DISEASE:
Oculocutaneous albinism type 2

NAME:	Oculocutaneous albinism type 2
DESCRIPTION:	A form of oculocutaneous albinism characterized by variable hypopigmentation of the skin and hair, numerous characteristic ocular changes and misrouting of the optic nerves at the chiasm.
ORPHACODE:	79432
SYNONYMS:	OCA2
XREF(S):	Orphanet ICD-10 OMIM MeSH
ANALYTE(S):	OCA2 MC1R
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RELATED CONTENT

Related Genetic Tests

- Ocular albinism and oculocutaneous albinism type 1, 2, 3, 4, 6, 7, 8 (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- melanocortin 1 receptor
- OCA2 melanosomal transmembrane protein

Related Gene Panels

- Ocular and oculocutaneous albinism - UGent