Osteogenesis imperfecta-retinopathy-seizures-intellectual disability syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Osteogenesis imperfecta-retinopathy-seizures-intellectual disability syndrome. ORPHA:2773

Osteogenesis imperfecta-retinopathy-seizures-intellectual disability syndrome is characterized by osteogenesis imperfecta, wormian bones, optic atrophy, retinopathy, seizures and severe developmental delay. It has been described in two sibs born to consanguineous parents.