

Noonan Syndrome



Knowledge
to grow by



Noonan syndrome is named for Dr. Jacqueline Noonan, a pediatric cardiologist, who first identified it in 1963. Noonan syndrome is caused by a **genetic mutation** that may cause **congenital heart disease**, distinctive facial features, short stature, and other conditions.

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

Noonan syndrome may present as short stature in up to 83% of patients. Features appear at birth or during early childhood. Boys and girls are affected equally. It is estimated that 1 in 1000 to 2500 infants are 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

If there is a medical condition, the doctor can suggest medicines that may help your child grow.

Children with Noonan syndrome usually have more than one medical condition. This means 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 or eye color and are passed down from generation to generation.

Gene mutation causes 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 many of these cases, this gene is passed down from a family member.

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

Features of Noonan syndrome

Some of the common physical traits and conditions of Noonan syndrome are:

- Short stature
- Heart defects
- Increased 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
- Frequent or forceful vomiting
- 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.

Short stature is one of the main features of Noonan syndrome. It affects up to 83% of children diagnosed. In children with Noonan syndrome, puberty can be delayed by about 2 years. The **pubertal growth** spurt is often reduced or absent. Bone development is also delayed by 2 years.

The average final height for patients with Noonan syndrome is generally less than the normal 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 unusual facial features. Afterwards a genetic test may be done. It is helpful if doctors know of any family members that 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.

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.



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 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 will 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 third percentile for height and weight.

This growth curve is made by comparing the child with children without Noonan syndrome.

Treatment of short stature in Noonan syndrome

Treatment with growth hormone may be beneficial in treating short stature caused by Noonan syndrome.

Treatment for short stature in Noonan syndrome is done through daily injections of synthetic (man-made) growth hormone that the body processes.

This is growth hormone produced in a lab. It is identical to human growth hormone. Human growth hormone is made naturally by a person's body.



Response to treatment depends on the following factors:

- Child's age at the beginning of treatment
- How long treatment is given
- Compliance with treatment

Follow-up visits

After diagnosing your child with Noonan syndrome, the endocrinologist will likely ask you for regular follow-up visits. He or she will monitor your child's growth to make sure growth hormone treatment is working well. If needed, the doctor will adjust the dose of growth hormone to ensure the best results.



In addition to watching your child's growth, other conditions associated with Noonan syndrome will 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

Supporting your child

The self-esteem of children with short stature has much to do with how they see their bodies. Children who feel good about themselves and who feel loved by their families may feel fine about their short stature.

But many children may face social challenges because of their height. Some may be teased by taller children about their small size. This may be hurtful.

For these reasons, please remember to support your child. Let your child know that any problems will pass. Treatment with growth hormone therapy may help him or her grow.

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 about support groups for Noonan syndrome.

GLOSSARY

Here are definitions for some words in this brochure that may be new to you. If there are other words 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.

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.


Lymphatic abnormalities

A malformed, sponge-like collection of channels and cystic spaces that contain clear fluid.

Pubertal growth

The growth period when a child becomes a young adult.

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