DISEASE:
Revesz syndrome

NAME:	Revesz syndrome
DESCRIPTION:	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.
ORPHACODE:	3088
SYNONYMS:	Dyskeratosis congenita with bilateral exudative retinopathy Retinopathy-anemia-central nervous system anomalies syndrome Revesz-DeBuse syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	TINF2
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