Ellis-Van Creveld Syndrome

Alternative Names
EVC
Chondroectodermal Dysplasia
Mesoectodermal Dysplasia

WHO International Classification of Diseases
Congenital malformations, deformations and chromosomal abnormalities

OMIM Number
225500

Mode of Inheritance
Autosomal recessive

Gene Map Locus
4p16, 4p16

Description
Ellis-van Creveld (EVC) syndrome or chondroectodermal dysplasia is a rare autosomal recessive disorder characterized by a variable spectrum of clinical findings. Classical Ellis-van Creveld syndrome comprises a tetrad of clinical manifestations of chondrodys trophy, polydactyly, ectodermal dysplasia, and cardiac defects. In several case reports, dysplasia involving other organs has also been identified. Oral manifestations tend to be pathognomonic such as multiple broad labial frenula and congenital missing teeth. Hematologic abnormalities have been rarely reported in patients with Ellis-van Creveld syndrome.

Ellis-van Creveld syndrome and Jeune's asphyxiating thoracic dystrophy are related disorders. Some patients have overlapping features of both disorders, indicating that these syndromes may be a part of a disease spectrum. Nephronophthisis has been occasionally reported in patients with asphyxiating thoracic dystrophy, but not with Ellis-van Creveld syndrome.

Molecular Genetics
Mutations in the Leucine-Zipper (EVC) gene have been reported in many individuals with Ellis-van Creveld syndrome as well as Weyers acrodental dysostosis. The EVC gene includes 21 coding exons encoding a 992-amino acid protein.

Epidemiology in the Arab World

Egypt
Mostafa et al. (2005) reported three Egyptian families with six cases of Ellis-Van Creveld syndrome. Mostafa et al. (2005) observed an unusual pattern of inheritance with father to son or to daughter transmission in two consanguineous families; thus, demonstrating pseudo-dominant inheritance, probably for the first time in the literature. A new consistent orodental anomaly found in all our cases was bifid tip of the tongue. Mostafa et al. (2005) emphasized the study of orodental anomalies in future cases for accurate diagnosis of Ellis-van Creveld syndrome and its probable differential diagnosis from Weyers acrodental dysostosis.

Jordan
Hattab et al. (1998) presented two siblings with Ellis-Van Creveld, a boy aged 9 years and a girl aged 7 1/2 years, a product of unaffected first cousin parents. The patients exhibited chondrodysplasia of tubular bones resulting in disproportionate dwarfism, polydactyly and syndactyly of hands and feet, severe dystrophic nails, multiple broad labial frenula with abnormal attachments, congenital missing incisors, anomalous teeth, bilateral partial clefts of the alveolar bone, and malocclusion. Other features noted in either cases were: congenital heart defect, median notch of the upper lip, shovel-shaped incisors and taurodontism. Of the unusual dental findings observed in the patients of Hattab et al. (1998) were talon cusp, reduced crown size, supernumerary tooth, and early
eruption of teeth. Hattab et al. (1998) pointed that because half of the cases with Ellis-Van Creveld syndrome have cardiac malformation, dental treatment must be performed under prophylactic antibiotic coverage. They also emphasized the important role of dentists in early diagnosis and control of dental problem of this condition.

Saudi Arabia
Reddy and Madelioglu (1967) reported two cases of Ellis-Van Creveld syndrome from Saudi Arabia. No further details could be obtained on these patients.

United Arab Emirates
In a 5-year prospective study for newborns at Al Ain Medical District, Al-Gazali et al. (2003) defined the pattern and birth prevalence of the different types of osteochondrodysplasias in the United Arab Emirates. Among the 38,048 births during the study period, 36 (9.46/10,000 births) had some type of skeletal dysplasia of which two had Ellis-Van Creveld syndrome. In one of these two cases, the parents were consanguineous. Al-Gazali et al. (2003) calculated the birth rate of this type of osteochondrodysplasia in the United Arab Emirates to be 0.52/10,000 births.

Al Talabani et al. (1998) studied the pattern of major congenital malformations in 24,233 consecutive live and stillbirth at Corniche hospital, which is the only maternity hospital in Abu Dhabi, between January 1992 and January 1995. A total of 401 babies (16.6/1,000), including 289 Arabs, were seen with major malformation. Single gene disorders accounted for 24% of the cases, 76% were due to autosomal recessive disorders. In their study, Al Talabani et al. (1998) observed one case of Ellis-Van Creveld syndrome born to a first cousin couple from the United Arab Emirates with no recurrent case in their family. Al Talabani et al. (1998) concluded that their study was very close to representing the true incidence of congenital abnormalities in the whole United Arab Emirates, as they investigated over 98% of deliveries in Abu Dhabi, the capital of United Arab Emirates.

References

Contributors
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