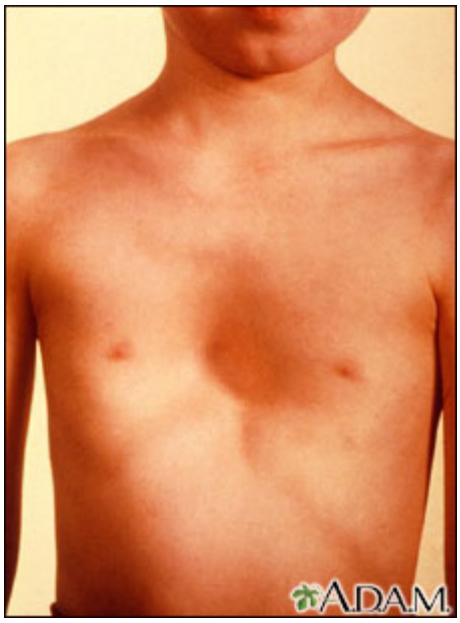
Noonan syndrome

Definition



Noonan syndrome is a disease that can be passed down through families (inherited). It causes many parts of the body to develop abnormally.

Causes

Noonan syndrome is linked to defects in several genes. In general, certain proteins involved in growth and development become overactive as a result of these gene changes.

Noonan syndrome is an autosomal dominant condition. This means only one parent has to pass down the nonworking gene for the child to have the syndrome. However, some cases may not be inherited.

Symptoms

Symptoms include:

- Delayed puberty
- Down-slanting or wide-set eyes
- Hearing loss (varies)
- Low-set or abnormally shaped ears
- Mild intellectual disability (only in about 25% of cases)
- Sagging eyelids (ptosis)
- Short stature
- Small penis
- Undescended testicles
- Unusual chest shape (most often a sunken chest called pectus excavatum)
- Webbed and short-appearing neck

Exams and Tests

The health care provider will perform a physical exam. This may show signs of heart problems the infant had from birth. These may include pulmonary stenosis and atrial septal defect.

Tests depend on the symptoms, but may include:

- Platelet count
- Blood clotting factor test
- ECG, chest x-ray, or echocardiogram
- Hearing tests
- Growth hormone levels

Genetic testing can help diagnose this syndrome.

Treatment

There is no specific treatment. Your provider will suggest treatment to relieve or manage symptoms. Growth hormone has been used successfully to treat short height in some people with Noonan syndrome.

Support Groups

The Noonan Syndrome Foundation is a place where people dealing with this condition can find information and resources.

Possible Complications

Complications may include:

- Abnormal bleeding or bruising
- Buildup of fluid in tissues of body (lymphedema, cystic hygroma)
- Failure to thrive in infants
- Leukemia and other cancers
- Low self-esteem
- Infertility in males if both testes are undescended
- Problems with the structure of the heart
- Short height
- Social problems due to physical symptoms

When to Contact a Medical Professional

This condition may be found during early infant exams. A geneticist is often needed to diagnose Noonan syndrome.

Prevention

Couples with a family history of Noonan syndrome may want to consider genetic counseling before having children.

References

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