Noonan Syndrome

Knowledge
to grow by

Models throughout are used for illustrative purposes only.

Noonan syndrome is named for Dr Jacqueline Noonan, a pediatric cardiologist, who first identified it in 1962. Noonan syndrome is caused by a **genetic mutation** that may cause **congenital heart disease**, distinctive facial features, short **stature**, and other conditions.

What does it mean if the doctor says that your child has short stature caused by Noonan syndrome? It means that your child is much shorter than average for his or her age group due to a rare genetic disorder that may affect growth.

Approximately 50% to 70% of patients with Noonan syndrome have short stature. Features appear at birth or during early childhood. Boys and girls are affected equally. It is estimated that 1 in 1,000 to 2,500 infants is born with Noonan syndrome.

If your child is shorter than average, he or she may be seen by an **endocrinologist**. This doctor is a specialist who is trained to diagnose and treat children with growth disorders. The endocrinologist will evaluate your child to find the cause of your child's short stature.

Noonan syndrome

Children with Noonan syndrome usually have more than one medical condition. This means that more than one doctor may be involved in helping them get better.

The entire body is made of tiny units called cells. Inside each cell are 23 pairs of **chromosomes**. Chromosomes contain a person's genes.

Genes control traits like hair color and eye color and are passed down from generation to generation.

Gene mutations are the cause of Noonan syndrome. A gene mutation is a permanent change in the makeup of a gene. Half of all cases of Noonan syndrome are caused by a mutation of a gene contained in chromosome 12.

In some cases, this gene is passed down from a parent.

Sometimes a gene will just change on its own. This is called a spontaneous gene mutation.

Features of Noonan syndrome

Some of the physical traits and conditions that may occur in Noonan syndrome are:

- Short stature
- Heart defects
- Abnormal bruising or bleeding
- Unusual facial features
  - > Wide-set eyes
  - > Down-slanting eyes
  - > Drooping eyelids
  - > Arched eyebrows
  - > Low-set ears with forward-facing lobes
- Delay in puberty
- Poor eyesight
- Chest and skeletal deformities
- Webbed neck
- A low hairline at the back of the neck
- Learning disability or mild retardation
- Lymphatic abnormalities
- Undescended testicles in males at birth
- Poor weight gain in infancy
- Brilliant blue or blue-green eyes
- Feeding difficulties in infancy
- Frequent or forceful vomiting in infancy
- Hearing problems
- Poor muscle tone in early development

Some children may have many of these features, while others have just a few.

As children grow older, facial signs of Noonan syndrome may begin to fade.

Approximately 50% to 70% of patients with Noonan syndrome have short stature. In children with Noonan syndrome, puberty can be delayed by about 2 years. The **pubertal growth** spurt is often reduced or absent. Bone development may also be delayed by 2 years.

The average final height for patients with Noonan syndrome is generally less than that of the general population.

Diagnosing Noonan syndrome

At present, there is no simple test that can diagnose a boy or girl with Noonan syndrome. At first, the diagnosis is made by physical features of Noonan syndrome being recognized. More commonly, this may be short stature, heart problems, and certain characteristic facial features. Afterward, a genetic test may be done. It is helpful if doctors know of any family members who have been diagnosed with Noonan syndrome.

Whenever problems associated with Noonan syndrome arise, it may be a good idea for children being treated by pediatricians and pediatric cardiologists to be sent to an endocrinologist for evaluation. After these symptoms are discovered, a **geneticist** may confirm the diagnosis with genetic testing.

Growth charts

Growth charts are used to track a child's height and weight. Charts are also used to compare a child's height and weight with the statistical norm. This is the average height and weight of other children who are the same sex and age. Separate growth charts are used for boys and girls.

Each chart has lines called percentile curves, or percentiles. These lines represent the percentage of children at the same height or weight for that age group.

A percentile is a way to show ranking. For example, if a 10-year-old girl is in the 50th percentile for height, that means 50% of 10-year-old girls are taller and 50% are shorter than she is.

On the other hand, if a 2-year-old boy is in the 5th percentile for height, that means 95% of 2-year-old boys are taller and 5% are shorter than he is.

Growth charts and Noonan syndrome

With Noonan syndrome, a child's weight and length may be fairly normal at birth. But during childhood they may fall behind in height, weight, and bone age or maturation compared with the general population. Because they may grow differently than other children with short stature, special charts are available for boys and girls with Noonan syndrome.

To record your child's growth, the doctor may draw a line connecting height measurements for your child at several points. When a child with short stature due to Noonan syndrome has information plotted on a growth chart, the curve generally follows on or below the 3rd percentile for height and weight.

This growth curve is made by comparing growth of a child with those of both children with and without Noonan syndrome.

Treatment of children with Noonan syndrome

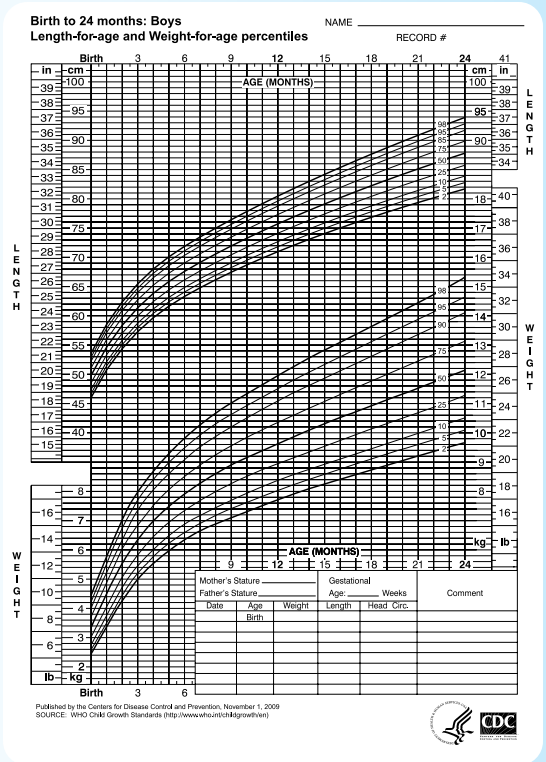
Treatment for Noonan syndrome depends on the individual child's symptoms. There are treatment options available that can successfully treat children with Noonan syndrome. To learn more about specific treatment options for your child, speak with your pediatrician, pediatric endocrinologist, or other pediatric specialist.

Follow-up visits

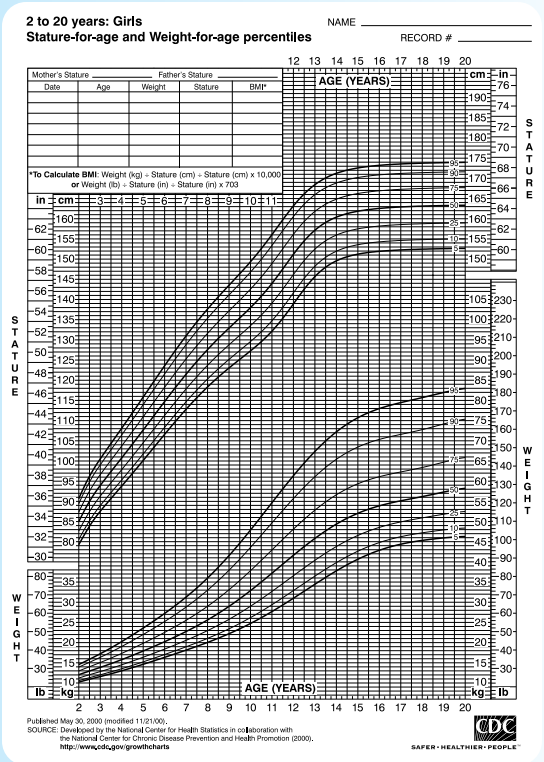
After diagnosing your child with Noonan syndrome, the endocrinologist or other pediatric specialist will likely ask you for regular follow-up visits. The endocrinologist will monitor your child's growth and other conditions.

In addition to watching your child's growth, other conditions associated with Noonan syndrome may need to be monitored. Page 4 lists the known conditions and features associated with Noonan syndrome. These may include:

- Congenital heart disease
- Blood clotting disorders
- Swelling in the lymph system



Growth chart for boys from birth to 24 months



Growth chart for girls aged 2 to 20 years

Supporting your child

The self-esteem of children has much to do with how they see their bodies. Children with Noonan syndrome may face social challenges because of their condition. Some may be teased by other children. This may be hurtful.

Remind your child that a person's worth has nothing to do with height. But it has everything to do with who that person is.

Talk to your doctors or see page 13 to learn more about support groups for Noonan syndrome.

Insurance

Questions about coverage

If your child is prescribed treatment, you may have questions about insurance coverage. There are programs that may help you get the assistance you need. In many cases, assistance is just a phone call away. The manufacturer of your child's treatment may be able to answer your questions about insurance coverage and reimbursement, and may be able to help you find additional coverage to ensure that your child's treatment continues as prescribed.

Use these lines for questions you may have for the doctor, and notes from your conversation.

ADDITIONAL RESOURCES

You can find information and support from these organizations:

The MAGIC Foundation®

6645 W. North Avenue
Oak Park, IL 60302
Tel: 1-800-3MAGIC3 (362-4423)
magicfoundation.org

Noonan Syndrome Community
A Division of the RASopathies Foundation

Tel: 1-866-875-8928
Email: info@teamnoonan.org
www.teamnoonan.org

GLOSSARY

Here are definitions for some words in this brochure that may be new to you. If there are other words that you need to understand better, your child's doctor or nurse can help you.

Chromosomes
Thread-like strands that contain a person's genes.

Congenital heart disease
A malformation of the heart or large blood vessels near the heart that people are born with.

Endocrinologist
A specially trained doctor who diagnoses and treats diseases of the glands and hormone imbalances.

Genetic mutation
A permanent change in the makeup of a gene. This may result in a trait or feature not found in relatives.

Geneticist
A scientist who studies genes. Genes are the functional units on a chromosome that transmit characteristics from parents to children.

Pubertal growth
The growth period during sexual maturity.

Stature
The standing height of a person.

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