Ellis-van Creveld Syndrome: Report of Two Cases

Altaf Hussain Chalkoo, Mohsin Muzaffar Tak

1Head, Department of Oral Medicine and Radiology, Government Dental College, Srinagar, Jammu and Kashmir, India
2Lecturer, Department of Oral Medicine and Radiology, Government Dental College, Srinagar, Jammu and Kashmir, India

Correspondence: Altaf Hussain Chalkoo, Head, Department of Oral Medicine and Radiology, Government Dental College Srinagar, Jammu and Kashmir, India, e-mail: drchalkoo_omar@yahoo.co.in

INTRODUCTION

Ellis-van Creveld syndrome was described by Richard WB Ellis (1902-1966) of Edinburg and Simon Van Creveld (1895-1971) of Amsterdam, McCusick et al (1964) followed up with its incidence in Amish population.

Ellis-van Creveld syndrome, also called chondroectodermal dysplasia or mesoectodermal dysplasia, is a rare genetic disorder of the skeletal dysplasia type. It involves numerous anomalies including postaxial polydactyly, congenital heart defects (most commonly an atrial septal defect producing a common atrium, occurring in 60% of affected individuals), prenatal tooth eruption, fingernail dysplasia, short-limbed dwarfism, short ribs, cleft palate and malformation of the wrist bones (fusion of the hamate and capitate bones). Ellis-van Creveld syndrome often is the result of founder effects in isolated human populations, such as the Amish and some small island inhabitants. Although relatively rare, this disorder does occur with higher incidence within founder effect populations due to lack of genetic variability. Observation of the inheritance pattern has illustrated that the disease is autosomal recessive, meaning that both parents have to carry the gene in order for an individual to be affected by the disorder.

Ellis-van Creveld syndrome is caused by a mutation in the EVC gene as well as mutation in a nonhomologous gene EVC2 located close to the EVC gene in a head to head configuration. The EVC gene maps to the chromosome 4 short arm. The function of a healthy EVC gene is not well understood at this time. Ellis-van Creveld syndrome is nowadays thought to be related with emerging class of diseases called ciliopathies. The underlying cause may be a dysfunctional molecular mechanism in the primary cilium structures of the cell organelles which are present in many cellular types throughout the human body. The cilia defects adversely affect numerous critical developmental signaling pathways’ essential to cellular development and thus offer a plausible hypothesis for the often multisystem nature of a large set of syndromes and diseases. Known ciliopathies include primary ciliary dyskinesia, Bardet-Biedl syndrome, polycystic kidney and liver disease, nephronophthisis, Alstrom syndrome, Meckel-Gruber syndrome and some forms of retinal degeneration.

CASE REPORTS

Case 1

A female patient aged about 10 years who was second child of her parents reported to department with the complaint of malposed teeth and needed correction for the same. Upon history/examination it was noticed that patient had undergone surgery for having single atrium at Apollo Hospital, New Delhi, when she was around 6 years of age. Patient had six digits instead of five in hands (Fig. 1). Nails were hypoplastic in both hands and feet (Fig. 2). Her legs were shortened...
Ellis-van Creveld Syndrome: Report of Two Cases


S435

DISCUSSION

All embryonic layers appear involved in Ellis-van Creveld syndrome. The signs of ectodermal dysplasia seem to affect nails, teeth and gums only although some cases show eye and neural involvement.\(^{11,12}\) Abnormalities of the bones, heart, kidneys indicate the involvement of mesodermal involvement.\(^{13,14}\) Endodermal involvement is not very common but some patients with lung and liver abnormalities have been reported in literature. Ellis-van Creveld syndrome presents phenotypic variations. Isolated findings in near relatives such as polydactyly, short stature or abnormalities of wrist bones without other stigmata of the syndrome have been frequently described. The patients present with partial harelip and maxillary alveolar clefts and continuous frenum could be related to a partial failure in the normal development of the embryonic median nasal process.\(^{15,16}\)

Caffey described constant and inconstant features. The constant features include hypoplasia of teeth and nails, progressive shortening of the bones of the arms and legs, bilateral polydactyly, polymetacarpalia and synmetacarpalia, hook-like bilateral fusion of the capitate and hamate bones, delayed maturation of the primary ossification centers of phalanges with accelerated maturation of the secondary epiphyseal cartilages and characteristic deformities of the proximal end of the tibiae. The inconstant features are alopecia, joining of the upper lip and gum, congenital cardiac anomalies, dislocation of the head of the radius, delayed maturation of sesamoid bones in the hand and pedal polydactyly, syndactyly and polymetatarsalism.\(^{17}\)

The cases which are reported also showed malocclusion, partial anodontia (deciduous and permanent dentition), genital abnormalities and no hepatic or renal abnormalities were noticed in this case.

Case 2

A female patient of around 16 years of age with retained deciduous anterior teeth in lower arch B and absent centrals in lower arch, malformed upper centrals (bell-shaped) (Fig. 5) and absent laterals in upper arch. Although other features of the patient appeared normal. Her digits in hands were six in each hand (Fig. 6) and nails of both hands and feet were hypoplastic (Fig. 7). There was marked deformity of knees. She was having congenital heart defect in the form of atrial septal defect, which was surgically corrected when the patient was 4 years of age. Other features of the patient like intelligence were normal and pelvic dysplasia was noticed in this case. Patient was also second child of her parents. Parents were normal and had four children, two males and two females. Only the patient had developed features as described. No natal teeth, no clefts at the time of birth. Normal skin and hair, no

Fig. 2: Hypoplastic nails of feet

(see Fig. 2) and her gait was abnormal. The color of skin and hair was normal with normal intelligence rate. The patient's parents were nonconsanguineous and normally developed and the patient was their second child. The first child was normal without any anomaly. Pregnancy and birth were uneventful. No natal teeth or genital abnormalities were found at the time of birth except newborn at that time showed polydactyly of hands (hexadactilia of both hands) and relatively short length of legs noticed. Her head morphology and facial appearance were normal as was the quantity of the hair growth (although shaved at the time of reporting to department but could be made out from hair outline). No renal or hepatic disorders were present in this case.

Her oral examination revealed anodontia (absence of permanent incisors, laterals in upper arch). Patient’s labial vestibule in upper jaw was not visualized because of adherence of alveolar mucosa with labial mucosa (Fig. 3), generalized caries and absence of permanent centrals and laterals and retained B. The OPG revealed absence of anterior permanent teeth (Fig. 4).

Fig. 3: Labio-alveolar mucosa adherence

Case 2
Altaf Hussain Chalkoo, Mohsin Muzaffar Tak

Retarded eruption, malformed teeth, fraenula hypertrophy (the midline puckering of upper lip is seen in case 1). General features included normal intelligence rates, short stature, polydactyly hands, dysplastic nails, congenital heart diseases, normal skin and hair color (pelvic dysplasia was seen in case 2 only).

In full-blown clinical form of Ellis-van Crevelled syndrome is easily diagnosed but in absence of some of the most relevant general manifestation some of the oral signs are so peculiar that they form the vital clue for the diagnosis and further counseling of such patients. Both the cases were properly counseled as they needed multidisciplinary dental treatment for rectification of their dental needs.

SUMMARY

Two cases of chondroectodermal dysplasia (Ellis-van Creveld syndrome) with a remarkable number of the classic oral, dental and general changes are described. This syndrome involves all embryonic tissue layers and is polysymptomatic. Some of the oral and dental manifestations are pathognomonic and must be considered in primary diagnostic criteria. However, in some patients the oral and dental manifestations are also minor and the oral medicine specialists must interpret them adequately.

REFERENCES


