

## Clinical Image

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# Looking beyond retinopathy of prematurity

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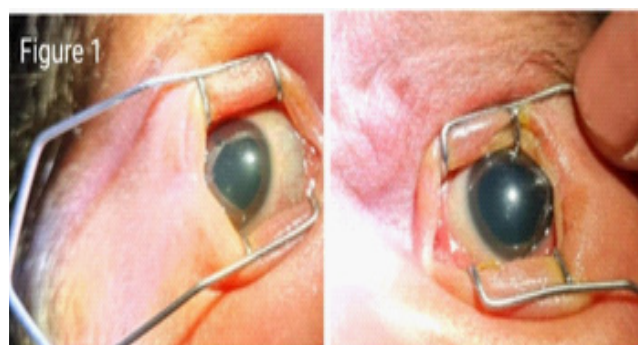
**Keywords:** Osteogenesis imperfecta; Retinopathy of prematurity.

### Case summary

A 2050 gm, 34 weeks gestational age term female with a family history of Osteogenesis Imperfecta (OI) was referred for ROP Screening. She was on oxygen support for respiratory distress. On examination, both eye sclera showed blue-grey discoloration (Figure 1). Fundoscopy of both eyes showed Stage 1, Zone II Retinopathy of Prematurity (ROP) (Figure 2). X-ray legs confirmed a fracture of the shaft of left femur with decreased mineralization of bone falling into type III Osteogenesis Imperfecta (OI) with anterior bowing of both tibia (Figure 3). The parents refused any genetic testing, as the elder male child had tested positive for OI and died 6 months after diagnosis.

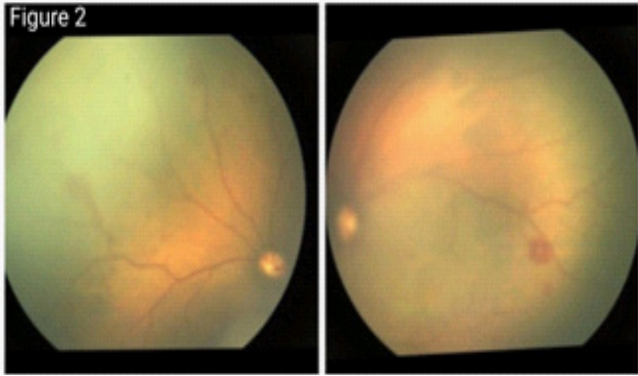
### Discussion

OI is a genetic connective tissue disorder due to abnormal synthesis or processing of type I collagen (90%); Type I-IV are autosomal dominant, Type VI-XIII are autosomal recessive [1]. Bone fragility in OI is not an isolated phenomenon but a part of the spectrum of abnormalities associated with basic collagen defect [2]. In the index case, an early diagnosis was clinched



**Figure 1:** Clinical picture at presentation showing blue- grey tinge of sclera in right and left eye.

due to a high index of suspicion of the condition as there was a family history of the disease in the elder sibling. The spectrum ranges from milder forms of bone susceptibility to the most severe forms like underdeveloped lungs incompatible with life. It is difficult to ascertain the cause of ROP in the present case as the baby had no high-risk factors like small gestational age and



**Figure 2:** RetCam fundus image with 130° lens of OD and OS showing stage 1 zone II ROP with dilated vessels multiple retinal hemorrhages, Roth spot.



**Figure 3:** X-Ray whole body showing anterior bowing of both tibia and fracture shaft of left femur and decreased mineralization of bone.

lower birth weight except oxygen support for respiratory distress. Patients with OI may also have an accompanying bleeding tendency secondary to capillary fragility caused by abnormal collagen support around blood vessels [3]. Multiple reports on bleeding diathesis in OI till date. We for the first time report ROP in osteogenesis imperfecta. Preterm babies with osteogenesis imperfecta may require an early eye examination for ROP screening.

**Conflict of interest:** No.

**Financial disclosure:** No.

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