

DISEASE:

Minimal pigment oculocutaneous albinism type 1

NAME:	Minimal pigment oculocutaneous albinism type 1
DESCRIPTION:	An extremely rare form of Oculocutaneous albinism type 1 with minimal pigment present, characterized by blond hair (white at birth), variable iris transillumination (blue irides at birth followed by minimal development of pigment during the first decade of life), visual acuity ranging from 20/80-20/200 and white skin, with or without skin nevi.
ORPHACODE:	352734
SYNONYMS:	MP OCA type 1 OCA1-MP
XREF(S):	Orphanet ICD-10
ANALYTE(S):	TYR
CREATED:	13 May 2019 - 01:02
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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

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- tyrosinase

Related Gene Panels

- Ocular and oculocutaneous albinism - UGent

Source URL: <http://gentest.healthdata.be/disease/1894>