

SHPRINTZEN-GOLDBERG SYNDROME

Shprintzen-Goldberg syndrome is a disorder that affects many parts of the body. People who have this syndrome have a combination of unique facial features, bone abnormalities, and brain deficiencies. Shprintzen-Goldberg syndrome is caused by genetic changes (mutations) in a gene that contributes to the formation of connective tissue.

Fewer than 50 cases of Shprintzen-Goldberg syndrome have been reported worldwide.



What other names do people use for Shprintzen-Goldberg syndrome?

Shprintzen-Goldberg syndrome (SGS) is also referred to as Marfanoid-craniosynostosis syndrome and Shprintzen-Goldberg craniosynostosis syndrome.

How prevalent is Shprintzen-Goldberg syndrome?

Shprintzen-Goldberg syndrome is an extremely rare disorder, but the exact prevalence is unknown. Fewer than 50 cases of Shprintzen-Goldberg syndrome have been reported to date worldwide.

What are the characteristics of Shprintzen-Goldberg syndrome?

Shprintzen-Goldberg syndrome can affect all parts of the body. A common feature is craniosynostosis, a premature fusion of certain skull bones (skull bones join together too early). This early joining together of the skull bones can impair skull growth.

Facial features of Shprintzen-Goldberg syndrome include:

- A long, narrow head (dolichocephaly)
- High prominent forehead
- Widely spaced eyes (hypertelorism)
- Protruding or bulging eyes (exophthalmos, ocular proptosis)
- Wandering eye (strabismus)
- Outside corners of the eyes point downward (down-slanting palpebral fissures)
- A high, narrow palate (roof of the mouth)
- Under-developed jaw bones (maxillary hypoplasia)
- Small lower jaw (micrognathia)
- Low-set ears that are rotated backward
- Increased angle of the eyelids (telecanthus)

Features related to the heart and blood vessels include:

- Mitral valve prolapse
- Mitral and aortic valve regurgitation
- Aortic root enlargement

Brain abnormalities include:

- Brain anomalies, including hydrocephalus (water on the brain)
- Dilatation (enlargement) of the lateral ventricles
- Chiari 1 malformation (brain tissue protrudes into the spinal canal)

Other characteristics include:

- One or more fingers are permanently bent (camptodactyly)
- Permanent bending of a muscle or joint (contractures)
- Unusually large range of joint movement (joint hypermobility)
- Weak muscle tone (hypotonia) in infancy
- A soft out-pouching around the belly-button (umbilical hernia) or lower abdomen (inguinal hernia)
- Flat feet (pes planus)

Shprintzen-Goldberg syndrome has several skeletal features that are similar to Marfan syndrome.

Shprintzen-Goldberg syndrome also shares many features with Loeys-Dietz syndrome, including all of the typical craniofacial and skeletal findings.

In addition, people with Shprintzen-Goldberg syndrome often have delayed development and mild to moderate intellectual disability (neurologic anomalies). This is rarely seen in Loeys-Dietz syndrome and it is not associated at all with Marfan syndrome.

What causes Shprintzen-Goldberg syndrome?

Most cases of Shprintzen-Goldberg syndrome are caused by a change (mutation) in the SKI gene.

This gene affects many cell types throughout the body and appears to play a role in the development of many tissues, including the skull, other bones, skin, and brain.

Rarely, a change in the FBN1 gene (fibrillin-1), which causes Marfan syndrome, can cause a condition that resembles Shprintzen-Goldberg syndrome. Other genes also may be involved in this condition, and in some cases, the genetic cause is unknown.

Shprintzen-Goldberg syndrome is an autosomal dominant disorder. This means that if one parent has the disorder, an altered copy of the gene can be inherited from the affected parent. Each child of someone with Shprintzen-Goldberg syndrome has a 50 percent chance of inheriting the abnormal gene and the condition.

However, most people with Shprintzen-Goldberg syndrome are the first in their family to have the condition. This means it was the result of a spontaneous mutation.

It is possible for unaffected parents to have more than one child with Shprintzen-Goldberg syndrome if one of the parents has a SKI mutation in a population of cells in the ovary or testes, but not in other cells throughout the body. This situation (called germ-line mosaicism) is very rare.

How is Shprintzen-Goldberg syndrome diagnosed?

The diagnosis of Shprintzen-Goldberg syndrome is made after a thorough examination and the identification of certain craniofacial, skeletal, cardiovascular, neurologic features, and brain anomalies. A gene test is available to test for a change in the SKI gene, the one gene known to date to cause this syndrome. However, a person can have Shprintzen-Goldberg syndrome and not test positive for the SKI gene.

How is Shprintzen-Goldberg syndrome managed?

Management of Shprintzen-Goldberg syndrome depends on the extent of the features. To evaluate severity, the following tests are recommended:

- Skeletal survey, including skull series
- Brain MRI
- Echocardiogram by a cardiologist
- Surgical evaluation for hernia repair, if a hernia is suspected
- Eye evaluation by an ophthalmologist

Treatments include:

- Surgical correction of craniofacial or chest problems are sometimes necessary or desirable.
- Shunting (surgical placement of a shunt to drain the accumulated fluid in the brain to the abdominal cavity to relieve pressure) may be required for patients with hydrocephalus (water on the brain).

- Orthopedic devices may be required for scoliosis or other bone problems.
- Surgical repair of abdominal hernias.
- Physiotherapy to increase mobility for those with joint contracture.

Anyone with Shprintzen-Goldberg syndrome should be managed by a cardiologist who is familiar with the condition.

Genetic counseling is recommended for families and individuals who have been diagnosed with Shprintzen-Goldberg syndrome.

What is the life expectancy for someone with Shprintzen-Goldberg syndrome?

Shprintzen-Goldberg syndrome does not alter lifespan, although complications from associated abnormalities, such as mental retardation or respiratory problems, may be expected.

Do you have questions? Would you like more information?

- Call our help center, 800-862-7326, ext. 126 to speak with a nurse who can answer your questions and send you additional information.
- Visit our website at marfan.org. You can print information that interests you and ask questions online.