Revesz syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Revesz syndrome. ORPHA:3088

Revesz syndrome is a rare severe phenotypic variant of dyskeratosis congenita (DC; see this term) with an onset in early childhood, characterized by features of DC (e.g. skin hyper/hypopigmentation, nail dystrophy, oral leukoplakia, high risk of bone marrow failure (BMF) and cancer, developmental delay sparse and fine hair) in conjunction with bilateral exudative retinopathy, and intracranial calcifications.