DISEASE:
X-linked dominant chondrodysplasia punctata

NAME:	X-linked dominant chondrodysplasia punctata
DESCRIPTION:	A rare genodermatosis disease with great phenotypic variation and characterized most commonly by ichthyosis following the lines of Blaschko, chondrodysplasia punctata (CDP), asymmetric shortening of the limbs, cataracts and short stature.
ORPHACODE:	35173
SYNONYMS:	CDPX2 CDPXD CPXD Chondrodystrophia calcificans congenita Conradi-Hünermann-Happle syndrome X-linked chondrodysplasia punctata type 2
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	EBP
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