Case Report

Aarskog-Scott syndrome a very rare congenital malformation

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Abstract

Aarskog-Scott syndrome, also known as faciogenital dysplasia. It is an X-linked disorder characterized by short stature, hypertelorism, ptosis, shawl scrotum, and brachydactyly. Although there is wide phenotypic variability and other features, such as joint hyperextensibility, short nose, widow's peak hairline, and inguinal hernia. Most patients do not have mental retardation, but some may have neurobehavioral features. Carrier females may present with subtle features, such as widow's peak or short stature. Similar to all genetic diseases Aarskog–Scott syndrome cannot be cured, although numerous treatments exist to increase the quality of life.

Keywords: Faciogenital, Aarskog, hypertelorism, brachydactyly, short stature, ptosis.

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INTRODUCTION

It is faciogenital dysplasia caused by mutation in a gene called FGDY1 in band p11.21 on the X chromosome.[4] The syndrome is named for Dagfinn Aarskog, a Norwegian paediatrician and human geneticist who first described it in 1970, and Charles Scott Jr., an American medical geneticist who independently described the syndrome in 1971. Arskog-Scott syndrome is a genetic disorder that affects the development of many parts of the body, Characterised by hypertelorism, a small nose, long philtrum, and a widow's peak hairline. They have mild to moderate short stature. Hand abnormalities are common in this syndrome and include brachydactyly, clinodactyly, 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 cleft palate. Most males with Aarskog-Scott syndrome have a shawl scrotum, Less often, they have undescended testes, umbilical hernia, 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.[5]

CASE REPORT

14 years male child brought by parents with complaint of unilateral drooping of eyelid, Undescended testes and dismorphic limbs since birth. In birth history, antenatal and postnatal history uneventful. Family history revealed second degree consanguenious marriage, presence of similar feartures to the 12 years old male sibling, mother is having unilateral ptosis with dismorphic extremities and short stature. On examination, He had short stature, left sided ptosis, crowding of teeth, scoliosis, contracture of hands and foot, undescended testes. The child is developmentaly delayed with average performance at school. Younger sibling is having mild developmental delay and CTEV. On systemic examination-per abdomen-no organomegaly, CVS- s1s2 WNL, RS- air entry equal on both sides, CNS-child conscious, oriented with gait abnormality due to contracture.

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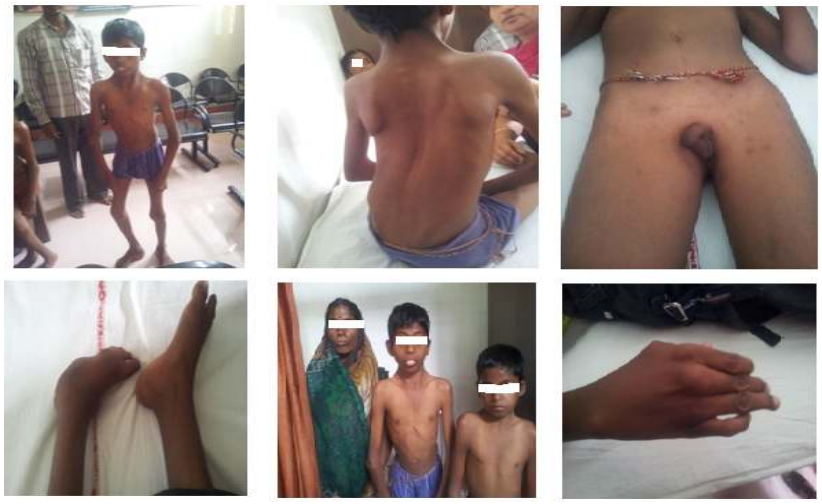

Figure 1:

Investigations

Routine blood investigations are within normal limit. X ray shows scoliosis of thoracic spine, USG abdomen shows atrophied testes in inguinal region.Genetic analysis shows mutation in FGDY1 in band p11.21 on the X chromosome.

DISCUSSION

It is faciogenital dysplasia, a very rare congenital malformation. Up till now less than 2oo cases reported in the world. Aarskog in 1970 described an X-linked disorder characterized by ocular hypertelorism, anteverted nostrils, broad upper lip, and undescended testis. These findings are seen in our case. Fernandez described 10 Japanese patients with Aarskog syndrome from 3 families. One of these patients had pulmonary stenosis, and another had ventricular septal defect. Fernandez suggested that cardiac evaluation is indicated for all children with Aarskog syndrome, but no CHD found in our case[6]. Vandenberg found abnormal venous drainage with raised ICT in one patient[7], Mikelsaar and Lurie (1992) described a boy with features typical of Aarskog syndrome who also had leg lymphedema extending to the knees when examined at the age of 10 years[8]. Logie and Porteous (1998) tested IQ in 21 males under 17 years of age with clinically confirmed Aarskog syndrome and found their IQs to lie within the normal range. The diagnosis of Arskog syndrome is mainly on clinical basis, it is confirmed by genetic testing.

CONCLUSION

It is faciogenital dysplasia caused by mutation in a gene called FGDY1 in band p11.21 on the X chromosome. It is very rare congenital malformation. It is x linked recessive, although autosomal dominant pattern can occur. As other congenital malformation, no cure is possible, only supportive treatment is available. Genetic counseling is necessary for prevention of such syndromes. Intellictual development varies from normal to severe mental retardation in Aarskog syndrome.

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