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CASE REPORT

Ellis-van Creveld Syndrome

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Abstract

Ellis-van Creveld syndrome, also known as chondroectodermal dysplasia, is a complex genetic disorder caused by the mutation of particular genes, characterized by dwarfism, polydactyly, abnormal nail structure, and dental malformations. Dental manifestations include enamel hypoplasia and hypodontia. Patients with Ellis-van Creveld syndrome may also have heart defects that would require specific diagnostic tools. The exact prevalence of this disorder remains poorly investigated because most risk groups are the Amish population, who refuse to share personal information voluntarily. We hereby present a case report of an eighteen-year-old young woman who presented with the chief complaint of poor appearance of teeth. The medical history includes ventricular hypertrophy and epilepsy. The patient had mandibular natal teeth that were removed. The first line of treatment was offered when the girl was seven, and standard check-ups and orthodontic manipulations were made within the last ten years. Overall, this case proves the importance of radiographic and physical examination, along with the multidisciplinary cooperation of dentists, surgeons, orthodontists, anesthetists, and cardiologists.

Keywords: Dental enamel hypoplasia; Dwarfism; Ellis-van Creveld syndrome; Epilepsy; Malformed nails; Natal teeth

Introduction

Ellis-van Creveld syndrome is an atypical and complex genetic disorder that is characterized by additional fingers or toes, abnormal development of nails and teeth, and skeletal transformations. The leading cause of this disorder is a mutation of genes (EVC1 and EVC2). These gene mutations result in the production of abnormally small EVC and EVC2 proteins. Ellis-van Creveld (EVC) syndrome is thought to be due to mutation in the EVC gene and a nonhomologous EVC2 gene located in chromosome 4p16 head-to-head configuration.[1]All these mutations are commonly observed in people who have the classic phenotype. In addition to physiological abnormalities in the person's fingers and toes, this syndrome's characteristic features include bilateral ulnar polydactyly and short, usually disproportionate, length of long bones.[2]

Ectodermal abnormalities such as the limited growth or absence of hair on some parts of the body (e.g., no eyebrows).[2] Heart defects are explained by the abnormal condition of the upper heart chambers.

The first case was reported by Ellis and van Creveld in 1940 and was termed chondroectodermal dysplasia or meso-ectodermal dysplasia.[2] As a genetic disorder, Ellis-van Creveld syndrome was thoroughly studied among siblings and relatives to identify common (partial cleft lip and partial anodontia) and distinctive (extra fingers or toes) features and symptoms.[3] Its prevalence varies from 1 to 7 in one million in the general population, but some communities (Amish) do not always report this disorder officially, and all the cases remain poorly investigated.[4] In a majority of patients with this diagnosis, lack of motor development and intelligence delays are observed.[4] However, nearly half of this population do not survive childhood due to cardiopulmonary malformations.[4] Other distinctive features of this condition may include bowed legs with several toes showing syndactyly and stunted growth.[3] In this report, attention will be paid to one particular patient to investigate the main signs, family history, diagnostic and assessment techniques, and available treatment plans for Ellis-van Creveld syndrome.

Case Presentation

An eighteen-year-old Arab girl was admitted to the Orthodontics clinic, complaining of the poor appearance of upper teeth. While the patient was the sixth child born to her parents, she was the third child who was born and diagnosed with Ellis-van Creveld syndrome in her family. The girl's elder brother was diagnosed with Ellis-van Creveld syndrome had passed away at birth. Parental consanguinity was reported. Informed consent was obtained from the patient, and her parent (father) signed the letter. In the letter, the patient gave her consent for images and other clinical information related to her case to be used by the researcher. She underlined that she understood that her name and initials would not be mentioned in the study, and her anonymity and confidentiality would be maintained. The material (medical records and photos) was defined as available to the general public, and no concerns from the patient or her family were reported.

History

The patient had mandibular natal teeth, which exfoliated around the age of three months. Since the patient presented with polydactyly, the additional digits were surgically removed immediately after birth. The child was treated in the neonatal intensive care unit for 40 days under observation due to ventricular hypertrophy. The patient was diagnosed with epilepsy at a younger age and prescribed appropriate medication for eight months; no further signs of epilepsy were noticed. The first visit to the dental and maxillofacial center was in 2008, at the age of 7 years. Multiple primary carious teeth, as well as hypodontia, were identified. Carious teeth were conservatively restored, and a primary mobile tooth was extracted.

Examination

The patient's clinical features were characterized by dwarfism, hypoplastic nails, short ribs, and facial asymmetry slightly to the left. The extra-oral examination of the patient's temporomandibular joint revealed clicking on the right side. Moderate skeletal base with a competent lip and increased lower facial height was observed. Intraoral examination showed that the patient had hypodontia with missing mandibular right and left central and lateral incisors and maxillary right lateral incisor and left central incisors. The child had retained deciduous maxillary left lateral incisors and mandibular right lateral incisors. Furthermore, the patient had a peg-shaped maxillary right central incisor while the upper right canine presented with a Talon cusp. The primary purpose of managing the talon cusp is the reduction of the cusp and the developmental grooves. Gradual grinding with air abrasion was recommended to treat this anomaly and promote orthodontic retraction of the teeth.

The patient had a wide maxillary and mandibular labio-gingival frenulum, prominent lower anterior edentulous ridge, normal overbite, and a reverse overjet of 3mm. Moreover, palatal displacement of the maxillary left second premolar, class III incisor relationship, and class 3 canine and molar relationships were observed. The patient was seen by an orthodontist at the age of 17 years in the year 2018 for her dental evaluation. Routine follow-ups

with an orthodontic clinic and consideration of orthognathic surgery were recommended. All the third molars (18, 28, 38, and 48) were extracted in the year 2019 under general anesthesia by a panel of specialists, *viz*. an oral surgeon, a cardiologist, and an anesthetist.

Radiographic Features

In the year 2015, dental panoramic tomography showed that the patient had all permanent teeth, while the mandibular incisors of both sides (41, 42 and 31, 32) and maxillary right lateral incisor (12) were missing. The patient had retained deciduous mandibular right lateral incisor and maxillary left deciduous lateral incisor. A dental follicle in the location of the mandibular right third molar was seen, and its radiographic appearance was suggestive of malformation of 48. In general, there are many internal and external factors that define the necessity of panoramic radiography in children, including their general health condition and inborn syndromes and abnormalities.[1] Still, in a majority of cases, the first routine check-up is recommended at the age of 7 or 8 years, which coincides with the eruption of permanent dentition.[5] Some patients need to take this procedure at the age of 5 years to track the growth of all teeth and determine abnormalities, if any.

A lateral cephalometric radiograph was taken in 2019; it showed class III incisor relationship to moderate skeletal three bases with increased lower facial height as described previously. Cone-beam computed tomography (CBCT) was taken in 2019. It revealed the presence of all permanent teeth except the mandibular left and right central and lateral incisors and the maxillary right lateral incisor. There were retained maxillary left deciduous lateral incisors (62) and mandibular right deciduous lateral incisors (82). It also showed the lower edentulous area in the mandibular incisor region, malformed upper right central incisor (11), and occlusal radio-opacity of maxillary right first molar (16) suggestive of a dental amalgam restoration.

At the initial diagnostic stages, several provisional diagnoses were revealed due to the presence of common symptoms. For example, Weyers syndrome is also associated with poor development of the patient's teeth, nails, and bones. However, in the case of Weyers disorder, the patient would present with small teeth or a reduced number of front teeth.[1,2] In the patient under analysis, all teeth were developed, and their extraction was due to extensive caries and physiological changes such as natural weakness. Therefore, the final diagnosis, Ellis-van Creveld syndrome, was adequately justified, relying on radiographic results and other diagnostic means.

Treatment

At the age of 7 years, the first line of treatment was carried out at the pedodontics clinic and followed up by the general dental practitioner for prevention of dental caries to improve oral hygiene and dietary analysis. At the age of 17 years, orthodontic treatment was initiated for the patient, following counsel regarding its risks and benefits. The girl was still undergoing fixed orthodontic appliances at the time of writing this case report. The care plan for this case involved a combination of several treatment approaches. Restorative treatment for malformed right central incisors and restoration of deciduous teeth to appear like permanent teeth was combined with prosthetic treatment for replacement of missing teeth and orthognathic surgery by the maxillofacial surgeon.

Discussion

The complexity of Ellis-van Creveld syndrome is explained by the emergence of several physiological changes in the patient. In most cases, symptoms were observed during the first physical examination of the child or the gestational period. These changes included the presence of extra fingers (polydactyly), dwarfism (Figure 1), nail hypoplasia (Figure 2), and facial abnormalities such as asymmetry of the right or the left side of the face.[1,5-6] Radiological prenatal ultrasound during the gestational period was the main diagnostic tool. Nevertheless, panoramic radiography could also be used at a certain age.[7,7] Medically, certain cardiovascular problems could be detected in about 50% of patients, and chest x-rays, electrocardiography, and electrocardiograms had to be recommended to analyze possible malformations and to offer a treatment plan.[8] In addition to the above-mentioned features, the patient with Ellis-van Creveld syndrome exhibited some dental

features such as hypodontia and supernumerary teeth (Figure 3).[1]

Dental panoramic tomography was an important aid in diagnosis dentally (Figure 4).[1] This method showed that the patient with the syndrome under consideration could have all permanent teeth, but the maxillary lateral incisors and canines were absent or removed in childhood.[1,9] In the case of patients with polydactyly or dwarfism, differential diagnoses such as Majewski syndrome, Beemer-Langer syndrome, Saldino-Noonan syndrome, and Asphyxiating Thoracic Dystrophy are to be considered during a clinical examination.[1]The reason for the offered syndromes' differentiation lay in their etiology, being related to mutations of the same genes. The differences of these conditions matter and have to be recognized regarding their main characteristics. For example, the Majewski syndrome is characterized by polydactyly, which is also observed in the syndrome under analysis.[1] However, its main features are a dysmorphic face, short ribs, and a disproportionate head. Children with the Beemer-Langer syndrome have narrow thorax and horizontally oriented ribs. In the Saldino-Noonan syndrome, hydropic appearance is observed, and Asphyxiating Thoracic Dystrophy presents with short legs, hands, ribs, and stature in general.

Patient and parent education, prevention strategies play a critical role in the management of oro-dental structures. It is advised to visit dental clinics regularly and evaluate the dental status.[7] Treatment in such cases would demand a multidisciplinary team (orthodontic, pediatric, oral surgery, and restorative). Therefore, early diagnosis and prudent treatment plans might contribute to a successful outcome. However, orthodontic input was the major element of dental health care. In this case, prevention was established in line with the Department of Health Preventative Toolkit, which included using fluoridated toothpaste with at least 1350ppm and reducing the frequency and amount of sugary food and drinks.[10]

Conclusion

The management of patients with Ellis-van Creveld syndrome is associated with an effort to gather a multidisciplinary team. This disorder was characterized by a complex nature, including several physiological changes like facial expression, extremity abnormalities, and heart-related problems, and associated emotional concerns. A majority of studies were developed to provide people with enough credible information but not to explain how to predict this disorder. However, with the help of the available diagnostic tools, examination, guidelines, and professionals of healthcare workers, it became possible to reveal other problems and identify the most effective treatment. To conclude, Ellis-van Creveld syndrome is a rare genetic disorder that produces significant physiological changes. Dental management aims to provide a high-quality operative treatment in collaboration with a panel of specialists.

Conflicts of interest

The authors of this study have no conflicts of interest to declare.

Ethical Approval

Approved by the Research Ethical Committee, Bahrain Defence Force Hospital, Bahrain.

Figure 1: Features of dwarfism

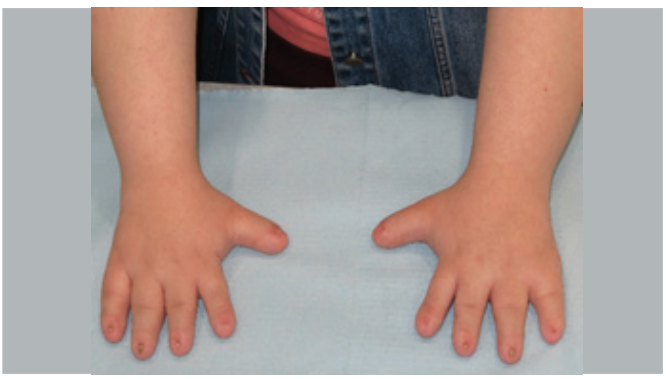

Figure 2: Hands with hypoplastic nails

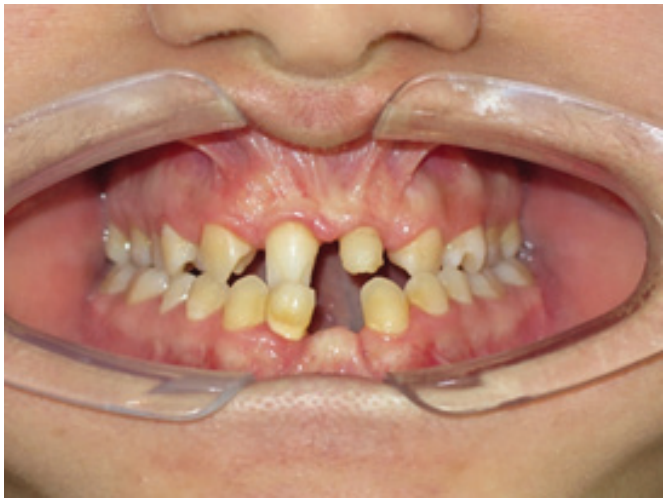

Figure 3: Malformed maxillary central incisors (11 and 21), missing mandibular right and left central and lateral incisors (31, 32 and 41, 42) as well as maxillary right lateral incisor (12) and left central incisors (21) peg-shaped maxillary right central incisor (11)

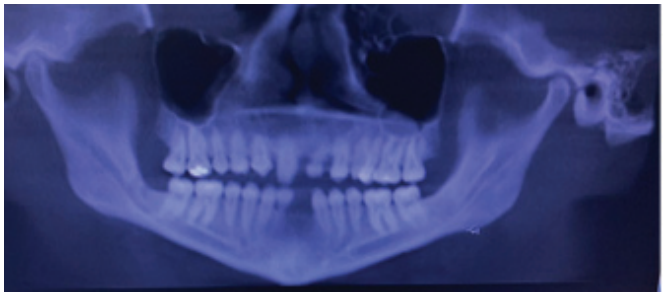

Figure 4: Dental panoramic tomography

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