Shprintzen-Goldberg syndrome

Shprintzen-Goldberg syndrome is a very rare disease that is congenital. It is often considered as *Marfan syndrome*, due to its many similar disorders, such as: **long limbs**, **scoliosis**, **abnormalities of the heart**, and **mental disorders**.

Occurence of the syndrome

This disease was discovered in 1979 by American doctors *Robert F. Shprintzen* and *Rosalie B. Goldberg*. It is a congenital disorder that is often confused with Shprintzen syndrome. It is a very rare syndrome, which occurs at a ratio of 1:1,000,000. The disorder is inherited in an autosomal dominant manner. It is a partial mutation in the SKI gene on chromosome 1, locus p36.33-p36.32 or FBN on chromosome 15 and locus q21.1.

Diagnosis

The diagnosis is based on clinical findings. Only a genetic examination, which detects the presence of a gene mutation, will definitely confirm this disease.

Symptoms

The symptoms can be visible during pregnancy on an ultrasound, where we can observe an elongated head (due to an enlarged fontanel).

- Premature fusion of cranial sutures (the skull cannot grow symmetrically)
- Long and thin face shape
- Prominent eyebrows, eyes are wide apart
- Exophthalmos
- Wide bridge of the nose
- Slightly raised mouth
- Marfanoid tongue
- Skeletal malformations
- Abnormalities of the cardiovascular system
- Hypotonia
- Omphalocele
- Hypoplasia
- Arachnodactyly

Treatment

The treatment is dependent on the symptoms. A tracheostomy is performed for better breathing and due to the development of sucking reflex, a tube is sometimes inserted.

References

Related articles

- Marfan syndrome
- DiGeorge syndrome

Used literature

- SADLER, T.W. *Langman's Medical Embryology.* 10. edition. vydavatel, 2006. ISBN 978-0-7817-9485-5.
- MUNTAU, Ania. *Pediatrie.* 2. edition. Grada, 2014. ISBN 978-80-247-4588-6.

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