Opis choroby *

Definicja

A rare chromosomal anomalie characterized by intellectual deficit, retinal and skin pigmentation disorders, seizures, and dysmorphic features, including flat occiput, epicanthal folds, downward slanting eyes, flat nasal bridge, upturned nostrils, short neck, and large low set ears.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Ring 14
Ring chromosome 14

Kod ORPHA
1440

Kod OMIM
616606

Kod ICD10
Q93.2

Kod ICD11
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*Źródło

orphanet