Aarskog-Scott syndrome

Aarskog-Scott syndrome is a genetic disorder that affects the development of many parts of the body. This condition mainly affects males, although females may have mild features of the syndrome.

People with Aarskog-Scott syndrome often have distinctive facial features, such as widely spaced eyes (hypertelorism (https://en.wikipedia.org/wiki/Hypertelorism)), a small nose, a long area between the nose and mouth (philtrum), and a widow's peak hairline (https://en.wikipedia.org/wiki/Widow%27s_peak). They frequently have mild to moderate short stature during childhood, but their growth usually catches up during puberty. Hand abnormalities are common in this syndrome and include short fingers (brachydactyly (https://en.wikipedia.org/wiki/Brachydactyly)), curved pinky fingers (fifth finger clinodactyly (https://en.wikipedia.org/wiki/Clinodactyly)), webbing of the skin between some fingers (syndactyly (https://en.wikipedia.org/wiki/Syndactyly)), and a single crease across the palm. Some people with Aarskog-Scott syndrome are born with more serious abnormalities, such as heart defects or a cleft lip with or without an opening in the roof of the mouth (cleft palate (https://en.wikipedia.org/wiki/Cleft_lip_and_cleft_palate#Cleft_palate)).

Most males with Aarskog-Scott syndrome have a shawl scrotum, in which the scrotum surrounds the penis. Less often, they have undescended testes (cryptorchidism) or a soft out-pouching around the belly-button (umbilical hernia) or in the lower abdomen (inguinal hernia).

The intellectual development of people with Aarskog-Scott syndrome varies widely among affected individuals. Some may have mild learning and behavior problems, while others have normal intelligence. In rare cases, severe intellectual disability has been reported.

How common is Aarskog-Scott syndrome?

Aarskog-Scott syndrome is believed to be a rare disorder; however, its prevalence is unknown because mildly affected people are often not diagnosed.

What genes are related to Aarskog-Scott syndrome?

Mutations in the FGD1 gene cause some cases of Aarskog-Scott syndrome. The FGD1 gene provides instructions for making a protein that turns on (activates) another protein called Cdc42, which transmits signals that are important for various aspects of embryonic development.

Mutations in the FGD1 gene lead to the production of an abnormally functioning protein. These mutations disrupt Cdc42 signaling, which causes the wide variety of developmental abnormalities seen in Aarskog-Scott syndrome.

Only about 20 percent of people with this disorder have identifiable mutations in the FGD1 gene. The cause of Aarskog-Scott syndrome in other affected individuals is unknown.

Read more about the FGD1 gene.

How do people inherit Aarskog-Scott syndrome?

Aarskog-Scott syndrome is inherited in an X-linked recessive pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause Aarskog-Scott syndrome. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. Females who carry one altered copy of the FGD1 gene may show mild signs of the condition, such as hypertelorism, short stature, or a widow's peak hairline. A striking characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

Where can I find information about treatment for Aarskog-Scott syndrome?

These resources address the management of Aarskog-Scott syndrome and may include treatment providers.

- MedlinePlus Encyclopedia: Aarskog syndrome

You might also find information on treatment of Aarskog-Scott syndrome in Educational resources and Patient support.

Where can I find additional information about Aarskog-Scott syndrome?

You may find the following resources about Aarskog-Scott syndrome helpful. These materials are written for the general public.

- MedlinePlus - Health information (4 links)
- Educational resources - Information pages (2 links)
- Patient support - For patients and families (4 links)

You may also be interested in these resources, which are designed for healthcare professionals and researchers.

- PubMed - Recent literature
- OMIM - Genetic disorder catalog

What other names do people use for Aarskog-Scott syndrome?

- Aarskog syndrome
- AAS
- Facio-digito-genital dysplasia
- Faciogenital dysplasia
- See How are genetic conditions and genes named? in the Handbook.

External links

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This article has been revived (https://web.archive.org/web/20130305083150/http://wiki.medpedia.com/Aarskog-Scott_syndrome) from the former medical wiki **Medpedia (https://en.wikipedia.org/wiki/Online_medical_wiki_encyclopedias#Medpedia)**.