

Aarskog-Scott syndrome: An unusual cause of scoliosis

ABSTRACT

A 16-year-old boy who had been diagnosed previously as Aarskog-Scott syndrome (AAS), referred to our clinic with shoulder asymmetry for 1 year. Results of spine examination showed a 52° right thoracic curve at T3-T11. Surgery was planned, and T1–L1 posterior instrumentation and fusion were performed. After surgery, satisfactory correction was achieved, and during 10 years follow-up, the patient had no complaints. AAS is a X-linked genetic disorder with facial, genital, and skeletal manifestations. Scoliosis is not reported as a typical finding of AAS, and there is no reported case in the English literature. Due to mutation affecting the developing skeleton tissue, spinal deformities may develop. In our case, concave side fusion was seen at the deformity. Although we do not know any specific pattern of the scoliotic deformity of this syndrome, surgical correction of the deformity can be difficult because of the premature fusion at these levels.

Keywords: Aarskog-scott syndrome, scoliosis, spinal deformity

INTRODUCTION

Aarskog-Scott syndrome (AAS) (facio-digital-genital) was first described in 1970 by Aarskog^[1] in seven males from one family with ocular hypertelorism, anteverted nostrils, a broad upper lip, and a shawl scrotum. A year later, Scott^[2] emphasized the occurrence of ligamentous laxity, genu recurvatum, and flat feet. It is a rare X-linked recessive disorder and related with a mutation on FDG1 found within region Xp11.21. Typical findings include short stature, distinct craniofacial abnormalities, brachydactyly with interdigital webbing, joint laxity, and shawl scrotum. A variety of other skeletal malformations is observed including cervical spine abnormalities such as odontoid hypoplasia, congenital vertebral anomalies, spina bifida occulta, and additional pairs of the rib. To the best of our knowledge, there has been no case about AAS with scoliosis in English literature. In this case report, we present an adolescent scoliosis patient who was previously diagnosed as AAS and treated by posterior instrumentation and fusion.

CASE REPORT

A 16-year-old boy is referred to our clinic with shoulder asymmetry for 1 year. He had been diagnosed as AAS

previously. He had short stature (145 cm), macrocephaly, broad upper lip with long philtrum, ocular hypertelorism, shawl scrotum, and brachydactyly. He had no mental retardation. Results of spine examination showed a right thoracic scoliosis with an elevation of the ipsilateral shoulder [Figure 1a]. Neurological examination was completely normal.

Standing anteroposterior spinal radiographs obtained and a 52° right thoracic curve at T3-T11 was observed [Figure 1b]. Concave side fusion was seen between T5 and T9 levels. Surgery was planned for this deformity. Preoperative lateral bending radiograph showed correction of the curve to 28°, and traction radiograph showed an angle of 24°. Posterior thoracic approach with transpedicular instrumentation, correction, and

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Figure 1: (a) Clinical photo of the spine and typical face findings of Aarskog-Scott syndrome (b) Preoperative X-ray

fusion was performed between T1 and L1 vertebrae. During surgery, fusion at the apical concave side was observed. After placement of the pedicle screws, correction was achieved by rod derotation maneuver. Satisfactory correction of the sagittal and coronal plane was obtained, and coronal Cobb angle was measured 16° after surgical correction and thoracic hump disappeared. Patient mobilized on 2nd day postoperatively, and shoulder asymmetry resolved [Figure 2a]. At 10 years follow-up, no shoulder and pelvic asymmetry were observed. Radiographs showed a slight increase of the Cobb angle, which was 18° . No adding-on was observed below the instrumentation level; however, slight right trunkal tilt and 3.3 cm coronal imbalance were observed [Figure 2b].

DISCUSSION

AAS is an X-linked genetic disorder with facial, genital, and skeletal manifestations. An autosomal recessive and autosomal dominant inheritance have also been reported. *FDG1* mutation is responsible for this syndrome, and this mutation appears to affect the developing skeleton tissue during the onset of ossification in regions of endochondral and intramebranous ossification including craniofacial bones, vertebrae, ribs, long bones, and phalanges, thus skeletal abnormalities dominate the clinical manifestations of the syndrome.^[3] Gorski *et al.*^[3] reviewed the spectrum of skeletal abnormalities reported in the literature more than 110 patients. Skeletal abnormalities of this syndrome include short stature, delayed ossification centers, hypertelorism, maxillary hypoplasia, broad nasal bridge, cervical spine anomalies, rib and sternal anomalies, short/broad hands, brachydactyly, hypoplastic phalanges, syndactyly, and joint laxity. No scoliosis patient was reported in this review. In the literature, there is no English report about AAS and scoliosis. To the best of our knowledge, only one case reported an AAS patient with scoliosis, in German, by Fehlow *et al.*^[4] They reported a 22-year-old patient with AAS who had a scoliosis causing a radicular impairment with resulting severe paresis of the right fibular nerve. The patient was also associated with mental retardation, epilepsy, premature craniosynostosis, and tardive dyskinesias. However, this mental retardation is atypical

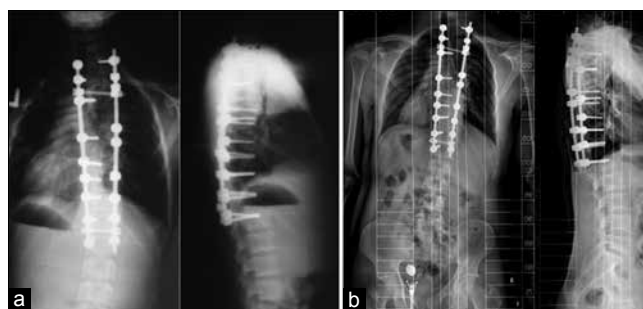


Figure 2: (a) Early postoperative X-ray (b) 10 years follow-up X-ray

for AAS because Logie and Porteous,^[5] reported normal IQ levels in 21 AAS patients. Our patient had a single thoracic curve recognized 1 year before admission. We do not know the nature of the curve whether it is progressive or not. We could not follow the patient because the curve was already 53° , thus surgery was addressed. Only thoracic fusion was performed similar to idiopathic-like curves and during 10 years follow-up, no decompensation was developed. The distinct feature of the curve was the concave side fusion. Although we do not know any specific pattern of the scoliotic deformity of this syndrome, surgical correction of the deformity can be difficult because of the premature fusion at these levels.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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