ASIDE Case Reports

Case Report

Ellis–van Creveld Syndrome with Common Atrium: A Clinically Diagnosed Case Report with Literature Review

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ABSTRACT

Ellis-van Creveld (EVC) syndrome is a rare, autosomal recessive disorder defined by a classic tetrad of features: chondroectodermal dysplasia causing disproportionate short stature, postaxial polydactyly, ectodermal defects affecting the nails and teeth, and congenital heart disease. The most significant cause of morbidity and mortality is the associated cardiac anomaly, most commonly a large atrial septal defect resulting in a functional common atrium. Early and accurate diagnosis is critical for managing cardiovascular risk and implementing appropriate long-term care.

We report the case of a 12-year-old female from Pakistan, with a weight of 26 kg (BMI 18.8 kg/m²), who presented with a classic EVC phenotype. Clinical examination revealed short-limb dwarfism, bilateral postaxial polydactyly of all four limbs, genu valgum, and ectodermal dysplasia. Her parents were non-consanguineous. Cardiovascular evaluation confirmed a common atrium via echocardiography, with corresponding ECG findings of right axis deviation and an incomplete right bundle branch block. Based on this distinct constellation of clinical and imaging evidence, a clinical diagnosis was made, as molecular genetic testing was not available.

This case confirms the importance of recognizing the distinct clinical phenotype of Ellis-van Creveld syndrome. In settings where molecular diagnostics are unavailable, a confident diagnosis can be made through thorough clinical assessment. Prompt identification is paramount for initiating multidisciplinary management, thereby improving the patient's long-term health and quality of life, with the patient currently awaiting surgical repair of her cardiac defect and referral for orthopedic and dental care.

1. Introduction

Ellis-van Creveld syndrome, also known as chondroectodermal dysplasia, is a rare autosomal recessive genetic disorder characterized by a classic tetrad of clinical features: disproportionate short stature, postaxial polydactyly of the hands, ectodermal dysplasia, and congenital heart defects [1, 2]. While its global incidence is low, it is found with significantly higher prevalence in founder populations, such as the Old Order Amish of Pennsylvania [3]. The syndrome is caused by homozygous or compound heterozygous mutations in either the EVC or EVC2 genes, which are located in a head-to-head configuration on chromosome 4p16 [4].

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It is essential to distinguish EVC from other disorders with overlapping features, such as Weyers acrofacial dysostosis, McKusick–Kaufman syndrome, and Jeune asphyxiating thoracic dystrophy, which share some skeletal or ectodermal anomalies but differ in their comprehensive clinical spectrum and genetic basis [5].

The clinical presentation of EVC is distinctive. Skeletal dysplasia manifests as acromelic and mesomelic shortening of the limbs, resulting in dwarfism, a long, narrow thorax, and genu valgum [6]. Polydactyly is a near-constant finding, typically presenting as a sixth digit on the ulnar side of the hands. Ectodermal signs are key to the diagnosis and include hypoplastic nails and sparse hair, as well as characteristic oral anomalies such as fusion of the upper lip to the maxillary gingiva, a V-shaped notch in the upper alveolar ridge, and hypoplastic or missing teeth [7].

Congenital heart defects are present in 50–60% of affected individuals and represent the primary cause of mortality, particularly in infancy [8]. The most common cardiac malformation is a large atrial septal defect, which often results in a functional or anatomical common atrium. Other reported defects include ventricular septal defects and abnormalities of the atrioventricular valves [9, 8]. While intelligence and motor development are typically normal, the cardiorespiratory complications necessitate the need for early and accurate diagnosis. This report presents a classic case of EVC

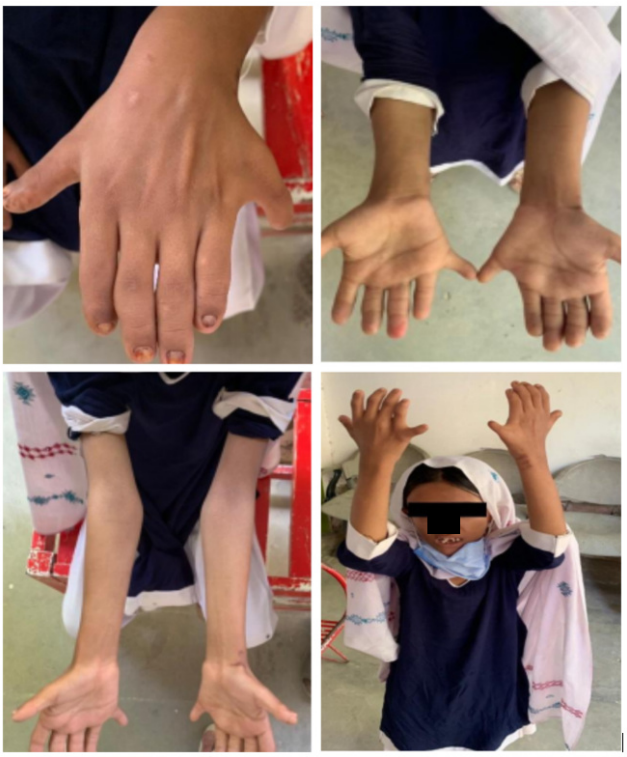

Figure 1: Several views of clinical photographs of the hands showing bilateral postaxial polydactyly, with a sixth digit present on the ulnar side.

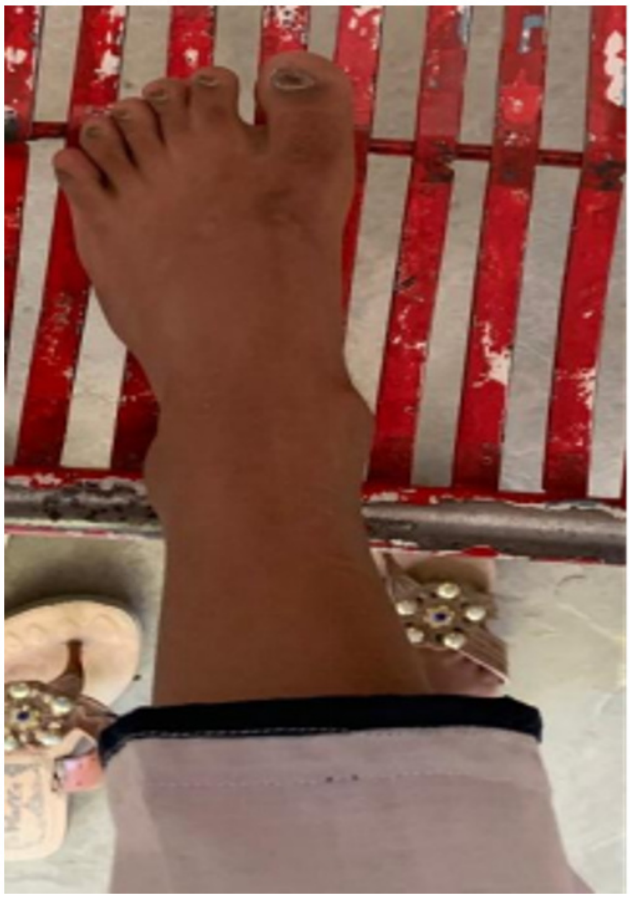

Figure 2: Characteristic postaxial polydactyly of the lower limbs in Ellis-van Creveld syndrome.

syndrome in an adolescent female, illustrating the constellation of skeletal, ectodermal, and cardiac findings that facilitate a clinical diagnosis, especially in settings where genetic testing is not readily available. Early and accurate diagnosis is crucial for initiating timely multidisciplinary management, including cardiology, orthopedic, and dental interventions, to mitigate complications and improve long-term prognosis.

2. Case Presentation

A 12-year-old female from a rural community in Pakistan was referred to the cardiology department for evaluation of a suspected congenital heart defect. She was born at full term via a normal vaginal delivery, and there was no history of natal or neonatal teeth. Her parents were non-consanguineous, and her five siblings (three younger brothers, two elder sisters) were reportedly healthy with no known congenital abnormalities. Genetic testing for EVC/EVC2 mutations was discussed with the parents; however, it was unavailable in our local setting due to resource limitations.

On general physical examination, the patient presented with disproportionate short-limb dwarfism, with a height of 118 cm (<3rd percentile for age), and a weight of 26 kg (BMI 18.8 kg/m²). Musculoskeletal examination revealed several characteristic findings. Bilateral postaxial polydactyly (hexadactyly) was evident in both the upper limbs, with a sixth digit on the ulnar side of each hand **(Figure 1)**, and in the lower limbs, with a sixth digit on the lateral aspect of each foot **(Figure 2)**. Further deformities included marked genu valgum (knock-knees) of the lower limbs, with an intermalleolar distance of 10 cm **(Figure 3)** and cubitus valgus of the forearms.

Ectodermal dysplasia was prominent. Oral examination revealed significant dental anomalies, including hypodontia (missing teeth), microdontia, and dysplastic, conical-shaped incisors **(Figure 4)**. Her fingernails were hypoplastic and dystrophic, and her scalp hair was noted to be thin and sparse. The head and trunk were of normal proportions for her age. Her psychomotor and cognitive development were within normal limits, and a neurological examination was unremarkable.

Cardiovascular evaluation confirmed a significant structural defect. A transthoracic echocardiogram revealed a common atrium, resulting from the complete absence of the interatrial septum **(Figure 5)**. The common atrium was unrestrictive, with significant left-to-right shunting. The atrioventricular valves were structurally normal, with no evidence of a cleft mitral valve or significant regurgitation. No evidence of pulmonary hypertension was noted, and no other defects, such as a ventricular septal defect or patent ductus arteriosus, were visualized. The left ventricular ejection fraction (LVEF) was preserved at 62%. An electrocardiogram (ECG) demonstrated right axis deviation and an incomplete right bundle branch block (RBBB) pattern, consistent with right ventricular volume overload **(Figure 6)**.

A posteroanterior chest X-ray showed cardiomegaly and a characteristically long, narrow thoracic cage with increased pulmonary vascularity, consistent with a left-to-right shunt. A precise cardiothoracic ratio was not formally measured. Abdominal and pelvic ultrasound examinations were normal, with no renal or hepatic

Figure 3: Marked genu valgum ('knock-knees'). Clinical photograph of the lower limbs showing the characteristic inward angulation of the knees, a common skeletal deformity in Ellis-van Creveld syndrome.

anomalies detected. Laboratory investigations, including a complete blood count (CBC), renal function tests (serum creatinine and blood urea), and liver function tests (LFTs), were all within normal ranges.

Based on the constellation of classic clinical and imaging findings, including chondroectodermal dysplasia (short-limb dwarfism, narrow thorax), postaxial polydactyly, characteristic oral and ectodermal features, and a congenital heart defect (common atrium), a clinical diagnosis of Ellis-van Creveld syndrome was made. The patient's clinical presentation and initial workup occurred over a period of two weeks. Following the diagnosis, the patient was referred for surgical evaluation for her common atrium and multidisciplinary consultations for orthopedic and dental care. She is currently awaiting cardiac surgery and long-term follow-up.

3. Discussion

This case report documents a classic presentation of Ellis-van Creveld syndrome in a 12-year-old female, whose constellation of findings aligns perfectly with the characteristic features described in the literature [1, 2]. The diagnosis was established clinically based on the presence of disproportionate short-limb dwarfism,

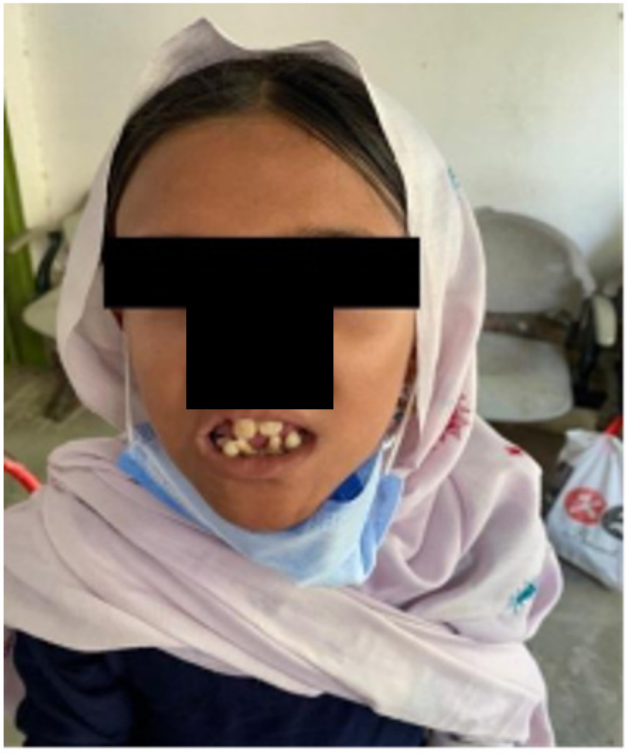

Figure 4: Oral manifestations of Ellis-van Creveld syndrome. The intraoral view shows characteristic dental anomalies, including significant hypodontia (multiple missing teeth), microdontia (abnormally small teeth), and dysplastic, conical-shaped incisors.

bilateral postaxial polydactyly, ectodermal dysplasia affecting the teeth and nails, and a signature congenital heart defect.

The skeletal dysplasia observed in our patient, including acromelic and mesomelic limb shortening, genu valgum, and a long, narrow thorax, is pathognomonic for EVC [6]. Postaxial polydactyly, a near-universal feature of the syndrome, was present in all four limbs of our patient. The absence of syndactyly in our patient is also consistent with typical presentations. These skeletal deformities, particularly genu valgum, often require orthopedic intervention to improve mobility and prevent long-term joint complications [6, 10].

Cardiovascular malformations are the most significant cause of morbidity and early mortality in EVC, affecting 50–60% of patients [8]. Our patient's finding of a common atrium, resulting from the complete absence of the interatrial septum, is the most frequently reported cardiac anomaly in this syndrome. [11, 12]. This defect leads to significant left-to-right shunting, causing right ventricular volume overload, which was clearly reflected in her electrocardiogram showing right axis deviation and an incomplete RBBB pattern [13, 8]. The preservation of her left ventricular function and absence of pulmonary hypertension are positive prognostic indicators, but lifelong cardiological follow-up is essential. The oro-dental and ectodermal findings are equally crucial for diagnosis. Our patient exhibited hypodontia, microdontia, and a partial fusion of the upper lip to the maxillary gingiva, which are hallmark features of EVC [7]. These oral signs are often present at birth. They can be a key clue for pediatricians and dentists to suspect the syndrome early in life, facilitating timely cardiac and genetic

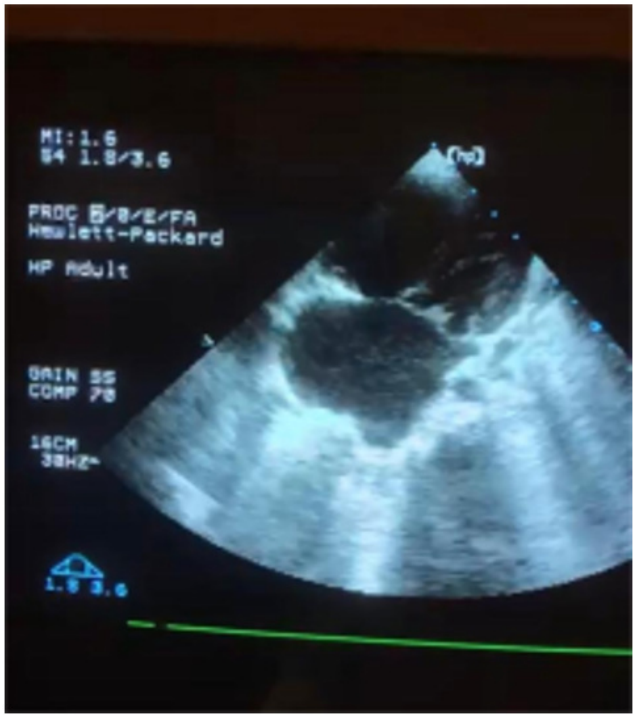

Figure 5: Echocardiographic evidence of a common atrium. A transthoracic echocardiogram (apical four-chamber view) demonstrates the complete absence of the interatrial septum, resulting in a single, functional atrial chamber. This is the most common congenital heart defect associated with Ellis-van Creveld syndrome.

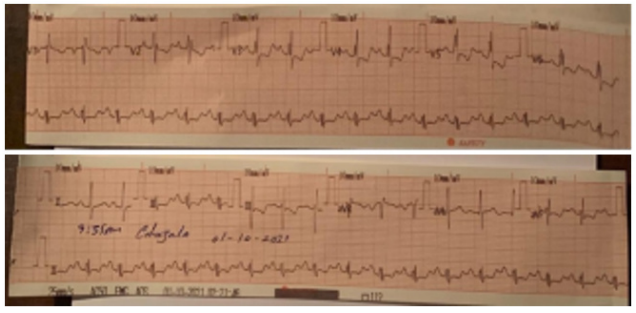

Figure 6: Electrocardiogram (ECG) demonstrating right ventricular volume overload. The 12-lead ECG shows a sinus rhythm with right axis deviation and an incomplete right bundle branch block (RBBB) pattern. These findings are characteristic of the hemodynamic effects of a large left-to-right shunt, consistent with the patient's common atrium.

evaluation [14]. The associated hypoplastic nails and sparse hair completed the picture of ectodermal involvement.

The definitive diagnosis of EVC is established through molecular genetic testing, which identifies mutations in the EVC or EVC2 genes on chromosome 4p16 [4]. Our patient's parents were non-consanguineous, which, for an autosomal recessive disorder, suggests heterozygosity in both parents for a pathogenic variant, potentially stemming from a shared distant ancestry within a localized community. It is also important to note that even with advanced sequencing, mutations are identified in only about two-thirds of clinically diagnosed patients, underscoring the continued importance of clinical assessment [15].

In resource-limited settings like ours, a diagnosis based on this classic constellation of symptoms remains a valid and necessary approach. This case reinforces the importance of recognizing the key clinical tetrad of EVC. The management of EVC is complex and requires a coordinated, multidisciplinary approach involving pediatricians, cardiologists, orthopedic surgeons, dentists, and genetic counselors [16]. For cardiac defects such as a common atrium, surgical repair is typically indicated to prevent progressive pulmonary hypertension and right ventricular dysfunction. The timing of intervention is crucial and often guided by the degree of shunting and the development of pulmonary vascular disease [13, 8]. Orthopedic management for genu valgum may involve bracing or corrective osteotomies to improve ambulation and prevent premature degenerative joint disease [6]. Staged dental care is crucial for addressing hypodontia, microdontia, and other oral anomalies, thereby improving mastication, speech, and aesthetics [14, 7]. The patient's preserved left ventricular function and mild pulmonary hypertension at presentation are positive indicators for a favorable outcome following appropriate surgical intervention, though lifelong cardiological follow-up remains essential.

4. Conclusions

This case report presents a classic clinical phenotype of Ellis-van Creveld syndrome, where the diagnosis was established through the recognition of its characteristic tetrad of features. In resource-limited settings, a clinical diagnosis of Ellis-van Creveld syndrome can be made from the characteristic phenotypic tetrad; molecular testing refines counseling and prognosis. The presence of hallmark findings, such as postaxial polydactyly and specific oro-dental anomalies, should prompt clinicians to conduct a comprehensive systemic evaluation, with special attention to cardiovascular screening. The successful long-term management of this complex genetic disorder hinges on early diagnosis and the coordinated efforts of a dedicated multidisciplinary team to address the patient's multifaceted health needs and improve their overall quality of life.

Conflicts of Interest

The authors declare that they have no competing interests.

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Informed consent

Consent for publication was obtained from the patient involved in this case report. All clinical photographs have been de-identified, and written informed consent for their publication was obtained from the patient's legal guardian.

Large Language Model

None

Authors Contribution

SMNA was the primary physician responsible for the patient's admission, management, and conceptualization of this case report. NG provided the crucial medical consultation and examination to exclude related syndromes. MFH and SMNA drafted the initial manuscript. SAHS, AS, and BK performed the comprehensive cardiological assessments, including all diagnostic tests, and were responsible for detailed data collection and obtaining informed consent from the patient's guardian. SAHS and BK provided critical revision for important intellectual content. All authors read and approved the final manuscript.

Data Availability

No datasets were generated or analyzed for this case report. All data supporting the findings of this study are included within the article. Additional clinical details are available from the corresponding author upon reasonable request, with appropriate safeguards to protect patient confidentiality.

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