Ellis-van Creveld Syndrome

Fahad S. Al Qooz, BDS, MFD RCS (Ireland)* Rana Al Ghatam, Orthodontics Doctorate (Bristol), MOrth RCS (Edinburgh), BDS (London), MFD RCS (Ireland), BSc (Boston)**

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.

Bahrain Med Bull 2018; 40(4): 242 - 244