Ellis–Van Creveld syndrome is a rare autosomal recessive disorder which was initially described by Richard Ellis and Simon Van Creveld in the year 1940. The syndrome is characterized by tetrad of clinical manifestations of chondrodysplasia, polydactyly, ectodermal dysplasia and congenital cardiac defects.

Reported incidence is 1,500,000 live births and in India it is even rare.

Case Report

A 4½-year-old female patient from parents of consanguineous marriage presented to our dental op with a chief complaint of missing teeth in the upper and lower front jaw region and unaesthetic appearance since birth. First tooth to erupt was the back tooth that too after 1½ years. There was difficulty in chewing and had slurred speech. Review of systems revealed that she was otherwise normal. Patient has undergone surgery 2 years back for bowed legs and her walk has improved.

On general examination, the patient had bowed legs. She showed stunted growth [Figure 1] and polydactyly in both the hands [Figure 2] and right leg with dysplastic nails. In the left leg the third and the fourth toes showed syndactyly [Figure 3]. Her brother who was 19 years old also had similar problem with stunted growth [Figure 1], polydactyly [Figure 2] in both the hands, syndactyly [Figure 3] of the left fourth and fifth toe with missing anterior teeth.

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On intraoral examination of the girl showed partial hare lip [Figure 4] with obliteration of upper labiogingival sulcus, the mandibular arch showed multiple, high frenal attachments [Figure 5]. The tongue showed depapillation on the dorsal surface. Partial anodontia with conical shaped mandibular lateral incisors was also present. The boy also showed partial hair lip [Figure 4], missing anterior teeth with multiple high frenal attachments [Figure 5].

Investigations were carried out. Examination of the heart revealed to be normal [Figure 6]. Complete skeletal survey was done for both of them which revealed postaxial bilateral polydactyly. The girl’s wrist joint anteroposterior view shows that the fourth and fifth metacarpals are fused on both sides with decreased bone density in the metacarpals [Figures 7 and 8]. Orthopantamograph of the boy and girl reveals missing maxillary and mandibular anterior teeth, conical mandibular laterals [Figure 9].

Discussion

Ellis–Van Creveld syndrome or chondroectodermal dysplasia syndrome is an uncommon disease inherited as an autosomal recessive trait with the incidence of 1:244,000 of the total population.[4] Chondrodystrophy is the most consistent clinical feature which is due to a defect in ossification that results in short stature and limb shortening which is more striking in the distal rather than proximal extremities.[5] Polydactyly is always present with typical postaxial hexadactyly of the hands and in few cases feet.[6]

All three embryonic layers appear involved in Ellis–Van Creveld syndrome.[7] The signs of ectodermal dysplasia is usually limited to nails, teeth, and gums. Endodermal involvement is not very common but sometimes affects the liver and lungs. Abnormalities of heart and kidneys indicate mesodermal involvement.[8]

Partial harelip, maxillary alveolar clefts, abnormal tooth shape (conical teeth), size (micro/macrodontia), structure
Gokulraj, et al.: A rare case report of Ellis Van Creveld syndrome in siblings

Summary

Ellis–Van Creveld syndrome or chondroectodermal dysplasia of the siblings showed bilateral ploydactyly, partial hare lip, multiple labial frenum, obliteration of the labiogingival sulcus, partial anodontia, conical teeth with stunted growth all of which are pathognomonic for the diagnosis. The management of patients with EVC requires a multidisciplinary approach which includes many specialties such as the pulmonologist, cardiologist, orthopedician, oral and maxillofacial surgeon, pediatrician, pedodontist, prosthodontist, and radiologist along with oral and maxillofacial radiologist. This article has presented a case of siblings of EVC syndrome. This case report enlightens the awareness of EVC syndrome though very rare is still a possibility.

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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Figure 7: The boys hand wrist radiograph shows fusion of fourth and fifth metacarpals on both sides

Figure 8: Anteroposterior view of feet shows decreased bone density in tarsal and metatarsal bones

Figure 9: Orthopantamograph

(enamel hypoplasia), number (missing teeth/fused teeth), site of implantation (disorderly arranged and irregular spacing), multiple labial frenum, obliteration of the labiogingival sulcus are all pathognomonic and should be used in primary diagnosis.[9]

Cardiac defects are found in 50–60% of patients with a common atrium and persistent atrioventricular canal being the most common defects.[10] Patient ductus arteriosus, ventricular septal defects and atrial septal defects could also be present.

Radiological features comprise a narrow thoracic cage and wide, spade-like anterior ends. The pelvis has square iliac ala and “trident acetabulum.” Limbs have shortened long bones, with wide diaphyses and metaphyses. Proximal and middle phalanges are short with cone shaped epiphyses.[11]

Ellis–Van Creveld syndrome or chondroectodermal dysplasia has been mapped to chromosome 4p16.[12] The differential diagnosis of Ellis–Van Creveld syndrome includes thoracic dysplasia of Jeune, Weyers acrofacial dysostosis, K/C kusick-kaufman syndrome, thoracic cage deformity, chondrodysplasia punctata, asphyxiating thoracic dystrophy.