

## Late Prenatal Diagnosis of Osteogenesis Imperfecta: A Case Report

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### INTRODUCTION/BACKGROUND

Osteogenesis imperfecta (OI) is a rare genetic disorder characterized by bone fragility and increased risk of fractures, most commonly resulting from mutations in the COL1A1 and COL1A2 genes, which disrupt type I collagen synthesis (1). Early detection is crucial for management and genetic counseling. Prenatal ultrasound is a key tool for identifying skeletal anomalies suggestive of OI (1,2).

### OBJECTIVES/AIMS

This case highlights the importance of prenatal ultrasound in identifying fetal anomalies suggestive of OI, particularly when the patient presents late in pregnancy and has no prior prenatal care.

### METHODS

This is a retrospective descriptive case report of a single patient seen at the Ben Arous Regional Hospital. Clinical history, ultrasound findings, radiographic evaluation, and neonatal outcomes were analyzed to assess the diagnostic approach and outcome.

### CONCLUSIONS

Osteogenesis imperfecta is often suspected based on prenatal ultrasound findings, though definitive diagnosis relies on genetic testing. This case highlights the importance of comprehensive prenatal screening, particularly when skeletal abnormalities are identified late in gestation. Early detection is key for effective counseling, clinical planning, and anticipating neonatal complications

### RESULTS

Mrs. HZ, a 26-year-old woman with no known family history of hereditary diseases, was Gravida II, Para II, and had previously delivered a healthy child. She sought prenatal care at 32 weeks of gestation, where an obstetric ultrasound revealed significant fetal anomalies, including micromelic dwarfism, a displaced fracture of the right femur, and an elongated skull shape, all suggestive of OI. Upon neonatal examination, a deformity of the right thigh, likely due to a pathological in-utero fracture, was observed. Radiographic imaging confirmed extensive ossification defects and limb deformities. The child survived for one year, experiencing multiple fractures but ultimately succumbed to respiratory distress at age one

Figure 1: ultrasound examination A: displaced fracture of the right femur, B: elongated skull

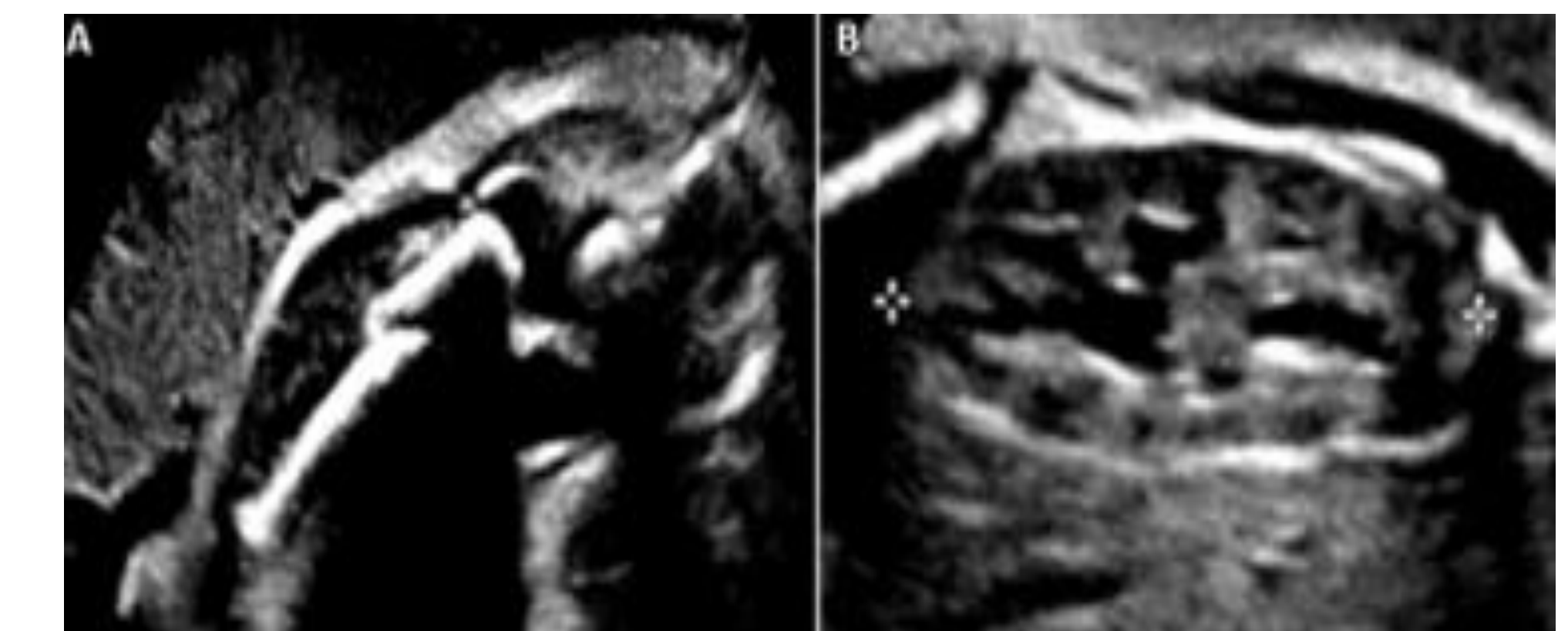


Figure 2: A: defect of ossification, short limbs, curved ribs; B: short limbs, displaced fractures



### REFERENCES

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