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Night blindness-skeletal anomalies-dysmorphism syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Night blindness-skeletal anomalies-dysmorphism syndrome. ORPHA:1390*

This syndrome is characterized by night blindness, skeletal abnormalities (sloping shoulders, joint hyperextensibility, minor radiological anomalies) and characteristic facies (periorbital anomalies, malar flatness, retrognathia).