tests to check blood cells and clotting
- Hearing and eye assessments

- Growth and developmental assessments
- Kidney ultrasound or other imaging when indicated

Treatment and management

There is no treatment that removes the underlying genetic change. Care focuses on monitoring for known problems, treating symptoms, and supporting development and daily life.

Heart care depends on the finding. A narrowed pulmonary valve may need monitoring, a catheter procedure, or surgery. Hypertrophic cardiomyopathy may require medicines, rhythm monitoring, or other specialist treatment. Even people without symptoms may need periodic heart review because some problems can change over time.

Infants with feeding difficulty may benefit from feeding support and nutrition assessment. Some need temporary tube feeding if they cannot safely take enough milk by mouth.

Children with speech, movement, or learning delays may receive speech and language therapy, physiotherapy, occupational therapy, or educational support. Hearing and vision problems should be treated because they can affect communication and school progress.

Growth is tracked over time. Growth hormone treatment may be considered for some children after specialist assessment. It does not treat every feature of Noonan syndrome and requires monitoring, particularly when heart problems are present.

Management may also include:

Genetic counselling can help explain test results, inheritance, family testing, and reproductive options.

- Assessment of unusual bruising or bleeding, especially before surgery
- Treatment for undescended testicles
- Support for delayed puberty
- Dental and orthodontic care
- Treatment of lymphatic swelling
- Psychological or school support when needed

Complications

Heart complications are among the most important long-term concerns. A narrowed valve can obstruct blood flow, while thickened heart muscle can interfere with the heart's filling or pumping. Irregular rhythms may also occur.

Some people have abnormal levels or function of clotting factors or platelets. Platelets are blood cells that help stop bleeding. This can lead to easy bruising or longer bleeding after injury, dental work, or surgery. Routine clotting tests do not identify every possible problem, so a history of unusual bleeding is important.

Lymphatic problems can cause swelling in the hands, feet, or other areas. In uncommon cases, lymph fluid can collect around the lungs or within the digestive system.

Children may experience ongoing feeding, growth, hearing, vision, or learning difficulties. Delayed puberty and undescended testicles can affect fertility, particularly in some males.

Certain genetic changes associated with Noonan syndrome increase the risk of particular blood disorders and cancers, including juvenile myelomonocytic leukaemia. Cancer remains uncommon overall. Monitoring is based on age, symptoms, and the gene involved rather than the same screening plan for everyone.

Prevention

Noonan syndrome cannot usually be prevented because it results from an inherited or new genetic change.

If the causative change has been identified, genetic counselling can explain the chance of it occurring in a future pregnancy. Prenatal testing or testing of embryos created through in vitro fertilisation may be options for some families. These are personal choices, and available options differ by location and individual circumstances.

Early diagnosis does not prevent the condition, but it can help identify heart, feeding, hearing, bleeding, and developmental problems sooner.

Living with it and what to expect

During infancy, feeding and weight gain may need close attention. Heart problems are sometimes detected before birth or soon after delivery. Developmental progress becomes clearer over the first few years.

During childhood, growth is often slower than average. Speech, coordination, attention, or school learning may require support. Many children attend mainstream school, sometimes with

extra help, while others need more specialised education.

Puberty may start later than usual. Some teenagers continue growing after their peers have stopped. Discussions about body image, independence, relationships, and fertility can become important during this period.

Facial features often become subtler with age, but the genetic condition does not go away. Heart disease, hearing loss, bleeding problems, and lymphatic symptoms may still require review in adulthood. Moving from children's to adult services should include a clear summary of previous heart findings, operations, genetic results, and bleeding history.

Many adults with Noonan syndrome work, study, form relationships, and live independently. Others need continuing practical or learning support. Life expectancy may be close to typical when serious complications are absent, but severe heart disease or other major health problems can affect outlook.

When to get medical help

Arrange routine medical follow-up for growth, development, hearing, vision, dental health, and any known heart problem. Seek a medical appointment sooner for new swelling, worsening exercise tolerance, repeated nosebleeds, unusual bruising, feeding difficulties, or concerns about development.

Prompt assessment is also appropriate for persistent paleness, fever without a clear cause, frequent infections, a swollen abdomen, or unexplained weight loss. These symptoms have many possible causes, but they should not be ignored.

Seek emergency care for:

- Severe difficulty breathing or blue-grey lips or skin
- Fainting during exercise or with a racing heartbeat
- Severe chest pain
- Bleeding that does not stop with firm pressure
- Sudden severe weakness, confusion, or loss of consciousness
- A baby who is struggling to breathe or cannot stay awake to feed

Common questions

Is Noonan syndrome a type of Down syndrome?

No. They are separate genetic conditions with different causes. Down syndrome usually results from an extra copy of chromosome 21. Noonan syndrome is usually caused by a change in one of several genes involved in cell signalling.

Can someone have mild Noonan syndrome?

Yes. Some people have only subtle facial features, shorter height, or a minor heart finding. They may not be diagnosed until adulthood, particularly after a child or other relative receives a diagnosis.

Is Noonan syndrome inherited from the mother or father?

It can be inherited from either parent. In many people, however, the genetic change is new and neither parent has it. Genetic testing can sometimes clarify how it arose.

Can people with Noonan syndrome have children?

Many can. Fertility may be reduced in some males, especially when undescended testicles were present. A person with a dominant form usually has a 1 in 2 chance of passing the genetic change to each child.

Does Noonan syndrome affect intelligence?

It can affect learning, but intellectual ability is often within the usual range. Some people have difficulties with language, attention, coordination, or particular school subjects. A smaller number have an intellectual disability.

Does Noonan syndrome get worse with age?

It is not usually a steadily worsening condition. Some childhood issues, such as feeding difficulties or distinctive facial features, may become less prominent. Heart, hearing, bleeding, or lymphatic problems can persist or develop over time, so ongoing follow-up remains important.

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