International Journal of Contemporary Pediatrics
Arunkumar P et al. Int J Contemp Pediatr. 2026 Apr;13(4):663-665
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20260831

Case Report

Chondroectodermal dysplasia with recurrent lower respiratory tract infections: a case report

Preethi Arunkumar[1]*, C. R. Vignesh Kumaran[2]

[1]Madras Medical College, Chennai, Tamil Nadu, India
[2]Department of Paediatrics, Madras Medical College, Chennai, Tamil Nadu, India

Received: 30 January 2026
Accepted: 05 March 2026

***Correspondence:**
Dr. Preethi Arunkumar,
E-mail: preethi.mmc7@gmail.com

ABSTRACT

Chondroectodermal dysplasia (Ellis–van Creveld syndrome), a rare autosomal recessive skeletal dysplasia, is characterized by disproportionate dwarfism, ectodermal dysplasia, polydactyly, and congenital cardiac anomalies. Respiratory involvement is recognized as a contributor to morbidity and mortality, but is under-emphasised. We report a 4-year-old male child born to consanguineous parents presenting with multiple admissions for lower respiratory tract infections (LRTIs) since birth. The physical examination revealed short stature, along with acromesomelic limb shortening, frontal bossing, a depressed nasal bridge, low-set ears, postaxial polydactyly, and dental anomalies. The echocardiogram showed an ostium secundum atrial septal defect. The radiology results showed a restricted thoracic cavity together with shortened rib bones. Through consistent supportive care, the child's health improved significantly. Currently, he continues to receive treatment from multiple healthcare specialists. This case highlights that recurrent LRTIs are a significant yet under-recognised clinical complication of chondroectodermal dysplasia, underscoring the importance of early respiratory surveillance and multidisciplinary care to improve outcomes.

Keywords: Ellis–van Creveld syndrome, Chondroectodermal dysplasia, Lower respiratory tract infection, Skeletal dysplasia, Case report

INTRODUCTION

Chondroectodermal dysplasia (CED), also known as Ellis–van Creveld syndrome, is a rare autosomal recessive disorder first described by Ellis and van Creveld in 1940.[1] It is characterised by a tetrad of chondrodysplasia, ectodermal dysplasia, postaxial polydactyly, and congenital cardiac defects, with an incidence of 1 in 60,000–200,000 live births.[2] Higher prevalence is noted among the Amish population due to genetic isolation.[3]

CED results from mutations in the EVC1 and EVC2 genes on chromosome 4p16, which are important parts of the Hedgehog signalling pathway responsible for skeletal and ectodermal morphogenesis.[4] Clinical features include short-limbed dwarfism, characteristic facial features consisting of frontal bossing, depressed nasal bridge, low-set ears, postaxial polydactyly, and dental anomalies.[2]

Cardiac defects, especially ostium primum or secundum atrial septal defect, are found in about half of all cases and have the most significant impact on prognosis.[2] Respiratory complications, such as recurrent lower respiratory tract infections (LRTIs), also play an important role in morbidity and mortality resulting from thoracic restriction and compromised pulmonary function.[5]

Here, we report a case of classical CED presenting with recurrent LRTIs since birth and highlight the importance of respiratory manifestations in disease prognosis.

CASE REPORT

A 4-year-old male child born to third-degree consanguineous parents presented with a history of recurrent cough, difficulty in breathing, and wheezing, requiring multiple hospital admissions for lower

respiratory tract infections since infancy. The child was delivered at term via vaginal delivery, with a birth weight of 2.6 kg. Antenatal and perinatal periods were uneventful, and developmental milestones were age-appropriate. No similar illness was present in siblings.

The child was alert and playful, with short stature less than the 3rd percentile, and had disproportionate limb shortening with an acromesomelic pattern. The child had frontal bossing, a depressed nasal bridge, and low-set ears. The rest of the general examination was significant for bilateral polyaxial polydactyly, bilateral genu valgum, and flat foot (Figure 1). Nails were thin and brittle. Intraoral examination showed hypodontia; teeth appeared conical in shape, and there was malformation in the alveolar ridges (Figure 2).

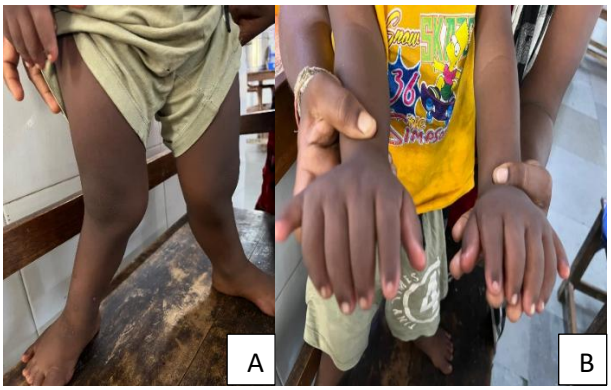

Figure 1 (A and B): Frontal view of the patient showing bilateral polyaxial polydactyly, bilateral genu valgum, and flat foot.

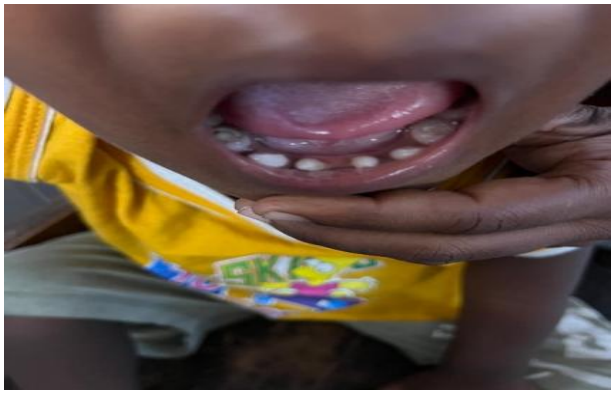

Figure 2: Oral view showing hypodontia, conical teeth and alveolar ridge malformation.

A grade II systolic murmur was heard at the left upper sternal border. Respiratory examination revealed bilateral crepitus and an occasional wheeze; a narrow, bell-shaped chest with reduced expansion.

Chest X-ray revealed a narrow thoracic cage, short ribs, and patchy bronchitic changes. 2D echocardiography showed ostium secundum atrial septal defect with L-R shunt, mild right atrium enlargement, and normal bi-ventricular function. The skeletal survey revealed limb shortening, flared metaphyses, and shortened ribs. All blood investigations were within normal limits.

During the acute phase of the illness, the child was treated with bronchodilators, antibiotics, and chest physiotherapy. A multidisciplinary strategy was employed for long-term treatment, which included pulmonary follow-up, influenza and pneumococcal vaccinations, dental and orthopaedic examinations, and family genetic counselling.

DISCUSSION

Chondroectodermal dysplasia is a congenital multisystem disorder characterised by skeletal, ectodermal, cardiac, and respiratory involvement.[2] Defect in endochondral ossification results in disproportionate short stature and limb shortening. Characteristic facial features, typical digit features, and dental anomalies help in establishing a clinical diagnosis.

Whereas cardiac anomalies, including mainly ostium secundum ASD, are significant causes of early morbidity, respiratory complications are equally essential but usually under-recognised.[5] Pulmonary expansions is seriously limited by the narrow thoracic cage and short ribs, predisposing to recurrent LRTIs and chronic hypoventilation. These infections further compromise the respiratory reserve, perpetuating a self-reinforcing cycle of respiratory morbidity.

Our patient presented with recurrent LRTIs since birth, showing significant thoracic restriction and predisposition to airway infection. Literature reports confirm that respiratory compromise, along with cardiac anomalies, is a leading cause of mortality in CED.[5]

The management of the condition should involve a multidisciplinary team including specialists such as paediatricians, pulmonologists, cardiologists, orthopaedists, and dentists. Emphasis on early detection of respiratory problems, early infection management, airway clearance therapy, vaccinations, and nutritional support is non-negotiable. Genetic counselling for these families is necessary on account of the condition's autosomal recessive mode of inheritance.[3]

This case highlights that recurrent respiratory infections are an integral complication of CED and not just an incidental occurrence, necessitating systematic examination and prevention.

CONCLUSION

Chondroectodermal dysplasia (Ellis–van Creveld syndrome) is a rare autosomal recessive disorder characterized by distinctive skeletal dysplasia, ectodermal anomalies, postaxial polydactyly, and cardiovascular anomalies. Although cardiac malformations are well-recognized contributors to early morbidity, recurrent

LRTIs are a frequently overlooked yet critical determinant of prognosis that stems from thoracic cage restriction, short ribs, and impaired pulmonary mechanics. This results in a vicious cycle of hypoventilation, atelectasis, and further respiratory compromise, often leading to repeated hospitalizations and heightened mortality risk in affected children.

Early, proactive respiratory surveillance in CED patients with particular emphasis on routine imaging, pulmonary function assessments, and aggressive preventive measures such as vaccinations, chest physiotherapy, and nutritional optimization is highlighted by this case. Holistic management can be achieved by a multidisciplinary approach with comprehensive focus on pediatric pulmonology, cardiology, orthopedics, dentistry, and genetics. Timely intervention not only mitigates acute exacerbations but also enhances long-term survival and quality of life. Future research should focus on respiratory outcomes to refine management protocols for this understudied complication.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Singh OP, Kumar V, Kumar R. Chondroectodermal dysplasia (Ellis-van Creveld syndrome). Int J Contemp Pediatr 2021;8:1599-601.Baujat G, Le Merrer M. Ellis-van Creveld syndrome. Orphanet J Rare Dis. 2007;2:27.
2. McKusick VA, Egeland JA, Eldridge R, Krusen DE. Dwarfism in the Amish I. The Ellis-van Creveld syndrome. Bull Johns Hopkins Hosp. 1964;115:306-36.
3. Ruiz-Perez VL, Goodship JA. Ellis-van Creveld syndrome and Weyers acrodental dysostosis are caused by mutations in the EVC and EVC2 genes. Am J Hum Genet. 2003;72(2):388-94.
4. Ruiz-Perez VL, Ide SE, Strom TM. Mutations in a new gene in Ellis-van Creveld syndrome and Weyers acrodental dysostosis. Nat Genet. 2000;24(3):283-6.
5. 'Pérez-Andreu J, Pérez-Andreu M, Carrió P. Ellis-van Creveld syndrome in adulthood. Orphanet J Rare Dis. 2005;1(Suppl 1):P108.

Cite this article as: Arunkumar P, Kumaran CRV. Chondroectodermal dysplasia with recurrent lower respiratory tract infections: a case report. Int J Contemp Pediatr 2026;13:663-5.