DISEASE:
Butterfly-shaped pigment dystrophy

NAME:	Butterfly-shaped pigment dystrophy
DESCRIPTION:	A rare patterned dystrophy of the retinal pigment epithelium characterized by abnormal accumulation of lipofuscin in a butterfly-shaped distribution at the retinal pigment epithelium level. Patients manifest with a slowly progressive loss of vision that often only becomes apparent in old age.
ORPHACODE:	99001
SYNONYMS:	Butterfly-shaped pattern dystrophy Butterfly-shaped pigmentary macular dystrophy
XREF(S):	Orphanet ICD-10 OMIM OMIM OMIM
ANALYTE(S):	PRPH2 OTX2 CTNNA1
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- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
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