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Case Report

Dentistry Section

Identifying Aarskog Syndrome

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ABSTRACT

Aarskog syndrome also known as Aarskog-Scott Syndrome, Facio-digito-genital Syndrome or Faciogenital Dysplasia is a rare, X-linked disorder predominantly affecting males, characterized by facial, skeletal and genital anomalies. This is a case report of a 15-year-old male patient who visited our college complaining of poor facial aesthetics. History revealed consanguinity and his sibling to be suffering from the same. A diagnosis of Aarskog syndrome was made based upon the detailed patient history, thorough clinical evaluation and identification of characteristic findings in radiographs. Professional counselling explained him the nature of his condition and treatment options to correct dental anomalies and congenital malformations were advised.

Keywords: Aarskog-Scott syndrome, Facial anomalies, Genital anomalies

CASE REPORT

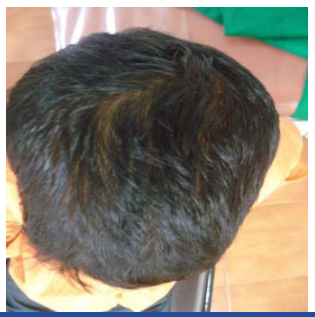
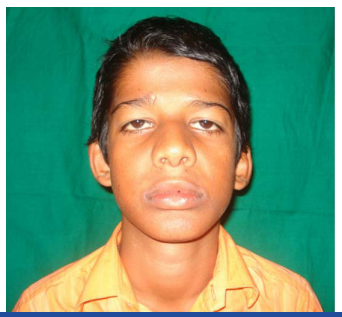
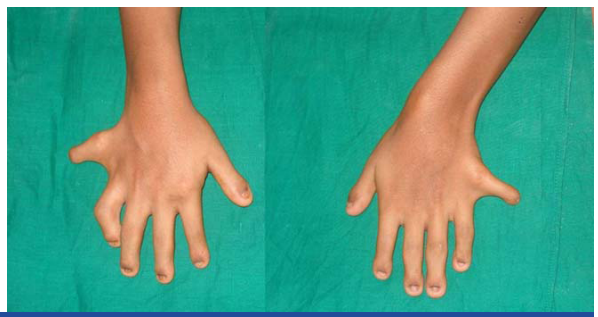
Syndromes not associated with severe medical problems or mental retardation can easily go unnoticed by physicians who may not be aware of the syndrome and co-related features. We report a case of 15-year-old male patient who came with a complaint of poor aesthetics because of missing teeth in the front region of the upper and lower jaws since childhood. He gave no history of extraction or exfoliation of teeth in front region of the jaw and there was no history of trauma either. He stated that he was the second son of consanguineous parents and had two brothers; the elder brother had a short stature, missing teeth and extra fingers. No relevant medical history was reported. On general examination he had short stature [Table/Fig-1], was well nourished and well oriented in time place and person. He had sparse scalp hair [Table/Fig-2]. His facial appearance was striking with abnormally large nose and ptosis of upper eyelids, hypertrophied and everted lower lip [Table/Fig-3]. The patient was 1.32 meter tall, weighed 46 kilograms and his vital signs were within the normal range. The patient had 6 digits on both his hands and the nails appeared smaller in size than normal [Table/Fig-4]. The patient could not adduct his thumb, index finger and the extra digit. Inward bowing of both lower limbs was noticed and both the feet had six toes [Table/Fig-5].

Intra-oral examination revealed an abnormally large tongue covering the occlusal surface of the mandibular dentition. The maxillary lateral incisors and all four mandibular incisors were missing. A high arched palate was observed with palatal positioning of second premolars [Table/Fig-6]. The maxillary central incisors were macrodontic with altered morphology and diastema [Table/Fig-7]. All the canines were conical in shape and the edentulous ridge of mandibular incisor region showed a soft tissue prominence and hyperplastic labial frenum [Table/Fig-7]. Based on the history and the clinical findings a provisional diagnosis of acrodental dysostoses was arrived. The other differential diagnosis considered were Weyers acrodental dysostosis which besides the dwarfism, postaxial polydactyly and dysplastic teeth is also characterized by cardiac problems but as patient did not report of any systemic problems and examination of vital signs did not reveal any abnormalities this was ruled out. Aarskog syndrome was also considered as similar features are noticed as in this case but a characteristic feature of genital abnormality had to be further examined. Leopard syndrome was excluded because of absence of multiple black macules on the skin and absence of sensorineural deafness.

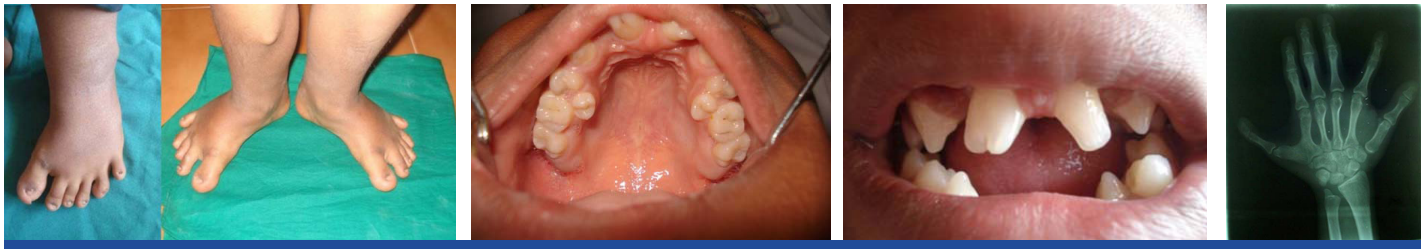
Investigations: An Orthopantomograph (OPG) of the patient was done which confirmed all the clinical observations made regarding the number, shape and position of teeth. The hand wrist radiograph showed an extra digit and star like pattern of the digits [Table/Fig-8]. Further physical and clinical examinations were conducted by a general physician who acknowledged that there was no cardiac problem but the examination of genitals revealed unusual shawl like tissue around glans penis. This characteristic feature made us to presume the most probable diagnosis as Aarskog syndrome. Further blood investigations and genetic investigation to search for mutation in FGD1 were advised to the patient but he was quiet unwilling for further investigations. Besides explaining the patient his condition, prosthetic rehabilitation was done for him which was his main concern.

DISCUSSION

In 1970 Aarskog, in Norway, described a syndrome associating short stature with certain anomalies of the face, hands, feet, and

[Table/Fig-1]: Short stature. **[Table/Fig-2]:** Sparse scalp hair. **[Table/Fig-3]:** Large nose, droopy upper eyelids, hypertrophied and everted lower lip. **[Table/Fig-4]:** Extra finger on both hands, small nails.



[Table/Fig-5]: Bowing of legs and extra toes. **[Table/Fig-6]:** High arched palate, missing lateral incisors and palatally positioned premolars. **[Table/Fig-7]:** Altered morphology of central incisors with midline diastema and missing mandibular central and lateral incisors. Hyperplastic labial frenum and conical shaped canines.
[Table/Fig-8]: Star like pattern of hand radiograph.

genitalia in seven males from two generations of the same family [1]. The following year Scott also described a similar case [2]; thus, acquiring the name: Aarskog-Scott syndrome. It is an X-linked disorder caused by mutations in the FGD1 gene and therefore males express all the clinical manifestations and female carriers only show minor manifestations of the disorder, especially in the face and hands [3]. Typical features of Aarskog syndrome have been presented in [Table/Fig-9]. Population surveys estimate that the Aarskog-Scott syndrome occurs with a recognized frequency of approximately 1 per one million in general population [4]. Most of the cases are clinically diagnosed and only 35 molecularly proven cases have been published worldwide [Table/Fig-9] [5].

There are no specific therapies for Aarskog syndrome. Some features such as inguinal or umbilical hernias, cryptorchidism and unusually severe craniofacial features may need surgical intervention [15]. Atypical features associated with Aarskog syndrome reported in the literature are shown in [Table/Fig-10].

CONCLUSION

Aarskog syndrome may be undiagnosed due to the rarity of the condition and lack of knowledge. Dental and orofacial features are strikingly characteristic and play a key roll in the diagnosis of conditions like Aarskog syndrome. Diagnosis and management of rare syndromes will require a multidisciplinary approach involving paediatrician, orthopaedic surgeons, dentists and neuropsychiatrist.

General features [5,6,7]	Extra-oral features [6]	Intra-oral features [7,8]	Radiographic features [9]
- Short Stature - Severly shortened distal extremities - Webbed appearance of hands and feet - Hyperflexible joints - Short and inwardly curved little finger - Simian crease in the palm of the hand - Shawl scrotum or bifid scrotum - Delayed puberty	- Round face - Hypertelorism - Ptosis - Down slanting palpebral fissure - Broad nasal bridge with anteverted nostrils - Long upper lip with a linear dimple below the lower lip	- Maxillary hypoplasia - Missing teeth (either impacted or hypodontia) - Decreased vertical facial height	- Lumber kyphoscoliosis - Pes planovalgus (flat foot)
All these features were appreciated in our patient except for Lumbar Kyphoscoliosis			

[Table/Fig-9]: Typical features of Aarskog syndrome reported in the literature.

Year of Publication	Author(s)	Uncommon findings associated with Aarskog syndrome
1983	Daniel LH [10]	The author reported a case with metatartus adductus (forefoot turned inwards) in Aarskog syndrome and emphasized on surgical correction of the same.
1988	Ahmad ST et al., [11]	The authors reported Aarskog syndrome in 5 siblings of consanguineous marriage. This is an exceptional case where the female siblings were also expressing the features of Aarskog syndrome.
1992	Ruth V-AM and Lurie IW [9]	The authors documented lymphoedema of the feet appreciated in maternal grandfather and grandson both of whom were affected with Aarskog syndrome.
1999	Waldo S et al., [12]	The authors describe a case of Aarskog syndrome identified prenatally by sonography at 28 weeks of pregnancy. Cystic hygroma and vertebral anomalies were diagnosed by ultrasonography which are not typical features of Aarskog syndrome.
2005	Mahmoud NS et al., [13]	The authors have published a case with congenital heart disease (aortic root dilation and sub-valvular aortic stenosis). They recommended cardiac screening for all patients with Aarskog syndrome.
2012	Raghavendra BN et al., [14]	The authors have published a case of Aarskog syndrome combined with temper tantrums, demanding behaviour, grandiose ideas, over familiarity, abusive assaultive behaviour and tobacco abuse in a 10 year old. Behavioural and personality disorders were not reported with the other cases.
2016	The present case	The atypical features noticed were polydactyl and alopecia.

[Table/Fig-10]: Atypical features associated with Aarskog syndrome reported in the literature.

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