

## Ellis-van Creveld syndrome in a neonate: a case report

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### Abstract

Ellis-Van Creveld Syndrome (EVC) is a rare genetic disorder with autosomal recessive inheritance, caused by mutations in two genes, EVC1 and EVC2 in the 4p16 chromosome. The exact prevalence of EVC is unknown and is estimated at approximately seven per million. It affects males and females equally. It is a constellation of four findings, including chondrodysplasia, polydactyly, ectodermal dysplasia, and congenital heart defects. Our case was unique as it had left inguinal hernia, short phallus, hyperpigmented scrotum, cryptorchidism, and other defining features of this syndrome. A multidisciplinary team managed this patient with regular follow up. Only six cases have been reported in Pakistan, and only one of them was reported in a neonate. This report highlights the importance of timely and proper multidisciplinary management of such disorders for better outcomes. It will also create awareness among medical professionals and will help them to identify promptly.

**Keywords:** Ellis-Van Creveld syndrome, Chondroectodermal dysplasia, Skeletal disorders.

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### Introduction

Ellis-Van Creveld Syndrome (EVC), also known as chondroectodermal dysplasia or meso-ectodermal dysplasia, is a rare genetic disorder with autosomal recessive inheritance. The pediatricians Richard W.B. Ellis and Simon Van Creveld made the first description of this syndrome.<sup>1</sup> It results from genetic mutations in EVC1 and EVC2 genes in the 4p16 chromosome.<sup>1</sup> The actual prevalence of EVC is unknown and is estimated at approximately seven per million.<sup>2</sup> Consanguinity in parents is reported for 30% of the cases.<sup>3</sup> The four principal manifestations include chondrodysplasia, polydactyly, ectodermal dysplasia, and congenital heart defects.<sup>4,5</sup> Characteristic oral manifestations include

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malformed teeth, supernumerary teeth, enamel hypoplasia, and neonatal teeth in about 30% of cases.<sup>6,7</sup> Malocclusion, absent vestibular sulcus, submucous clefts, premature exfoliation of the teeth, hypertrophic frenula, and labial frenula are also seen.<sup>6,7</sup> We report a case of a neonate with similar features suggestive of Ellis-Van Creveld Syndrome. The patient was presented with several clinical features, typical to Ellis-Van Creveld Syndrome. Publishing this case report would benefit health professionals in the region and lead to early diagnosis and management of the disorder.

### Case Report

A one-day-old male baby, delivered at term by C-section, presented to the paediatric emergency department of Nishtar Hospital, Multan in July 2021 with complaints of cyanosis and respiratory distress. The patient was the seventh child of healthy parents of consanguineous marriage, and family history was significant for miscarriage of unknown reason. Anthropometric measurements of the baby at birth were; length 45 centimeters (cm), weight 2.5 kilograms (kg), and head circumference 33cm. Physical examination of the patient revealed widely open anterior and posterior fontanelles, inadequately fused sagittal sutures, low set ears, high arched palate, depressed nasal bridge, dysplastic nails, widely spaced nipples, rhizomelic shortening,



**Figure-1:** Rhizomelic shortening, depressed nasal bridge, widely spaced nipples, undescended testes.

undescended testis, inguinal hernia, short phallus, and contracture at the knee and hip joints. (Figure1).

Echocardiogram revealed pulmonary atresia with subaortic ventricular septal defect, transposition of great arteries, and moderate patent ductus arteriosus. Ultrasonography of abdomen and pelvis showed epiphyseal dysplasia, slightly heterogeneous and ill-defined femoral heads, ill-defined labrum and acetabular roof, alpha angle on right side 51 degrees and left side 54-degree representing physiological immaturity. A skeletal survey of the patient revealed ileal spur, short proximal long bones, metaphyseal widening, slightly enlarged cardiac silhouette, and mild thickening of clavicles. (Figure 2) Baseline blood tests and EKG reports were unremarkable. The septic screen was negative, and blood counts were normal Based on clinical examination, echocardiographic and radiological findings, a diagnosis of Ellis Van Creveld was made.



**Figure-2:** Skeletal survey of the patient with Ellis-Van Creveld Syndrome. There is a bony spur at the acetabulum, short proximal long bones, and metaphyseal widening

Initially, the patient was treated with Intravenous (IV) fluids, IV antibiotics, and oxygen with nasal prongs for 3 days. The patient was shifted to intermittent oxygen and Oro-gastric (OG) feeding for two days when cyanosis and respiratory distress improved. After five days, the patient was oxygen-free and was shifted to oral feeding. The orthopaedic surgeon was called for correction of contracture of the knee and hip joints, and he advised exercises. The paediatric surgeon was also consulted for

an evaluation of hernia and cryptorchidism, and he suggested an operation at one year of age. We discharged the patient on the seventh day of life, and he was referred to the outpatient care for further better management of cardiac defects. Parents were also counseled regarding the nature of the disease. Unfortunately, the patient died on the 12th day of discharge due to problems secondary to his complex cardiac disease. This shows the unfavourable prognosis of this disorder involving complex cardiac defects.

## Discussion

Chondroectodermal dysplasia, another name for Ellis van Creveld syndrome, is a rare autosomal recessive disorder affecting males and females equally. They are more common in families with a history of consanguinity.<sup>8</sup> Sughra Wahid et al. reported n postaxial polydactyly involving both hands' ulnar side and other typical features in a neonate. While in our case only significant finding in hands was hypoplastic nails with long fingers. The fate of the case by Sughra Wahd et al. was not mentioned.<sup>8</sup>

In a case report by Ikramullah Khan et al., oral manifestations including small peg-shaped teeth, high arched palate, obliterated sulcus between upper lip and gum, and multiple frenula were observed in a 15-year-old girl with EVC syndrome. While in our case, we found only a high arched palate with a small tongue.<sup>9</sup> Our case was unique as it had left inguinal hernia, short phallus, hyperpigmented scrotum and cryptorchidism, and other defining features of this syndrome. The fate of this case by Ikramullah Khan et al. was not mentioned.

The comprehensive management of the patient with EVC syndrome should be carried out by a multidisciplinary team involving paediatrician, neonatologist, cardiologist, orthopaedic surgeon, paediatric surgeon, and odontologist. Ideally, it should be started from the prenatal stage, identifying phenotypic characteristics suggestive of the syndrome. It is essential to identify this disease early for better outcomes. As Ellis Van Creveld syndrome follows an autosomal recessive inheritance, it is essential to carry out early genetic counseling of the parents. The insufficient local literature about such conditions either means physicians' ignorance about this condition or a decrease incidence of such disorders.

## Conclusion

EVC syndrome is a rare genetic disorder with a high mortality rate in early life. As in this case, the patient died twelve days after discharge due to complex cardiac defects. Therefore, early diagnosis and timely intervention by the multidisciplinary approach is essential

for survival.

**Consent:** Consent was obtained from parents for publication of this case report and accompanying images.

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