Aarskog-scott syndrome (AAS): a case report

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Abstract

Background Aarskog-Scott syndrome (AAS) is a rare developmental disorder characterised by facial dysmorphism, genital and limb anomalies as well as disproportionate acromelic short stature. Clinical diagnosis is based on physical examination and the presence of the most characteristic clinical signs. The diagnosis can be finally confirmed by molecular tests, which identify mutations in the FGD1 gene.

Case Report The report outlines the orthodontic treatment of a 6-year-old male patient, who was diagnosed with AAS syndrome. He presents all facial and oral clinical signs of this syndrome. The extent of maxillary hypoplasia and early dental crowding are so significant that immediate expansion therapy is required.

Conclusions Dental management of patients with AAS syndrome represents a challenge for paediatric dentists. The key to improving a patient's aesthetic, functional and psychological condition is making the correct orthodontic decision.

KEYWORDS Aarskog-Scott syndrome, Faciogenital dysplasia, Faciodigitolgenital syndrome, case report

Introduction

Aarskog-Scott syndrome (AAS), also named faciogenital dysplasia, is a X-linked recessive genetic condition characterised by facial, genital and digital symptoms [Scott CI, 1971]. This syndrome is caused by mutations in the FGD1 gene. The gene maps to the short arm of the X chromosome and encodes a guanine nucleotide exchange factor (GEF), which, activating Cdc42, participates directly in cytoskeletal organisation, growth regulation, and normal embryonic development [Zanetti Drumond et al., 2021]. AAS primarily affects males with an estimated population prevalence equal or slightly lower than to 1/25.000. The syndrome is clinically heterogeneous, but the most common clinical findings are craniofacial anomalies, short stature, a sunken chest, brachydactyly and genital dysmorphism such as shawl scrotum and cryptorchidism [Spiegel et al, 2009]. Additionally, some types of neurocognitive disabilities and behaviour disorders could be present. These disorders range from attention deficit and hyperactivity disorder (ADHD) to severe intellectual disability [Zanetti Drumond et al, 2021; Orrico et al, 2005]. Dysmorphic facial features include a round face with broad forehead, ptosis, a short and upturned nose, a well-formed philtrum and ocular hypertelorism. There are also some characteristic ear features of AAS like low-set ears and thickened, "fleshy" earlobes.

From the oral point of view, the clinical signs are ankyloglossia, a space between the upper teeth (diastema) and maxillary hypoplasia. There could also be a variety of abnormalities affecting the teeth like missing teeth at birth, delayed eruption of teeth, enamel hypoplasia and consequently an increased risk of dental caries [Halse et al, 1979]. Finally, congenital heart defects, cleft palate and cleft lip are possible symptoms that occur less frequently.

Case Report

E. is a 6-year-old male patient who comes to our attention,

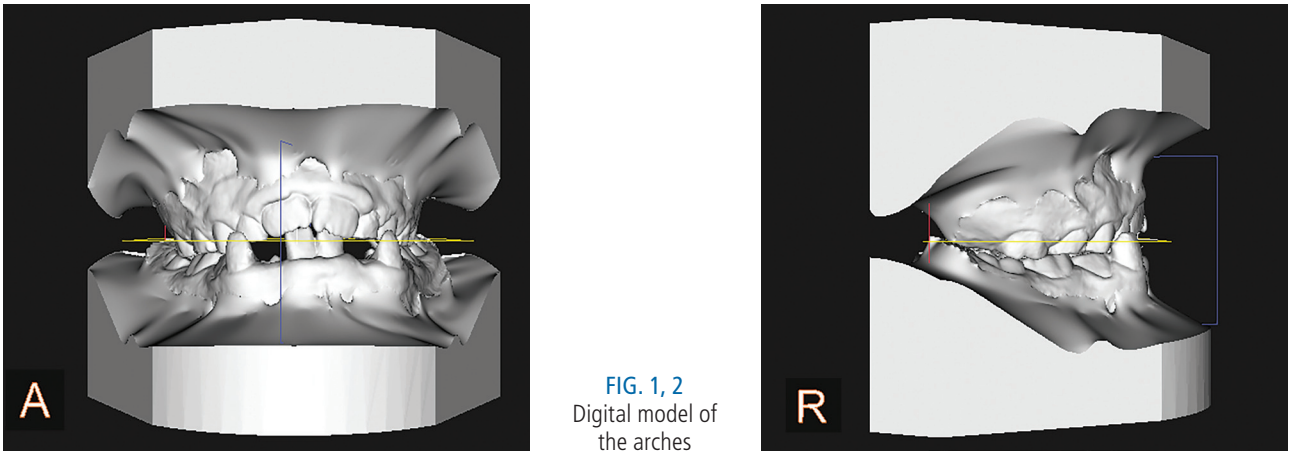

FIG. 1, 2
Digital model of the arches

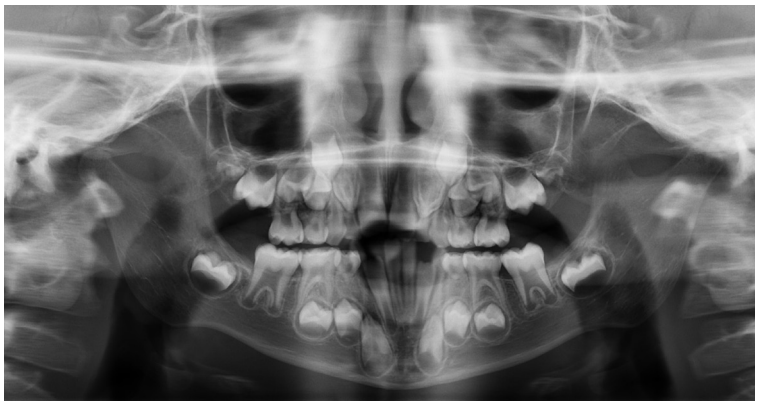

FIG. 3 Initial orthopanoramic X-ray

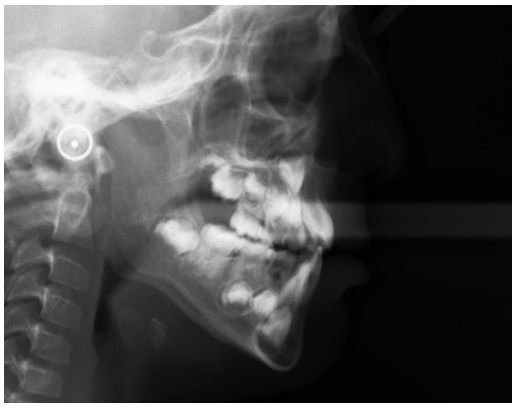

FIG. 4 Lateral teleradiography for cephalometric analysis

for the first time, in 2019. He was previously diagnosed with Aarskog-Scott syndrome. The patient's general state of health is good with regular psychomotor development. The anamnesis is negative for congenital heart defects and neurocognitive disabilities. Ocular problems, such as strabismus and ophthalmoplegia, are not evident. No food or drug allergies are reported.

Due to extraoral examination, some clinical characteristics of AAS such as short stature, a round face with broad forehead, low set ears and ocular hypertelorism are observed. Our patient presents broad and short hands with short, stubby fingers.

Intraoral examination shows early mixed dentition, anterior and bilateral cross bite, centred midlines and an anterior crowding in the upper arch (Fig. 1-2).

In the patient's orthopanoramic X-ray, all the buds of permanent teeth are present. In the upper arch, the risk of canine inclusion is evident due to the lack of adequate space for their eruption. This radiological investigation also shows an undefined dental anatomy and dimension of the superior lateral incisors (Fig. 3).

Cephalometry analysis diagnoses a skeletal class III in a hyperdivergent, biretrused patient with a growth pattern of clockwise mandibular rotation and mandibular asymmetry (Fig. 4).

Discussion

After cephalometric analysis, the chosen orthodontic treatment includes expansion and anterior traction of maxilla. The level of maxillary hypoplasia and early dental crowding are so severe that two expansion phases are probably required, with an orthodontic revaluation at the end of the first stage dental eruption (when the upper permanent central/lateral incisors and molars are completely erupted) [Mummolo S. et al., 2014]. The first phase starts before the eruption of permanent molars and lasts nine months. The orthodontic appliance presents external arms for Delaire's mask. This initial treatment results in an improvement of transverse diameters and anterior defects of the maxilla. E.'s cooperation with respect to hygiene and treatment compliance represents a positive prognostic factor. In the case of our patient, the delayed eruption of lateral incisors prevents the orthodontic revaluation and the beginning of the second phase of expansion. In September 2022, the patient comes to our clinic for an emergency visit. The clinical examination shows an abscess in the apical and buccal region, between the elements 6.2 and 6.3. The deciduous lateral incisor is mobile and painful on percussion. The parents report a fever and an exacerbation of symptoms two days before the appointment.

The radiological investigation includes orthopanoramic (Fig. 5) and periapical end-oral X-rays at site 6.2 – 6.3 (Fig. 6). From these radiological examinations, it is possible to observe an unphysiological shape and size of the buds of elements 1.2 and 2.2.

A second level X-ray investigation (CBCT scan) is suggested due to the unclear shape of lateral permanent incisors, shown by the first level X-ray. At the same time, antibiotic therapy is

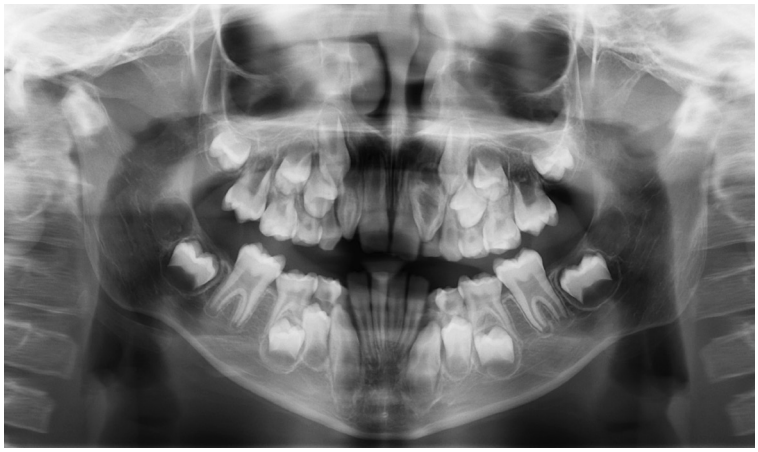

FIG. 5 First level X- ray

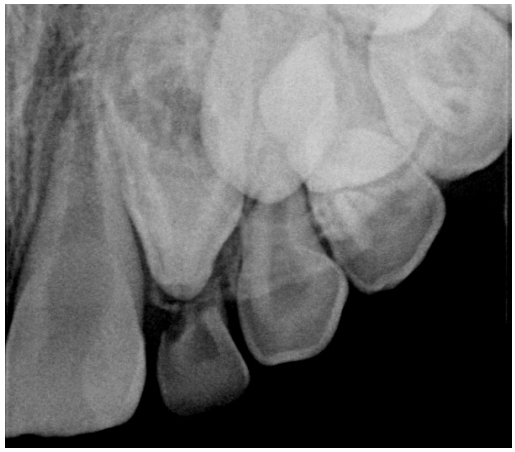

FIG. 6 Periapical end-oral X-ray (site 6.2 - 6.3)

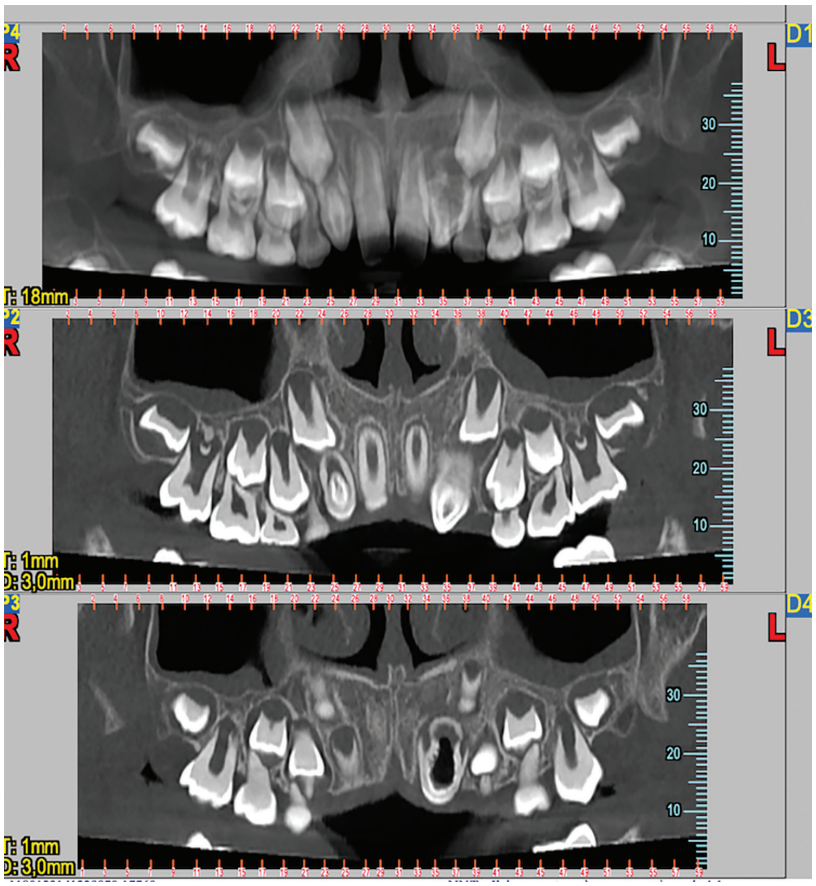

FIG. 7 Second level X-ray

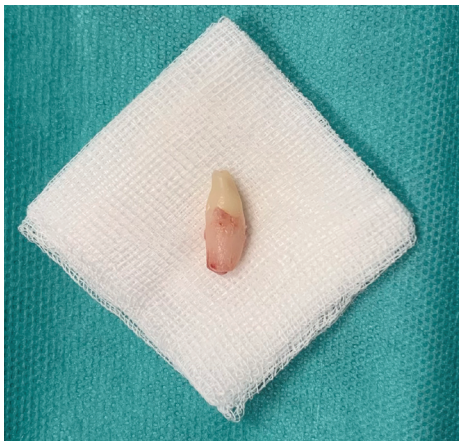

FIG. 8 Element 1.2 post extraction

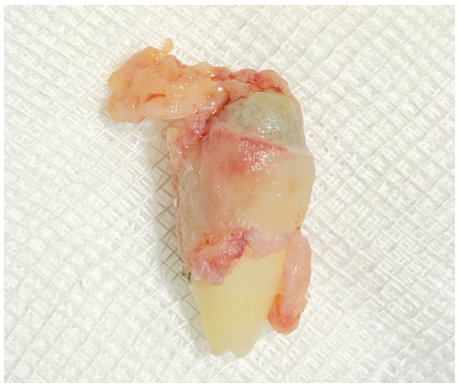

FIG. 9 Element 2.2 post extraction

prescribed for six days. The CBCT shows a shape and dimension alteration of the elements 1.2 and 2.2 (Fig. 7).

In agreement with E.'s parents, it is decided for the surgical extraction of both elements. This choice avoids the risk of worsening the mandibular and dental asymmetry already present and the possibility of any problems related to the eruption of other contralateral incisors. The surgery is performed under general anaesthesia and, in only one session, the elements 1.2, 2.2 and 6.2 are extracted (Fig. 8-9) [Mummolo S. et al., 2020]. These elements are subjected to histological examination. According to the histological report, part of the samples are identified as teeth, with discrete dentine thickening and substantial share of dental pulp, and other parts are referred to as the compound odontoma.

Conclusion

The aim of this case report is to show the paedo-orthodontic treatment chosen for a patient diagnosed with AAS. It must be remembered that the pathognomonic signs of the disease appear early during first childhood. The careful detection of the clinical signs allows an early diagnosis, which, in turn, helps clinicians choose the most appropriate treatment for these patients. In patients affected by faciogenital dysplasia, the dental abnormalities and, more generally, those of the entire orofacial region are such that orthodontic intervention is almost always required. Their high prevalence of caries makes the interdisciplinary approach between paediatric dentist and orthodontist essential during all treatment periods. The histological report of the extracted dental elements refers to the compound odontoma. Based on these clinical findings, the development of more studies that investigate the presence of odontoma as dental abnormalities in patients with AAS is suggested.

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