Opis choroby *

Definicja

A rare genetic disease characterized by global developmental delay with language and cognition deficiencies, behavioral problems, osteopenia, joint laxity, skin defects consisting of hyperkeratosis and sweat gland and melanocyte abnormalities with hypopigmented areas, and abnormal hair structure. Mild facial dysmorphism (prominent forehead, thick eyebrows, epicanthal folds, broad nasal bridge, long philtrum, and micrognathia), abnormalities of the teeth, and skeletal and cardiac anomalies have also been described.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
73223

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet