

EP05.131 Prenatal Diagnosis and Outcomes of Fetal Rib Anomalies: A Cohort Study

39,149 Singleton Pregnancies (2021–2024) | 514 Cases of Fetal Rib Developmental Anomalies

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01. Background & Methodology

Rib anomalies stem from Hox gene dysregulation, often acting as soft markers for 22q11.2 microdeletions or skeletal ciliopathies. This study fills the gap of large-scale data integrating 3D ultrasound, genetics, and perinatal outcomes.

Core Research Objectives

- 1. Classify anomalies and determine population incidence.
- 2. Correlate genetic findings with isolated/non-isolated defects.
- 3. Establish a risk-stratified prenatal management protocol.

Study Design Details

Retrospective cohort (IRB-approved) of 39,149 pregnancies (20–24⁺⁶ wks). Imaging via Voluson E10 3D ultrasound; genetic testing included karyotyping, CMA, and WES. Outcomes defined as live birth or termination (TOP).

02. Key Epidemiological Findings

1.31%

Total Incidence
(514/39,149)

80.9%

Isolated Anomalies
vs 19.1% Complex



Risk Profile: Isolated vs. Non-isolated

Genetic Abnormality: 18.5% (Iso) vs 23.7% (Non-Iso)

Adverse Outcome: 0% (Iso) vs 17.4% (Non-Iso)

03. Clinical Management & Conclusion

Step 1: Detection & Survey → 3D ultrasound + complete fetal anatomical scan to rule out other anomalies.

Step 2: Genetic Testing → CMA for isolated cases; CMA + WES (trio) mandatory for non-isolated/complex defects.

Step 3: Counseling → Provide recurrence risk assessment and discuss prognosis based on genetic and structural findings.

Key Takeaways

Isolated rib anomalies carry low adverse outcome risk but are not entirely benign (18.5% genetic risk). Non-isolated cases demand urgent genetic workup due to high association with severe syndromes and poor outcomes.