

Second Trimester Medical Termination For Severe Fetal Skeletal Dysplasia Following Prenatal Diagnosis: A Case Report

Dr. Simran Sharma

(Junior Resident, Dept. Of Obstetrics And Gynaecology)

Dr. Shriya Ladiwala

(Junior Resident, Dept. Of Obstetrics And Gynaecology)

Dr. Mahenaz Preeen

(Senior Resident, Dept. Of Obstetrics And Gynaecology)

Dept. Of Obstetrics And Gynaecology, MGMIHS, Kamothe, Navi Mumbai, India

Abstract

Skeletal dysplasias are a heterogeneous group of genetic disorders affecting bone and cartilage development. This case highlights prenatal diagnosis of a lethal skeletal dysplasia leading to medical termination of pregnancy in the second trimester. A 21-year-old primigravida at 19–20 weeks of gestation presented with ultrasound findings suggestive of severe skeletal abnormalities. Features included short long bones, polydactyly, narrow thorax, and congenital heart defect. Based on the poor prognosis and findings incompatible with postnatal life, termination was performed using mifepristone and misoprostol in accordance with the Medical Termination of Pregnancy (MTP) Act. Fetal findings were consistent with Ellis-van Creveld syndrome. Early prenatal diagnosis and genetic counseling are crucial for management and future pregnancy planning.

Keywords: *Skeletal dysplasia, Ellis-van Creveld syndrome, polydactyly, prenatal diagnosis, second trimester termination, MTP*

Date of Submission: 02-05-2026

Date of Acceptance: 12-05-2026

I. Introduction

Skeletal dysplasias comprise a large and heterogeneous group of over 450 disorders characterized by abnormalities in bone and cartilage growth, with significant phenotypic variability ranging from mild to lethal forms. They may be inherited in autosomal dominant or recessive patterns and often present with short stature, limb deformities, and thoracic abnormalities. Lethal forms typically result in pulmonary hypoplasia due to a critically narrowed thoracic cage, rendering postnatal survival impossible.

Ellis-van Creveld (EVC) syndrome is a rare autosomal recessive skeletal dysplasia caused by mutations in the EVC and EVC2 genes located on chromosome 4p16. The syndrome has an estimated incidence of 1 in 60,000 to 200,000 live births. It is clinically characterized by disproportionate short stature, postaxial polydactyly, narrow thorax, ectodermal dysplasia affecting nails, teeth, and hair, and congenital cardiac anomalies, most commonly atrioventricular septal defects. The condition is associated with consanguinity and carries a 25% recurrence risk in subsequent pregnancies.

Prenatal diagnosis of EVC syndrome is possible through targeted ultrasonography, which may reveal features such as short limbs, polydactyly, narrow chest, and cardiac defects. Early diagnosis facilitates appropriate counseling regarding prognosis, pregnancy continuation, and future reproductive planning. In the context of the Indian MTP Act, termination of pregnancy is legally permissible in cases of severe fetal anomalies likely to result in a substantially handicapped child or where the anomaly is incompatible with life.

II. Case Report

Mrs. Ritu Kawate, a 21-year-old female, married primigravida with a gestational age of 19–20 weeks by dates (biparietal diameter 20.2 weeks; biometric scan 19.4 weeks), presented to MGM Hospital, Kamothe on 30/01/2026 for medical termination of pregnancy in view of an ultrasound report suggestive of lethal skeletal dysplasia, likely achondroplasia.

The patient had conceived spontaneously, and the antenatal period had been uneventful until the time of presentation. There was no history of exposure to teratogenic drugs, radiation, or infections during pregnancy. The couple reported a history of consanguineous marriage. There was no known family history of congenital

anomalies or skeletal disorders. First-trimester screening had been within normal limits. Routine antenatal investigations including complete blood count, blood group, and serological tests were unremarkable.

III. Ultrasound Findings (28/02/2026)

All long bones were markedly shortened for gestational age, with bowing noted in several long bones. The metacarpals of the hands were short and broad, giving a trident hand appearance. Supernumerary digits of the hand and foot were observed, suggestive of polydactyly. The thoracic cavity appeared narrow and disproportionate relative to the abdominal circumference. A defect involving the atria and ventricles of the heart was noted, suggestive of atrioventricular septal defect (AVSD) with absence of the crux and small left ventricular size.

Clinical Suspicion: Lethal skeletal dysplasia — likely achondroplasia, short rib-polydactyly syndrome, or a ciliopathy with skeletal dysplasia; Ellis-van Creveld syndrome also considered.

After detailed counseling regarding the poor prognosis and potential complications associated with the condition, the couple opted for termination of pregnancy in view of the ultrasound findings suggestive of lethal dysplasia. Written informed consent was obtained.

Medical termination of pregnancy was performed as follows: Tablet Mifepristone 400 mg orally administered on 30/01/2026 at 8:00 PM. Tablet Misoprostol 200 mcg orally administered at 4:00 AM on 02/02/2026, followed by 200 mcg sublingually, and then 200 mcg per vaginal smear at 10:00 AM. The abortus was delivered in breech presentation.

Fetal examination was performed and the baby was shown to the relatives. Fetal examination confirmed features consistent with the suspected diagnosis. The fetus was donated to the anatomy department for further genetic evaluation, specifically genomic array. Two doses of Tablet Cabergoline 0.5 mg were administered for lactation suppression. Urine and stools were passed. The patient remained vitally stable and was subsequently discharged.

Genetic counseling was provided to the couple regarding the autosomal recessive inheritance pattern of the condition and the estimated recurrence risk of approximately 25% in future pregnancies. They were advised to undergo early targeted anomaly scanning and genetic evaluation in subsequent pregnancies.

IV. Figures



*Fig. 1: Postaxial polydactyly — supernumerary digit of the hand and foot noted in the abortus
Fig. 2: Abortus showing disproportionate short stature, postaxial polydactyly, narrow thorax, and features consistent with ectodermal dysplasia*

V. Discussion

Management of pregnancies complicated by lethal skeletal dysplasia primarily involves detailed counseling regarding the poor prognosis and high likelihood of perinatal mortality. Second-trimester medical termination of pregnancy is a well-established and safe option in such cases. In accordance with the Medical Termination of Pregnancy (MTP) Act, termination is permissible in cases of severe fetal anomalies incompatible with life. In this case, termination was carried out using a standard regimen of mifepristone followed by misoprostol, which is a safe and effective method for second-trimester termination.

The combination of short long bones, postaxial polydactyly, narrow thorax, and an atrioventricular septal defect on prenatal ultrasound is highly suggestive of Ellis-van Creveld syndrome, particularly in the setting of consanguinity. The finding of a trident hand appearance and supernumerary digits further supports this diagnosis. While EVC syndrome is not uniformly lethal, the cardiac defect identified in this case (AVSD with a small left ventricle) carries a high risk of perinatal death and significant morbidity if the pregnancy were continued.

Post-termination evaluation of the fetus plays a crucial role in confirming the diagnosis and guiding future management. Genetic counseling is essential, particularly in autosomal recessive conditions like EVC syndrome, where the recurrence risk is 25% in subsequent pregnancies. Couples should be advised regarding early targeted anomaly scans, first-trimester screening, and, where available, molecular genetic testing in future pregnancies. Genomic array, as planned for this fetus, may provide additional confirmation and guidance for prenatal diagnosis in future pregnancies.

This case highlights the importance of early antenatal registration, timely anomaly scanning at 18–20 weeks, and multidisciplinary management involving obstetricians, radiologists, and genetic counselors. Early diagnosis not only facilitates informed decision-making but also helps reduce maternal morbidity and psychological distress associated with late recognition of severe fetal anomalies.

VI. Diagnosis And Outcome

Diagnosis: Ellis-van Creveld Syndrome (provisional, pending genomic array confirmation)

Indication for MTP: Severe fetal anomaly incompatible with normal postnatal life

Gestational Age at Termination: 19–20 weeks

Method: Mifepristone followed by Misoprostol (MTP Act compliant)

Consent: Obtained

Fetal Findings:

- Postaxial polydactyly (hands and feet)
- Short long bones with bowing
- Narrow thorax
- Trident hand appearance
- AVSD with small left ventricle
- Normal placenta

VII. Conclusion

This case underscores the vital role of detailed second-trimester anomaly scanning and multidisciplinary counseling in the management of suspected lethal skeletal dysplasias. A combination of characteristic ultrasonographic features — including short limbs, polydactyly, narrow thorax, and cardiac defects — in the setting of consanguinity should raise strong suspicion for Ellis-van Creveld syndrome. Timely prenatal diagnosis allows families to make informed decisions, reduces the burden of ongoing a pregnancy with a poor prognosis, and enables appropriate planning for future pregnancies with proper genetic counseling and targeted screening.

References

- [1]. Bacino CA, Lee B. Ellis-Van Creveld Syndrome. In: Adam MP, Feldman J, Mirzaa GM, Et Al., Editors. Genereviews® [Internet]. Seattle (WA): University Of Washington, Seattle; 1993–2024. Updated 2023 Oct 26. Available From: <https://www.ncbi.nlm.nih.gov/books/NBK596643>
- [2]. Baujat G, Le Merrer M. Ellis-Van Creveld Syndrome. Orphanet J Rare Dis. 2007;2:27. Doi:10.1186/1750-1172-2-27. PMID: 17559660.
- [3]. NORD. Ellis-Van Creveld Syndrome. Last Edit: Aug 14, 2024. Available From: <https://rarediseases.org/rare-diseases/ellis-van-creveld-syndrome>
- [4]. Medlineplus Genetics. Ellis-Van Creveld Syndrome. U.S. National Library Of Medicine. Available From: <https://medlineplus.gov/genetics/condition/ellis-van-creveld-syndrome>
- [5]. Cleveland Clinic. Skeletal Dysplasia. Last Reviewed: 9/7/2023. Available From: <https://my.clevelandclinic.org/health/diseases/skeletal-dysplasia>