DISEASE:
Syndromic microphthalmia type 5

NAME:	Syndromic microphthalmia type 5
DESCRIPTION:	Syndromic microphthalmia, type 5 is characterized by the association of a range of ocular anomalies (anophthalmia, microphthalmia and retinal abnormalities) with variable developmental delay and central nervous system malformations.
ORPHACODE:	178364
SYNONYMS:	MCOPS5 Syndromic microphthalmia/anophthalmia due to OTX2 mutation
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	OTX2
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RELATED CONTENT

Related Genetic Tests

- Microphthalmia, syndromic 5; Retinal dystrophy, early-onset, and pituitary dysfunction

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- orthodenticle homeobox 2

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