

Opis choroby *

Definicja

Trichomegaly-retina pigmentary degeneration-dwarfism syndrome, also known as Oliver-McFarlane syndrome, is an extremely rare genetic disorder characterized by hair abnormalities, severe chorioretinal atrophy, hypopituitarism, short stature, and intellectual disability.

Dane

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|-----------------------|---|
| Klasyfikacja | Synonimy |
| Zespół wad wrodzonych | Long eyelashes-intellectual disability syndrome |
| | Długie rzęsy - niepełnosprawność intelektualna |
| | Zespół Olivera i McFarlane'a |
| | Oliver-McFarlane syndrome |

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|------------------|-----------------|------------------|
| Kod ORPHA | Kod OMIM | Kod ICD10 |
| 3363 | 275400 | Q87.1 |

Kod ICD11
LD27.0Y

*Źródło

orphanet