DISEASE:
Autosomal dominant popliteal pterygium syndrome

NAME:	Autosomal dominant popliteal pterygium syndrome
DESCRIPTION:	A rare genetic, multiple congenital anomalies syndrome characterized by cleft lip, with or without cleft palate, pits in the lower lip, contractures of the lower extremities, abnormal external genitalia, syndactyly of fingers and/or toes, and a pyramidal skin fold over the hallux nail.
ORPHACODE:	1300
SYNONYMS:	Facio-genito-popliteal syndrome Popliteal web syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	IRF6
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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