

## Opis choroby \*

### Definicja

A severe form of oculocutaneous albinism type 1 (OCA1) characterized by complete absence of melanin and manifesting as white hair and skin, blue, fully translucent irises, nystagmus and misrouting of the optic nerves.

### Dane

<b>Klasyfikacja</b>	<b>Synonimy</b>
Podtyp kliniczny	OCA1A Albinizm oczno-skórny tyrozynazo-ujemny OCA1A Tyrosinase-negative oculocutaneous albinism

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
79431	203100	E70.3

**Kod ICD11**  
EC23.20

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### \*Źródło

orphanet