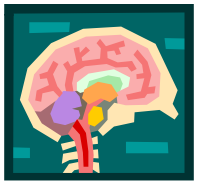

Disability Awareness Begins With You: Robinow Syndrome

Robinow Syndrome is a genetic disorder that causes abnormal development of the skeleton, face, and genitalia. It is also known as Fetal Facies Syndrome and Robinow Dwarfism due to its characteristic facial features and short stature.

This syndrome affects almost all ethnic groups, but it is less common in the Japanese and Afro-Caribbean populations. Robinow Syndrome occurs in approximately 1 out of every 500,000 people and affects males and females equally.

People with Robinow Syndrome will have several of the following symptoms.

- Short stature
- Hemivertebrae - vertebrae (bones of the spine) are only half the normal size
- Mesomelic shortening (shortening of the middle bones of the arms and legs, especially the forearms) •
- Genital hypoplasia (underdevelopment of the genitals)
- females: small size of the clitoris and labia minora
- males: small penis with normal-sized scrotum and testes
- Macrocephaly (large head)
- Prominent eyes with hypertelorism (increased distance between the eyes)
- Broad and prominent forehead
- Depressed or flat nasal bridge, upturned nose
- Upper lip has an appearance of an inverted “V”
- Midline cleft of the lower lip
- Overgrowth of the gums, irregular or crowded teeth
- Ankyloglossia (shortened frenulum, which connects the tongue to the floor of the mouth and gives the appearance of “tongue-tie”) •
- ow-set ears
- Brachydactyly (shortened fingers and toes) with small nails
- congenital heart defects
- Mental retardation (20% of cases)

Possible Causes

Robinow Syndrome can be inherited in both an autosomal dominant and autosomal recessive manner. In the autosomal dominant type, the child inherits a mutated or altered gene from one parent and this one change is enough to cause the disease. It is not known which gene causes this form of Robinow Syndrome. A gene named ROR2, which is located on chromosome 9, has been identified as the gene that causes the autosomal recessive form of Robinow Syndrome. In this form, a child receives a mutated copy of the gene from each parent and two mutated copies are needed to cause disease. The autosomal recessive type has been shown to have more severe symptoms than the autosomal dominant type. For example, height is either normal or slightly shorter in the autosomal dominant type, but height is greatly affected in the autosomal recessive type.

Diagnosis

The diagnosis of Robinow Syndrome is based on clinical examination which shows the physical characteristics described above, identification of familial inheritance and genetic testing. prenatal diagnosis of Robinow Syndrome can be made through fetal ultrasound examination. The length of the long bone and the ulna (lower arm bone) to humerus (upper arm bone) ratio is especially useful in prenatal diagnosis.

Treatment

Children diagnosed with Robinow Syndrome may require extensive surgical care for skeletal abnormalities. Additionally, orthotics or braces may be needed to help with walking and to stabilize the arms and legs. Orthodontic care may be needed if the teeth are crowded together. In a few cases, facial reconstructive surgery may be needed. The autosomal recessive form of this disorder is associated with poor growth during the pregnancy and this continues after birth. Boys may have low testosterone levels, which causes poor growth and underdevelopment of the genitalia. Hormone therapy with testosterone and/or human chorionic gonadotropin will help increase growth. For both boys and girls, therapy with recombinant human growth hormone has been shown to increase growth rates. Approximately 10-15% of cases involve heart defects that are present at birth. The type of heart defects associated with this disorder can be very serious so children who have heart defects need to be followed very closely by a cardiologist. Surgical intervention may be needed.

Prognosis

The prognosis of Robinow Syndrome is variable due to the wide range of symptoms. Children must be checked for congenital heart defects immediately after birth since it is the major cause of death within the first few years of life. Mental retardation or developmental delay occurs in about 10-20% of the cases. However, most children will have normal intelligence. Those diagnosed with the autosomal recessive type will have more severe symptoms than those with the autosomal dominant type, but surgical intervention and hormone therapy will greatly help with the prognosis. Overall, the prognosis is good. There are many treatments and therapies available to help relieve symptoms of Robinow Syndrome.

Connect with other parents

In the spirit of community and support, Madisons Foundation offers the unique service of connecting parents of children with rare diseases. If you would like to be connected to other parents of children with this disease, please fill out this brief form.

Weblinks

Robinow Syndrome Foundation:
A great site created by families of children with Robinow Syndrome.
Children’s Craniofacial Association : This organization provides information and support to individuals and families affected with craniofacial abnormalities.
Little People of America:
Provides education and peer support for people of short stature.
MAGIC Foundation:
A foundation that provides information about diseases and syndromes that affect children’s growth.
Google Search for Robinow Syndrome

References/Sources: Hosalkar HS, Gerardi J, Shaw BA (2002). Robinow syndrome. Journal of Postgraduate Medicine. 48, p. 50-51. Patton MA, Afzal AR (2002). Robinow Syndrome. Journal of Medical Genetics. 39, p. 305-310. Sleesman JB, Tobias JD (2003). Anaesthetic implications of the child with Robinow syndrome. Paediatric Anaesthesia. 13, p. 629–632.

http://www.madisonsfoundation.org/index.php/component/option,com_mpower/