About Aarskog Syndrome

Aarskog syndrome is an extremely rare genetic disorder marked by stunted growth that may not become obvious until the child is about three years of age. Broad facial abnormalities, musculoskeletal and genital anomalies, and mild to moderate intellectual disability.

Signs & Symptoms

Aarskog syndrome primarily affects males who exhibit a characteristic set of facial, skeletal, and genital abnormalities. In some cases, female carriers may develop a mild form of the disorder. Symptoms may vary from case to case. Males with Aarskog syndrome often have a rounded face with a broad forehead. Additional characteristic facial features include widely spaced eyes (ocular hypertelorism), drooping (ptosis) of the eyelids, downwardly slanting eyelid folds (palpebral fissures), a small nose with nostrils that are flared forward (anteverted nares), an underdeveloped upper jawbone (maxilliary hypoplasia), and a widow's peak.

Affected individuals may also have an abnormally long groove in the upper lip (philtrum) and a broad nasal bridge. Affected individuals may also have a variety of abnormalities affecting the ears and teeth. Ear abnormalities include low-set ears and thickened, "fleshy" earlobes. Dental abnormalities include missing teeth at birth, delayed eruption of teeth, and underdevelopment of thehard outer covering of teeth (enamel hypoplasia). Males with Aarskog syndrome also develop characteristic malformations of the skeletal system including disproportionate short stature; broad, short hands and feet; short, stubby fingers (brachydactyly) with permanent fixation of the fifth fingers in a bent position (clinodactyly); abnormally extendible finger joints; and wide flat feet with bulbous toes.

In addition, affected individuals may have a sunken chest (pectus excavatum), protrusion of portions of the large intestine through an abnormal opening in the muscular lining of the abdominal cavity (inguinal hernia) and a prominent navel (umbilicus).
Approximately 50 percent of individuals with Aarskog syndrome have spinal abnormalities such as incomplete closure of the bones of the spinal column (spina bifida occulta), fusion of the upper bones of the spinal column (cervical vertebrae), and underdevelopment of the "peg-like" projection of the second cervical vertebra (odontoid hypoplasia).

Males with Aarskog syndrome develop genital abnormalities including an abnormal fold of skin extending around the base of the penis ("shawl" scrotum) and/or failure of one or both of the testes to descend into the scrotum (cryptorchidism).
In addition, the urinary opening (meatus) may be located on the underside of the penis (hypospadias)and the scrotum may appear clefted or divided (bifid scrotum).
Mild intellectual disability is a consistent feature of the disorder. In some cases, affected children may exhibit hyperactivity, fail to gain weight and grow at the expected rate (failure to thrive), and develop chronic respiratory infections.

Additional symptoms may occur less frequently including congenital heart defects; abnormal side-to-side curvature of the spine (scoliosis); additional pairs of ribs; incomplete closure of the roof of the mouth (cleft palate) and/or a vertical groove in the upper lip (cleft lip); mild webbing of the fingers; and a short neck with or without webbing. Additional eye abnormalities may be present including crossed eyes (strabismus), farsightedness (hyperopia), and paralysis of certain eye muscles (ophthalmopleg)

Diagnosis

A diagnosis of Aarskog syndrome is made based upon a thorough clinical evaluation, a detailed patient history and identification of characteristic findings. X-ray studies can identify distinctive characteristics of Aarskog syndrome and help to differentiate it from similar disorders.

Treatment

The treatment of Aarskog syndrome is directed toward the specific symptoms that are apparent in each individual.
Treatment may require the coordinated efforts of a team of specialists. Paediatricians, surgeons, cardiologists, dental specialists, speech pathologists, specialists who assess and treat hearing problems (audiologists), eye specialists, and other healthcare professionals may need to systematically and comprehensively plan an affected child's treatment.
Surgery may be necessary to treat specific congenital or structural malformations sometimes associated with Aarskog syndrome. Individuals with Aarskog syndrome should receive complete eye and dental evaluations.