Ellis-van Creveld Syndrome

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Ellis-van Creveld Syndrome (EVC) or Chondroectodermal Dysplasia is a rare congenital genetic disorder caused by autosomal recessive traits. All embryonic layers are involved, but an uncommon presentation of endodermal could be seen. The syndrome presents with general manifestations, such as postaxial polydactyly, ectodermal imperfections, short ribs and heart defects. The oral manifestations of Ellis-van Creveld syndrome are enamel hypoplasia, hypodontia, and malocclusion of teeth. The mutation of the genes EVC1 and EVC2 is responsible to cause the syndrome.

A nineteen-year-old female presented with a complaint of oddly-shaped teeth. She had a history of patent ductus arteriosus, which was resolved 3 years after birth. Her general features revealed that she was short in height, short-ribbed, and had hypoplastic fingernails. An extraoral examination of the patient revealed a class III skeletal base. The intraoral examination revealed multiple missing teeth, enamel hypoplasia and hypertrophic mandibular labial frenulum. Radiographic examination confirmed EVC in this young female, which is reasonably uncommon.

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Ellis-van Creveld Syndrome or Chondroectodermal Dysplasia is a rare congenital disorder caused by mutation of the EVC1 and EVC2 genes[1]. In 1940, the syndrome was described by Ellis and van Creveld[3]. It has a prevalence of 7 in one million live-births[3]. Thirty percent of patients were reported to have parental consanguinity[3].

In Ellis-van Creveld Syndrome, all embryonic layers are involved[10]. The ectodermal layers are mainly affected, such as the nails, teeth, and gums[10]. Some would present with eye and neural involvements. Cardiac and bone complications have been shown to involve the mesodermal layer[10]. Endodermal involvement of the liver and lungs are rare, but have been described in literature[10].

The syndrome has multiple general features: postaxial polydactyly, short ribs, ectodermal malformations and heart defects[4]. Approximately 50% of infants have a high mortality rate due to cardiorespiratory problems, whereas survivors may have average lifespan[3,4]. Rare presentations could include congenital cataracts, hypospadias and cryptorchidism[4].

Oral anomalies may present with delayed eruption of teeth. Studies revealed anomalies such as the presence of neonatal teeth in 25-30% of the cases at birth, submucous clefts, mandibular and maxillary alveolar process notching with a wide or a continuous hypertrophic labial frenulum, malocclusion, enamel hypoplasia, dystrophic philtrum, microdontia, peg-shaped lateral incisors, taurodontism, and hypodontia[3-5].

To our knowledge, this is the first case of EVC reported in the Kingdom of Bahrain and the Arabian Gulf region.

The aim of this report is to present a rare case of Ellis-van Creveld Syndrome with uncommon dental and oral appearances.

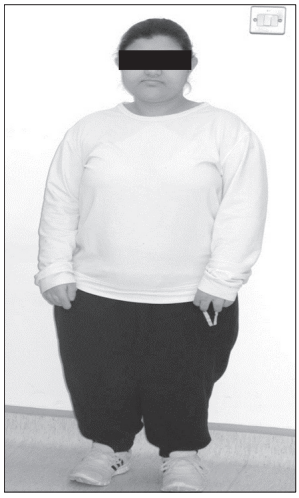
THE CASE

A nineteen-year-old female presented with chief complaint of spaces in the front region of her upper and lower jaws. The patient was the second child of a consanguineous marriage and was a known case of Ellis-van Creveld Syndrome, which was diagnosed at an early age. She had a history of patent ductus arteriosus, which resolved after 3 years without medical intervention.

She had dental treatment for multiple carious teeth at an early age. She had multiple dental extractions. The patients' parent reported that her 16-year-old sister was also diagnosed with Ellis-van Creveld Syndrome

The patient's mother reported that the patient had two mandibular deciduous central incisors present at birth, which exfoliated few months after she was born.

General examination revealed that the patient had all the features of dwarfism, see figure 1. She is a short-ribbed patient with hypoplastic nails, see figure 2.



Consent has been obtained

Figure 1: Full Body Image showing Features of Dwarfism

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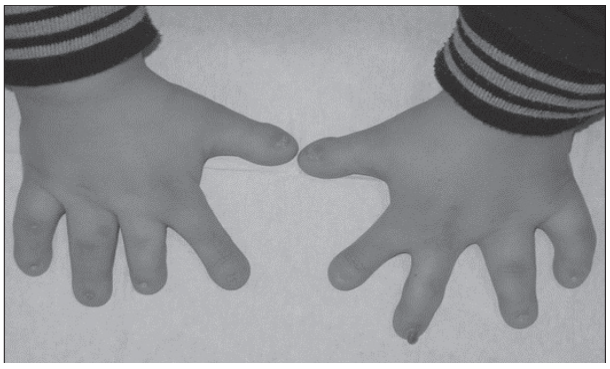

Figure 2: Hypoplastic Fingernails

The patient had slight facial asymmetry to the right. No temporomandibular joint problems were found. The patient had a moderate class III skeletal base with incompetent lips and an increase in lower facial height.

The patient had the oral features of hypodontia with missing maxillary lateral incisors and canines as well as missing mandibular central and lateral incisors. Enamel hypoplasia, hypoplastic maxilla with a class III malocclusion and an open bite from the right permanent first molars to the contralateral side of the same teeth were also found. The patient's lower frenulum was attached to the labial gingiva which is a typical feature of the disorder, see figure 3. The patient was seen by an orthodontist at the age of 10 years and was advised to routinely attend the dental clinic to review the dentition development.

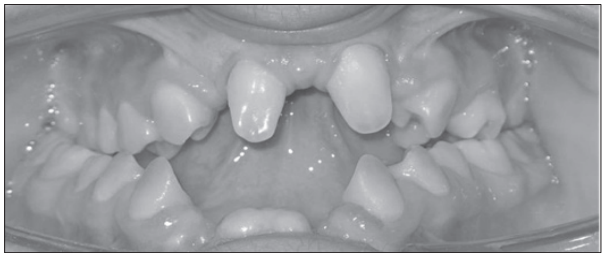

Figure 3: Frontal Intraoral Image Showing Malformed Maxillary Central Incisors, Hypodontia, and a Hypertrophic Labio-gingival Frenulum

Dental panoramic tomography (DPT) revealed that the patient had all permanent teeth except for the maxillary lateral incisors and canines, mandibular central and lateral incisors, see figure 4.

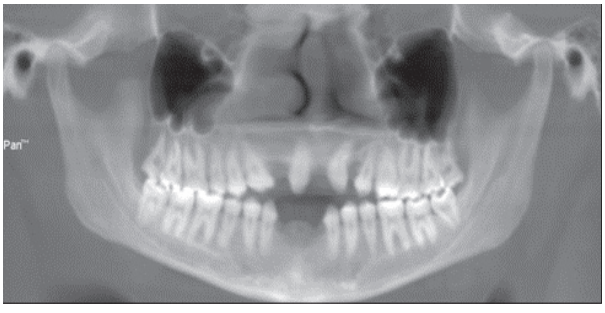

Figure 4: Dental Panoramic Tomography Showing Hypodontia and Malformed Maxillary Central Incisors

Both mandibular third molars were closely in contact to or penetrating the inferior dental canal. Taurodontism of the permanent dentition (maxillary and mandibular permanent molars) was found on DPT.

A lateral cephalogram radiograph confirmed the diagnosis of class III incisal relationship on class III skeletal base with an open bite as described.

Cone-beam computed tomography (CBCT) showed an anteroposterior view of the patient's head revealing hypoplastic maxillary incisors, missing maxillary lateral incisors and canines, mandibular permanent central and lateral incisors**.**

At the age of 10, intraoral examination revealed multiple carious teeth, poor oral hygiene, enamel hypoplasia and malocclusion. The first line treatment by the pedodontist, orthodontist and maxillofacial surgeon was the prevention of dental disease by oral hygiene, fluoride gel application, diet analysis, scaling, restorative treatment of carious teeth, extraction of poor carious teeth, periodic review of developmental dentition and definitive plan when she reaches 16 years of age

At the age of 17, examination revealed premolar supernumeraries and a reversed overjet. The treatment of this case was a combination of restorative treatment of malformed permanent dentition, prosthetic replacement of missing teeth, orthodontic decompensation and future orthognathic surgical correction. Orthodontic fixed appliances were placed after discussing the risks and benefits with the patient. At 18 years of age, the patient started the treatment and is currently under orthodontic appliances until final surgical treatment is contemplated.

DISCUSSION

Ellis-van Creveld Syndrome is a rare genetic disorder mainly caused by the mutation of EVC1 and EVC2 genes that is located on 4p16 chromosome, which is an autosomal recessive trait that mainly affects skin and skeleton[4,6].

The lower extremities are affected predominantly due to the mesomelic shortening[4]. Knock-knees or genu valgum may be seen, which needs surgical interventions[4].

The diagnosis could be attained by radiological prenatal ultrasonography at 18 weeks in the gestational period, which would reveal the fetal anomalies. In addition, it could be revealed through family history and dental features[2,3,6]. A conclusive diagnosis may be attained by DNA mapping which is the most reliable to detect this syndrome[2]. At the neonatal period, patients suffering from EVC syndrome may require critical care for respiratory distress or cardiac failure[8].

Oral signs of the syndrome may present as submucous clefts, malocclusion, enamel hypoplasia, hypodontia, multiple wide or continuous labial frenulums causing alveolar process notching[2,4,5].

The cardiac anomalies could be mitral and tricuspid valve defects, single atrium, patent ductus, ventricular septal defect, atrial septal defect, or hypoplastic left heart syndrome[7].

The differential diagnoses of EVC syndrome could be Saldino-Noonan syndrome, Majewski syndrome, Verma-Naumoff syndrome, Beemer-Langer syndrome, Jeune Dystrophy, McKusick-Kaufman syndrome and Weyers syndrome[2].

It has been recommended that an antibiotic cover be prescribed and administered to patients who have a history of infective endocarditis, a prosthetic heart valve, mitral valve prolapse

with thickened leaflets or valvular regurgitation, who are at risk of bacteremia during surgical interphases or dental procedures[3].

Composite restorations or crowns may be the treatment of choice for malformed dentition[9]. Partial dentures may be fabricated for the restoration of missing teeth for esthetics, function and speech[9]. Orthodontic treatment is also a treatment of choice to treat malocclusion[9].

CONCLUSION

The management of such cases requires a joint clinic in conjunction with a high quality healthcare. Patients with Ellis-Van Creveld syndrome could be subjected to psychological distress. A thorough medical, family, and social history with proper examination would reveal other disorders. Prevention of dental disease is the key to success in dental treatment. An urgent referral to a specialist is indicated for further investigations and treatment options.

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