DISEASE:
Brittle cornea syndrome

NAME:	Brittle cornea syndrome
DESCRIPTION:	A rare, hereditary connective tissue disease characterized by severe ocular manifestations due to extreme corneal thinning and fragility with rupture in the absence of significant trauma, often leading to irreversible blindness. Extraocular manifestations comprise deafness, developmental hip dysplasia, and joint hypermobility.
ORPHACODE:	90354
SYNONYMS:	Ehlers-Danlos syndrome type 6B
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	ZNF469 PRDM5
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