

Aarskog Syndrome: Genetic and Rare Disease

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Editorial

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

Aarskog syndrome is a rare genetic condition which is characterized by short stature and multiple facial, limb and genital abnormalities. Additionally, some types of cognitive disorders may occasionally be present. The *FGD1* gene on the X chromosome is the only gene known to be associated with Aarskog syndrome.

Aarskog-Scott syndrome is believed to be a rare disorder. Its prevalence is unknown because mildly affected people may not be diagnosed. Mutations in the *FGD1* gene are the known genetic cause of Aarskog-Scott syndrome. The *FGD1* gene provides instructions for making a protein that turns on another protein called *Cdc42*, which transmits signals which are important for various aspects of development before and after birth.

Aarskog syndrome primarily affects males. Affected boys exhibit a characteristic set of facial, skeletal, and genital abnormalities. Clinical signs may vary from person to person, even within families. Males with Aarskog syndrome have a rounded face with a broad forehead. Additional characteristic facial features includes ocular hypertelorism, ptosis, palpebral fissures, a small nose with nostrils that are flared forward, maxillary hypoplasia, and a widow's peak. Female carriers often show some minor manifestations of the disorder

in the face and hands. Penoscrotal transposition is a rare abnormality of the external genitalia in which the scrotum is malpositioned superior to the penis.

Mutations in the *FGD1* gene lead to the production of an abnormally functioning protein. These mutations disrupt *Cdc42* signaling, leading to the wide variety of abnormalities that occur in people with Aarskog-Scott syndrome. Similar to all genetic diseases Aarskog-Scott syndrome cannot be cured, although numerous treatments exist to increase the quality of life.

Surgery may be required to correct some of the anomalies, and orthodontic treatment which may be used to correct some of the facial abnormalities. Trials of growth hormone have been effective to treat short stature in this disorder.

Aarskog-Scott syndrome (AAS) or Faciogenital Dysplasia (FGD) is an X-linked syndrome with a recessive mode of inheritance. This condition is characterized by a distinguishing combination of short stature, genital, facial and skeletal anomalies. The latter include brachydactyly, short nose, hypertelorism, syndactyly and shawl scrotum. Additional features include mental retardation, joint hyperextensibility and ptosis. Clinical presentations of patients vary to a great extent which impedes reaching a clear-cut diagnosis.

Genetic and biochemical analyses shows that *FGD1* encodes a Guanine Nucleotide Exchange Factor (GEF), or activator, for *Cdc42*, a member of the Rho family of Raslike GTPases. Rho proteins comprises a family of at least eight distinct proteins which are involved in the control of a wide variety of cellular functions, including the organization of the actin cytoskeleton, the control of cellular division and the transcriptional regulation of gene expression.

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