CONDITION

Aarskog syndrome

Learn about Aarskog syndrome, including its genetic cause, common facial, growth, hand, and genital features, diagnosis, treatment, and follow-up.

WHAT IT IS
Rare genetic condition affecting growth and development

BODY SYSTEM
Growth and development

MAIN SYMPTOMS
Short stature, distinctive facial features, hand differences, genital differences

WHAT CAUSES IT
Inherited FGD1 gene variant

HOW IT IS DIAGNOSED
FGD1 genetic testing

MAIN TREATMENT
Supportive and symptom-based care

CAN IT BE CURED?
No, genetic change cannot be corrected

IS IT AN EMERGENCY?
Sometimes, painful firm groin or abdominal bulge

LONG-TERM OUTLOOK
Life span not generally shortened, depending on specific needs

Overview

Aarskog syndrome is a rare genetic condition that affects growth and development. It is also called Aarskog-Scott syndrome or faciogenital dysplasia. "Faciogenital" refers to differences involving the face and genitals.

The condition can affect facial appearance, height, hands and feet, bones, teeth, and genital development. Learning, attention, vision, and hearing may also be affected, although many people have typical intelligence and few medical problems.

Aarskog syndrome is usually caused by a change, also called a variant, in the FGD1 gene. This gene helps control signals involved in development before birth. The condition is inherited in an X-linked pattern, so it is often more noticeable in males. Females with an FGD1 variant may have mild features or no obvious signs.

The combination and severity of features vary widely, even among relatives. A person does not need to have every characteristic associated with the syndrome. Some people are diagnosed in childhood because of short stature, distinctive physical features, or developmental concerns. Others are diagnosed later, sometimes after another family member receives a diagnosis.

Aarskog syndrome is not usually a progressive disorder. This means that it does not typically cause continuing loss of physical or thinking abilities. However, individual concerns such as dental problems, an undescended testicle, hernias, or learning difficulties may need treatment.

Symptoms

Features may be present at birth, but some become easier to recognize as a child grows. Facial characteristics can also become less noticeable in adulthood.

Possible facial and head features include:

Growth is often slower than expected during childhood. Short stature may become more noticeable during the early years, and the adolescent growth spurt may occur later than usual. Head size may appear relatively large compared with height. Adult height varies, and not everyone with the condition is short.

Hand and foot differences may include short fingers, curved fifth fingers, mild webbing between fingers, or a single crease across the palm. Joints may be unusually flexible. Some people have broad feet or toes that are shaped differently.

Skeletal findings can include an indented or prominent breastbone, rounded shoulders, or curvature of the spine. These differences are often mild but may affect posture or comfort.

In males, genital findings may include a shawl scrotum , in which the scrotal skin surrounds the base of the penis. One or both testicles may not descend into the scrotum. This is called cryptorchidism. Less commonly, the opening of the urethra may be on the underside of the penis, a condition called hypospadias. Groin or umbilical hernias can also occur.

Females may have subtle facial, growth, or hand features. Because genital signs are usually absent and symptoms may be mild, the condition may be harder to recognize in females.

Development is variable. Some children have delayed speech, mild learning difficulties, attention problems, or challenges with social and behavioral regulation. Others have typical development and school performance. Vision problems such as crossed eyes, known as strabismus, may occur. Hearing concerns have also been reported.

- A broad or rounded forehead

- Widely spaced eyes
- Drooping upper eyelids, called ptosis
- A wide bridge of the nose or nostrils that point forward
- A long groove between the nose and upper lip
- A widow's peak hairline
- Ears that are folded or shaped differently
- Delayed growth of teeth or crowded teeth

Causes and risk factors

Aarskog syndrome is most often linked to a disease-causing variant in the FGD1 gene. The gene provides instructions for a protein involved in cell signaling and the development of bones and other tissues.

FGD1 is located on the X chromosome. Males usually have one X chromosome, so a disease-causing variant in their single copy of FGD1 can produce clearer features. Females usually have two X chromosomes. Their second working copy of the gene may reduce the effects, although some females have noticeable symptoms.

A female who carries an FGD1 variant can pass it to children. An affected male passes his X chromosome to all of his daughters and none of his sons. The exact chance for a particular family depends on who carries the variant and whether it was inherited or occurred for the first time in that person.

Not everyone with clinical features of Aarskog syndrome has an identifiable FGD1 variant. This may be because current tests cannot detect every relevant genetic change, or because another condition has similar features. A negative genetic test therefore does not always settle the diagnosis.

Aarskog syndrome is not caused by anything a parent did during pregnancy. Diet, exercise, parenting, and common childhood illnesses do not cause it. The main known risk factor is having a disease-causing FGD1 variant in the family, although a new variant can occur without a known family history.

Diagnosis

Diagnosis begins with a medical history and physical examination. A clinician may review

growth patterns, developmental milestones, school progress, and family history. The examination may look for the characteristic combination of facial, hand, skeletal, and genital findings.

Because no single physical sign confirms Aarskog syndrome, the overall pattern matters. A clinician may also consider other genetic conditions that can cause short stature, widely spaced eyes, developmental differences, or genital findings. Examples include Noonan syndrome, Robinow syndrome, and some chromosome conditions.

Genetic testing can look for an FGD1 variant. Testing may involve sequencing the gene and checking for larger missing or duplicated sections. In some situations, a multigene panel or a broader test such as exome sequencing may be considered, particularly when the features do not clearly point to one condition.

A positive result can confirm the diagnosis and may help with family testing. A negative result does not completely exclude Aarskog syndrome when the clinical features strongly support it.

Additional evaluations depend on the person's symptoms. These may include:

A clinical geneticist or genetic counselor can explain test choices, uncertain results, inheritance, and what a result may mean for relatives.

- Careful measurement of height, weight, and head size
- Developmental, speech, or educational assessment
- Eye or hearing evaluation
- Dental and orthodontic examination
- Examination for hernias or undescended testicles
- Assessment of the spine, chest, joints, and walking
- Other tests when the examination suggests a specific concern

Treatment and management

There is no treatment that corrects the underlying genetic change. Care focuses on the health and developmental needs of the individual.

Growth should be followed over time rather than judged from one measurement. If growth is much slower than expected, a clinician may look for other causes, such as nutrition problems, thyroid disease, or growth hormone deficiency. Growth hormone has been used in some affected children, but evidence specific to Aarskog syndrome is limited. Decisions about growth treatment require evaluation by a pediatric endocrinologist and should not be based on

the syndrome alone.

Early developmental support may help children with speech, movement, learning, attention, or behavior. Services can include speech-language therapy, occupational therapy, physical therapy, or an individualized school plan. Support should be based on the child's actual strengths and needs.

Eye problems such as strabismus or ptosis may be treated with glasses, patching, or surgery, depending on the cause and effect on vision. Hearing concerns should be evaluated because untreated hearing loss can affect speech and learning.

Dental care may address delayed tooth eruption, crowded teeth, an uneven bite, or missing teeth. Treatment can involve preventive dental care, orthodontics, or restorative work.

An undescended testicle should be assessed by an appropriate specialist. Surgery may be recommended to move it into the scrotum. Hypospadias and significant genital differences may also be evaluated by pediatric urology. Hernias can be repaired if they cause symptoms or are considered likely to create problems.

Musculoskeletal care may include monitoring of spinal curvature, posture, joint flexibility, and foot alignment. Physical therapy can improve strength or function when needed. Surgery is generally reserved for structural problems that cause significant symptoms or impairment.

Emotional support can be important, especially when short stature, facial appearance, genital differences, or learning needs affect self-esteem. Age-appropriate explanations and attention to privacy can help children participate in their care.

Complications

Many people with Aarskog syndrome have a good quality of life. Complications depend on the features present rather than the diagnosis itself.

Potential concerns include persistent dental crowding, vision problems, learning difficulties, spinal curvature, and discomfort related to joint or chest-wall differences. An undescended testicle requires follow-up because it carries health risks beyond Aarskog syndrome. Hernias can occasionally trap part of the intestine or other tissue.

A painful, firm groin or abdominal bulge that does not go back in, especially with vomiting or abdominal swelling, needs emergency assessment. Sudden severe scrotal or groin pain also requires urgent care because it may indicate twisting of a testicle or another time-sensitive problem. Trouble breathing, loss of consciousness, or other severe symptoms should be treated as an emergency.

Less urgent concerns-such as a change in growth rate, new school difficulties, worsening vision, dental crowding, or gradual spinal curvature-should be discussed during routine or prompt outpatient follow-up.

Aarskog syndrome itself is not generally associated with a shortened life span. Outlook is influenced by the person's specific medical and developmental needs.

Follow up

Follow-up should be individualized. Children may benefit from coordinated care involving primary care, clinical genetics, dentistry, eye care, and developmental or educational professionals. Endocrinology, urology, orthopedics, or other specialties may be added when a specific concern is present.

Routine visits may review growth, development, school progress, vision, hearing, dental health, puberty, and emotional well-being. Clinicians may also recheck for hernias, spinal curvature, or joint problems as the child grows.

Genetic counseling can help families understand test results and inheritance. It may also be useful when relatives are considering testing or when an affected person is planning a family.

As adolescents move into adult care, they should gradually receive information about their diagnosis, previous procedures, genetic test results, and any continuing health needs. Copies of genetic reports and specialist records can make this transition easier.

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