Noonan syndrome
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.

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 between 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.

Effects of Noonan syndrome

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 165 cm (5’ 5”) and women 152 cm (5’) in height
- low muscle tone and impaired coordination
- deformity of the breastbone (sternum)
- curvature of the spine (scoliosis).

Intelect 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.

Other problems with Noonan syndrome

Depending on the severity, problems may include:

- 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 in males
- decreased fertility in males who have had undescended testicles
- delayed puberty.

Cause of Noonan syndrome

Noonan syndrome is a genetic condition caused by a fault in one of at least seven different genes. In genetic testing, a mutation in the PTPN11 gene causes Noonan syndrome in about 50 per cent of affected people.

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 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.

Genetic testing is available and can be discussed with your doctor.

Treatment for Noonan syndrome

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

- 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 (called 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
- Victorian Clinical Genetics Services, 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.

References

- Noonan syndrome, Centre for Developmental Disability Health Victoria, Australia. More information here.

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More information

Genes and genetics

The following content is displayed as Tabs. Once you have activated a link navigate to the end of the list to view its associated content. The activated link is defined as Active Tab

- A-Z of genetic conditions
- Genes and genetics explained
- Genetic testing

A-Z of genetic conditions

- Ambiguous genitalia
  The causes of ambiguous genitalia include genetic variations, hormonal imbalances and malformations of the fetal tissues that are supposed to evolve into genitals.
- Angelman syndrome
  The characteristic features of Angelman syndrome are not always obvious at birth, but develop during childhood.
- Ankylosing spondylitis
  Ankylosing spondylitis (AS) is a type of inflammatory arthritis that targets the joints of the spine.
- Barrett's oesophagus
  Symptoms of Barrett's oesophagus are similar to regular heartburn, which means many people don't seek treatment until their condition is quite advanced.
- Bipolar disorder
  Bipolar disorder is a type of psychosis, which means the person’s perception of reality is altered. It is characterised by extreme mood swings.
- Central nervous system birth defects
  Folic acid taken before conception, and during at least the first four weeks of pregnancy, can prevent around seven out of 10 cases of neural tube defects.
- Charcot-Marie-Tooth disease (CMT)
  Charcot-Marie-Tooth disease is the most common inherited disorder affecting the peripheral nervous system.
- Cleft palate and cleft lip
  Most cleft palates and cleft lips can be repaired so that appearance and speech develop normally.
- Congenital adrenal hyperplasia (CAH)
  CAH is a rare genetic disorder, but it is well understood and treatment is readily available.
- Creutzfeldt-Jakob disease (CJD)
  Creutzfeldt-Jakob disease is characterised by physical deterioration of the brain, dementia and walking difficulties.
- Cri du chat syndrome
  Most children born with cri du chat syndrome have moderate intellectual disability, with varying degrees of speech delay and some health problems.
- Cystic fibrosis (CF)
  When a person has cystic fibrosis, their mucus glands secrete very thick sticky mucus that clogs the tiny air passages in the lungs and traps bacteria.
- Digestive tract birth defects
  Too much amniotic fluid surrounding the baby during pregnancy (polyhydramnios) may indicate the presence of defects of the digestive tract.
- Down syndrome
  With the support and opportunities available to them today, most people with Down syndrome are able to achieve and participate as valued members of their community.
- Dwarfism
  Dwarfism refers to a group of conditions characterised by shorter than normal skeletal growth.
- Eczema (atopic dermatitis)
  Eczema can vary in severity, and symptoms may flare up or subside from day to day.
**Essential tremor**
Essential tremor causes involuntary shaking or trembling of particular parts of the body, usually the head and hands, but it is not Parkinson's disease.

**Fragile X syndrome**
The facts about fragile X syndrome are complicated, and parents and family members are invited to ask their doctor to refer them to a genetics clinic.

**Friedreich's ataxia**
To the casual observer, a person with Friedreich ataxia may seem to be drunk.

**Genetic factors and cholesterol**
Familial hypercholesterolaemia is an inherited condition characterised by higher than normal levels of blood cholesterol.

**Haemochromatosis**
Haemochromatosis (iron overload disorder) tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses.

**Haemophilia**
All children with severe haemophilia are given preventative treatment with infusions of blood products before they have a bleed.

**Hair**
Human hair grows one centimetre every month.

**Hearing problems in children**
The earlier that hearing loss is identified in children, the better for the child's language, learning and overall development.

**Huntington's disease**
The symptoms of Huntington's disease usually, but not always, first appear when the person is approaching middle age.

**Kabuki syndrome**
Kabuki syndrome affects males and females equally and there is no cure.

**Kennedy's disease**
Kennedy's disease is a rare inherited neuromuscular disorder that causes progressive weakening and wasting of the muscles, particularly the arms and legs.

**Kidneys - medullary cystic kidney disease**
Medullary cystic kidney disease causes the growth of abnormal cysts in the kidneys.

**Kidneys - polycystic kidney disease (PKD)**
Polycystic kidney disease is a common cause of kidney failure in Australia and equally affects men and women of different ethnic backgrounds.

**Klinefelter syndrome**
Klinefelter syndrome is often diagnosed at puberty, when the expected physical changes don't occur.

**Leukodystrophy**
Leukodystrophy refers to a group of inherited disorders that affect the white matter of the brain, which causes loss of normal brain functions.

**Long QT syndrome**
You should be investigated for long QT syndrome if you faint for no apparent reason, during or after exercise or emotional excitement.

**Marfan syndrome**
Some people may not realise they have Marfan syndrome, because their features are either mild or not obvious.

**McCune-Albright syndrome**
The severity of symptoms or how a child with McCune-Albright syndrome will be affected throughout life is difficult to predict.

**Muscular dystrophy**
People affected by muscular dystrophy have different degrees of independence, mobility and carer needs.

**Neurofibromatosis**
Neurofibromatosis is caused by faulty genes, which may be inherited or have spontaneously mutated at conception.

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• Osteoporosis in children
  Osteoporosis in children is rare and usually caused by an underlying medical condition.

• Phenylketonuria (PKU)
  PKU is an inherited disorder that prevents the normal breakdown of a protein found in some foods.

• Prader-Willi syndrome
  A feature of Prader-Willi syndrome is the child's excessive appetite, which often leads to obesity.

• Premature and early menopause
  The symptoms of premature or early menopause are the same as for menopause at any age.

• Rett syndrome
  People with Rett syndrome have a keen desire to communicate.

• Spinal muscular atrophy (SMA)
  A child with spinal muscular atrophy type 1 rarely lives beyond three years of age.

• Tay-Sachs disease
  Tay-Sachs disease is a serious genetic disorder common in Ashkenazi Jews and French-Canadians.

• Thalassaemia
  Thalassaemia is an inherited blood disorder that can cause anaemia or death if not treated.

• Tourette syndrome
  Milder forms of Tourette syndrome can be misdiagnosed, as it often occurs at the same time as attention deficit hyperactivity disorder (ADHD) and other disorders.

• Treacher Collins syndrome
  Treacher Collins syndrome is a genetic disorder that affects growth and development of the head, causing facial defects and hearing loss.

• Tuberous sclerosis
  Tuberous sclerosis is a genetic disorder that affects various parts of the body to varying degrees of severity.

• Turner's syndrome
  Turner's syndrome is a random genetic disorder that affects females, causing short stature and infertility.

• Usher syndrome
  Services aim to help a person with Usher syndrome prepare for the dual loss of sight and hearing.

• Von Willebrand disease
  A person with von Willebrand disease may have frequent nosebleeds, heavy menstruation or excessive bleeding from the mouth.

• Williams syndrome
  Williams syndrome often goes undiagnosed, which means that some people with the disorder fail to get the support and treatment they need until later in life.

• Wilson disease
  In Wilson's disease, a build-up of copper damages organs including the liver, nervous system, brain, kidneys and eyes.

Genes and genetics explained

• Gene therapy
  Gene therapy is an experimental form of treatment that targets the faulty genes that cause genetic diseases.

• Genetic disorders
  Genetic disorder is caused by an altered or faulty gene or set of genes.

• Genetic services in Victoria
  Genetic services can help people who are affected by, or who are at risk of, inherited conditions or birth defects, to make informed choices about their healthcare.

Genetic testing

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DNA profiling

DNA profiling is a way of establishing identity and is used in a variety of ways...

Egg freezing

You can freeze your eggs for medical reasons or for reasons that are more to do with your life circumstances...

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Genetic testing for inherited cancer

A predisposition to certain cancers can be inherited via altered genes...

Newborn bloodspot screening

Every newborn baby in Australia is offered a newborn bloodspot screening test to identify those at risk of rare, but serious, medical conditions...

Pregnancy tests – chorionic villus sampling

Chorionic villus sampling (CVS) is a pregnancy test that checks the baby for some abnormalities...

Pregnancy tests - maternal serum screening

Maternal serum screening can indicate increased risk of abnormalities in the unborn child, but is not a diagnosis...

Related Information

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Related information on other websites

- Centre for Developmental Disability Health (CDDHV)
- Centre for Genetics Education
- Newlife (foundation for disabled children UK)
- NSW Council for Intellectual Disability
- Spine Society of Australia - National Self-Detection Program for Scoliosis
- The Noonan Syndrome Support Group Inc.
- Victorian Clinical Genetics Services (VCGS)

Support Groups

- Noonan Syndrome Support Group

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