DISEASE:
Autosomal recessive cutis laxa type 2B

NAME:	Autosomal recessive cutis laxa type 2B
DESCRIPTION:	A rare, hereditary, developmental defect with connective tissue involvement characterized by cutis laxa of variable severity, in utero growth restriction, congenital hip dislocation and joint hyperlaxity, wrinkling of the skin, in particular the dorsum of hands and feet, and progeroid facial features. Hypotonia, developmental delay, and intellectual disability are common. In addition, cataracts, corneal clouding, wormian bones, lipodystrophy and osteopenia have been reported.
ORPHACODE:	357064
SYNONYMS:	ARCL2, progeroid type ARCL2B Autosomal recessive cutis laxa type 2, progeroid type
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PYCR1
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