Congenital osteogenesis imperfecta-microcephaly-cataracts syndrome

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Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital osteogenesis imperfecta-microcephaly-cataracts syndrome. ORPHA:2772

Congenital osteogenesis imperfecta-microcephaly-cataracts syndrome is characterised by multiple fractures in the prenatal period, microcephaly and bilateral cataracts. It has been described in three infants all of whom died in utero or a few hours after birth. The mode of inheritance appears to be autosomal recessive.