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 and 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
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].

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 role 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.

### References

[12] 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.
[13] Mahmoud NS et al., [13]. 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.
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FINANCIAL OR OTHER COMPETING INTERESTS: None.

