Oculotrichodysplasia

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Oculotrichodysplasia. ORPHA:2718

Oculotrichodysplasia is characterised by retinitis pigmentosa, trichodysplasia, dental anomalies, and onychodysplasia. It has been described in two siblings (brother and sister) born to first cousin parents. Transmission appears to be autosomal recessive.