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# Ellis-van Creveld Syndrome: Case Report and Review of the Literature

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*Ellis-van Creveld syndrome (EVCS) is a rare, autosomal recessive disorder. We present a 9-year-old boy who was referred to the dermatology clinic for evaluation of congenital nevi. His history was consistent with the classic tetrad of EVCS. We discuss this potentially serious condition with congenital heart malformations that can result in failure to thrive and even death if not recognized early.*

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## Case Report

A 9-year-old boy was referred to the dermatology clinic for evaluation of congenital nevi. The patient's medical history was notable for genu valgum, cardiac surgery to repair complex congenital heart defects, and surgical repair of polydactyly of both hands. The patient was not born of consanguineous parents but did have a family history of distant relatives with stillborn infants.

On physical examination, the patient's height was well below the one-third percentile. Extensive scarring was evident on the anterior chest and bilateral lower extremities due to surgical repair of genu valgum and cardiac defects (Figure 1). There was no circumoral or distal extremity cyanosis. The patient's fingernails were small and dystrophic (Figure 2). The ulnar aspect of both hands demonstrated surgical scars overlying bony protrusions due to surgical repair of polydactyly. In comparison to the patient's torso, all extremities were shorter than

expected. The patient was unable to make a full fist. Oral examination revealed missing and cone-shaped teeth. The anterior upper labiokingival sulcus was obliterated by multiple frenula (Figure 3).

## Comment

First recognized by Ellis and van Creveld in 1940,<sup>1</sup> Ellis-van Creveld syndrome (EVCS) is a rare, autosomal recessive, chondroectodermal dysplasia caused by mutations of the Ellis-van Creveld syndrome genes, *EVC* and *EVC2*, in a chromosomal region of several other skeletal disorders (achondroplasia).<sup>2</sup> Although the exact prevalence remains unknown, the syndrome is most prevalent in the Amish population in Lancaster County, Pennsylvania, occurring in 5 per 100 live births.<sup>3</sup>

The characteristic tetrad of EVCS consists of chondrodysplasia, ectodermal dysplasia, polydactyly, and congenital heart disease.<sup>1</sup> Small stature with symmetric distal limb shortening is typical in patients with EVCS. Due to the shortened distal phalanges, patients frequently are unable to make a full fist, as was the case with our patient. Distal rather than proximal shortening differentiates EVCS from the achondroplasia phenotype.

Ectodermal dysplasia is evident in 70% of cases.<sup>4</sup> A vast array of dental abnormalities include cone-shaped teeth, hypodontia, enamel hypoplasia, fused incisors, molars with extra cusps, dental fissures and pits, and neonatal teeth. Fingernails are small and dystrophic. Sparse thin hair seems to be quite variable among cases. Although not of ectodermal origin, associated oral cavity anomalies such as multiple labiokingival frenula are common findings, as was seen in our patient. The frenula obliterate the anterior upper labiokingival sulcus. Additional oral manifestations include malocclusion, labiokingival adherences, gingival hypertrophy, and labiokingival frenulum hypertrophy.<sup>4</sup>

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**Figure 1.** A 9-year-old boy with scarring on the anterior chest and bilateral lower extremities due to surgical repair of genu valgum and cardiac defects.

Bilateral postaxial (ulnar aspect) polydactyly of the hands is almost invariable and 10% of cases have polydactyl feet.<sup>5</sup> Congenital heart malformations are present in 50% to 60% of cases, including common atrium, defects of the mitral and tricuspid valves, patent ductus arteriosus, ventricular septal defect, atrial septal defect, and hypoplastic left heart syndrome. Cardiac anomalies are the major determinant of longevity.<sup>6</sup>

Ellis-van Creveld syndrome can affect other organs, as there have been additional findings of strabismus, thoracic wall, and pulmonary malformations resulting in respiratory problems, hypospadias and epispadias, and cryptorchidism. Reports of rare renal abnormalities include agenesis, nephrocalcinosis, and megaureters.<sup>4</sup> Hematologic abnormalities have only been reported in 2 cases, one with dyserythropoiesis<sup>7</sup> and the other with perinatal myeloblastic leukemia.<sup>8</sup> Although there have been reports of central nervous system anomalies and mental retardation in patients with EVCS, most patients have unaffected intelligence.<sup>9</sup>

Differential diagnosis includes several syndromes that cause dwarfism. Cartilage-hair hypoplasia is an autosomal recessive syndrome that consists of



**Figure 2.** The patient's fingernails appeared small and dystrophic due to surgical repair of polydactyly.



**Figure 3.** The anterior upper labiogingival sulcus was obliterated by multiple frenula.

short-limb dwarfism, sparse hair, soft doughy skin, small dystrophic fingernails, and T-cell immunodeficiency. A history of cytopenia and susceptibility to frequent viral infections with an absence of polydactyly and cardiac defects assist in differentiating cartilage-hair hypoplasia from EVCS.

Diagnosis of EVCS is based on clinical manifestations and radiologic evaluation of skeletal features. Molecular diagnosis for mutations of the *EVC* and *EVC2* genes is definitive.<sup>4</sup> Management of patients with EVCS requires a multidisciplinary team including pediatric, orthopedic, orthodontic, cardiac, and possibly pulmonary care.

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