



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), a small nose, a long area between the nose and mouth (philtrum), and a widow's peak hairline. 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), curved pinky fingers (fifth finger clinodactyly), webbing of the skin between some fingers (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).

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.

Frequency

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

Genetic Changes

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.

Inheritance Pattern

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 characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

Other Names for This Condition

- Aarskog syndrome
- AAS
- Facio-digito-genital dysplasia
- Faciogenital dysplasia

Diagnosis & Management

These resources address the diagnosis or management of Aarskog-Scott syndrome:

- Genetic Testing Registry: Aarskog syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0175701/>
- MedlinePlus Encyclopedia: Aarskog syndrome
<https://medlineplus.gov/ency/article/001654.htm>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Aarskog syndrome
<https://medlineplus.gov/ency/article/001654.htm>
- Health Topic: Cleft Lip and Palate
<https://medlineplus.gov/cleftlipandpalate.html>
- Health Topic: Congenital Heart Defects
<https://medlineplus.gov/congenitalheartdefects.html>
- Health Topic: Learning Disorders
<https://medlineplus.gov/learningdisorders.html>

Genetic and Rare Diseases Information Center

- Aarskog syndrome
<https://rarediseases.info.nih.gov/diseases/4775/aarskog-syndrome>

Educational Resources

- Disease InfoSearch: Aarskog Syndrome
<http://www.diseaseinfosearch.org/Aarskog+Syndrome/58>
- MalaCards: aarskog-scott syndrome
http://www.malacards.org/card/aarskog_scott_syndrome
- Orphanet: Aarskog-Scott syndrome
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=915

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/aarskog-syndrome/>
- Resource list from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/aarskog.html>

Genetic Testing Registry

- Aarskog syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0175701/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?term=%22Aarskog-Scott+syndrome%22+%5BDISEASE%5D+OR+NCT00916903+%5BID-NUMBER%5D>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28aarskog-scott+syndrome%5BTIAB%5D%29+OR+%28aarskog+syndrome%5BTIAB%5D%29+OR+%28faciogenital+dysplasia%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- AARSKOG-SCOTT SYNDROME
<http://omim.org/entry/305400>

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- OMIM: AARSKOG-SCOTT SYNDROME
<http://omim.org/entry/305400>
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