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Case Report

Neonatal Diagnosis of Ellis-Van Creveld Syndrome: A Case Report

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Received: July 16, 2026; **Published:** July 30, 2026

Abstract

Ellis-Van Creveld syndrome (EVCS) is a rare autosomal recessive disorder characterized by disproportionate short stature, postaxial polydactyly, ectodermal dysplasia, and congenital heart defects in approximately 50% - 60% of affected individuals. Early diagnosis is essential for appropriate neonatal management and genetic counseling.

We report the case of a premature female neonate born at 34 weeks of gestation to first-degree consanguineous parents. At birth, she presented with postaxial polydactyly, nail dysplasia, sparse light-colored hair, a short neck, a narrow thorax, and moderate ascites. Laboratory investigations were unremarkable, and both cardiac and abdominal ultrasonography showed no abnormalities. The diagnosis of Ellis-Van Creveld syndrome was confirmed by genetic testing. The newborn received supportive neonatal care with close respiratory and nutritional monitoring and remained clinically stable during hospitalization.

This case highlights the importance of recognizing the characteristic clinical features of Ellis-Van Creveld syndrome during the neonatal period, even in the absence of congenital heart disease, to facilitate early diagnosis, multidisciplinary management, and appropriate genetic counseling.

Keywords: *Ellis-Van Creveld Syndrome; Neonate; Postaxial Polydactyly; Ectodermal Dysplasia; Congenital Anomalies*

Abbreviation

EVCS: Ellis-Van Creveld Syndrome

Introduction

Ellis-Van Creveld syndrome (EVCS), also known as chondroectodermal dysplasia, is a rare autosomal recessive disorder characterized by disproportionate short stature, postaxial polydactyly, ectodermal abnormalities (including nail and dental dysplasia), short ribs with a narrow thorax, and congenital heart defects, which are present in approximately 50% - 60% of affected individuals. The clinical presentation is highly variable, ranging from mild skeletal abnormalities to severe thoracic hypoplasia associated with respiratory compromise. The syndrome is caused by pathogenic variants in the EVC and EVC2 genes and demonstrates considerable clinical variability.

Prenatal diagnosis may be suspected by ultrasonography from the second trimester based on characteristic skeletal abnormalities, including short limbs, a narrow thorax, and postaxial polydactyly. Postnatal diagnosis relies on the recognition of the characteristic clinical features and may be confirmed by genetic testing. Early diagnosis is essential because the prognosis mainly depends on the severity of respiratory compromise related to thoracic hypoplasia and the presence of associated congenital cardiac anomalies.

Here, we report the case of a premature neonate diagnosed with Ellis-Van Creveld syndrome shortly after birth and discuss its clinical presentation, diagnostic evaluation, and management.

Case Presentation

A female preterm neonate was born at 34 weeks of gestation to first-degree consanguineous parents following an unsupervised fourth pregnancy (G4P4). Her birth weight was 2,000g, length was 44 cm, and head circumference was 33 cm. At birth, physical examination revealed multiple congenital anomalies, including postaxial polydactyly, nail dysplasia, sparse light-colored hair, a short neck, a narrow thorax, and moderate ascites (Figure 1-3). Her respiratory rate was 31 breaths/min and heart rate was 122 beats/min.

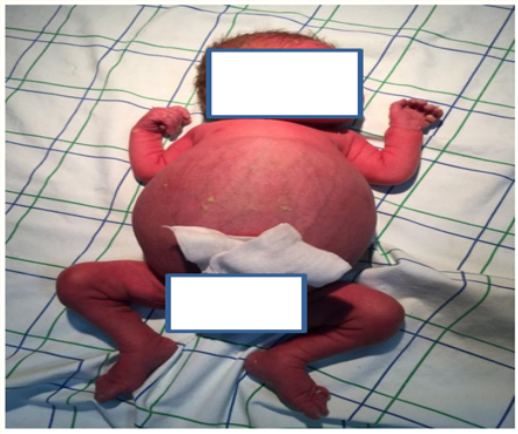

Figure 1: *Anterior view of the neonate showing marked abdominal distension due to ascites and the characteristic body habitus.*

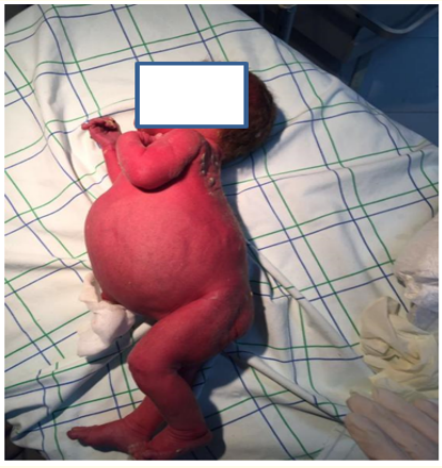

Figure 2: *Lateral view demonstrating a narrow thorax and abdominal distension.*

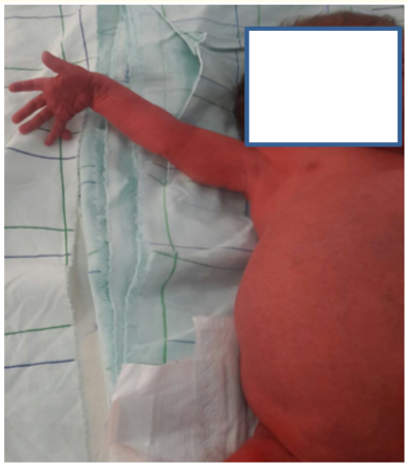

Figure 3: *Postaxial polydactyly of the hand.*

Initial laboratory investigations, including a complete blood count, C-reactive protein, renal and liver function tests, were within normal limits. Analysis of the ascitic fluid revealed no significant abnormalities. Chest radiography demonstrated short ribs, supporting the clinical findings of a narrow thorax. Cardiac and abdominal ultrasonography were unremarkable. Serological tests for toxoplasmosis, rubella, cytomegalovirus, herpes simplex virus, and syphilis were negative.

Considering the presence of multiple congenital anomalies, a genetics consultation was requested. Genetic testing confirmed the diagnosis of Ellis-Van Creveld syndrome by identifying a pathogenic variant in the EVC2 gene.

The neonate was admitted to the neonatal unit for close respiratory and nutritional monitoring. Her clinical course was favorable, and she was discharged after 28 days of hospitalization. Genetic counseling was provided to the family because of the autosomal recessive inheritance pattern of the disorder.

Discussion

Ellis-Van Creveld syndrome (EVCS), also known as chondroectodermal dysplasia, is a rare autosomal recessive disorder characterized by disproportionate short stature, postaxial polydactyly, ectodermal abnormalities, short ribs, and congenital heart defects, which occur in approximately 50% - 60% of affected individuals [1,5]. Recent data from the largest published EVCS cohort have further expanded the phenotypic spectrum of the syndrome and highlighted its marked clinical variability [12].

The diagnosis in our patient was suspected shortly after birth based on the combination of postaxial polydactyly, ectodermal abnormalities, a narrow thorax, and short ribs. Genetic testing confirmed the diagnosis. Similar neonatal and pediatric presentations have recently been reported, emphasizing the importance of early recognition to facilitate prompt diagnosis and multidisciplinary management [3,10,11].

Pathogenic variants in the EVC and EVC2 genes are responsible for Ellis-Van Creveld syndrome. These genes encode proteins involved in primary cilia function and Hedgehog signaling, a pathway essential for normal skeletal and ectodermal development. Disruption of this signaling pathway leads to the characteristic clinical manifestations of EVCS, including short stature, postaxial polydactyly, thoracic abnormalities, and ectodermal defects. Although pathogenic variants in both EVC and EVC2 produce a similar phenotype, considerable inter- and intrafamilial clinical variability has been reported, suggesting that additional genetic and environmental factors may influence disease expression [12-14].

Respiratory complications resulting from thoracic hypoplasia and congenital heart defects remain the main determinants of prognosis in patients with EVCS [5,8]. Although congenital cardiac anomalies are reported in approximately half of affected individuals, echocardiography was normal in our patient, illustrating the considerable phenotypic variability of this syndrome [12].

An unusual finding in our patient was moderate ascites at birth. Although ascites is not considered a classical manifestation of EVCS, extensive investigations, including analysis of the ascitic fluid, infectious screening, renal and liver function tests, and cardiac evaluation, failed to identify an alternative cause. Whether this finding represents an uncommon manifestation of EVCS or an incidental association remains uncertain.

Prenatal diagnosis may be suspected during the second trimester by ultrasonography when characteristic skeletal abnormalities, including short limbs, a narrow thorax, and postaxial polydactyly, are identified [2,6]. Following birth, confirmation by genetic testing and appropriate genetic counseling are essential because of the autosomal recessive inheritance pattern and the 25% recurrence risk in future pregnancies [4,7,12].

Management of EVCS requires a multidisciplinary approach involving neonatologists, pediatric cardiologists, orthopedic surgeons, dentists, geneticists, and rehabilitation specialists according to the patient's clinical manifestations. Recent case reports have also emphasized the importance of early diagnosis, multidisciplinary care, and regular follow-up to optimize patient outcomes [9-11].

Conclusion

Ellis-Van Creveld syndrome is a rare genetic disorder with variable clinical manifestations that should be considered in neonates presenting with postaxial polydactyly, ectodermal abnormalities, and a narrow thorax. Early recognition, appropriate genetic evaluation, and multidisciplinary management are essential for optimizing patient care and providing accurate genetic counseling.

This case highlights the phenotypic variability of the syndrome, particularly in the absence of congenital heart disease, and emphasizes the importance of considering Ellis-Van Creveld syndrome in the differential diagnosis of neonates with characteristic skeletal and ectodermal abnormalities.

Conflict of Interest

The authors declare no conflict of interest.

Funding Support

The authors received no financial support for the research, authorship, and/or publication of this article.

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Volume 15 Issue 8 August 2026