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REVIEWER'S REPORT

Manuscript No.: **IJAR-53023** Date: 30-07-2025

Title: Syndrome d' Ellis Van Creveld : A propos d' un cas

Recommendation:
Accept as it is .....
Accept after minor revision.....
Accept after major revision .....
Do not accept (*Reasons below*) .....

Rating	Excel.	Good	Fair	Poor
Originality		✓		
Techn. Quality		✓		
Clarity	✓			
Significance		✓		

Reviewer Name: Dr Aamina

Reviewer's Comment for Publication.

Summary of Content:

The article presents a case study of a 13-year-old female patient diagnosed with Ellis-van Creveld syndrome (EvC). The report highlights the classical clinical features of EvC, including dwarfism, polydactyly, nail dysplasia, and dental agenesis, with the notable absence of congenital heart malformations. The text emphasizes the phenotypic variability of the syndrome and the significance of consanguinity as a risk factor. The multidisciplinary management involving orthopedic and dental care, along with genetic counseling, is well-documented.

Strengths:

- The abstract provides a concise yet comprehensive overview of the clinical presentation, diagnosis, and management approach.
- The inclusion of epidemiological data, particularly the reference to higher prevalence in the Amish population, contextualizes the rarity of the condition in non-Amish populations.
- The case description is clear and focuses on the clinical variability of EvC, contributing valuable data to existing literature.
- The discussion of consanguinity as a key risk factor adds relevance for populations with higher consanguinity rates.

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Clinical Relevance:

The case contributes to the understanding of EvC's phenotypic spectrum, particularly the absence of cardiac malformations, which are typically prevalent. The report underscores the importance of multidisciplinary care and genetic counseling in managing rare autosomal recessive disorders.

Scientific Quality:

The manuscript is factually accurate and uses appropriate terminology related to genetic disorders. The inclusion of epidemiological figures and clinical findings strengthens the report's scientific rigor.

Overall Evaluation:

This case report provides a meaningful addition to the literature on Ellis-van Creveld syndrome. The detailed clinical description, emphasis on phenotypic variability, and documentation of genetic risk factors enhance its academic and clinical value.