Opis choroby *

Definicja

A rare sterol biosynthesis disorder characterized by microcephaly, bilateral congenital cataract, mild developmental delay, growth delay with short stature, psoriasiform dermatitis of variable severity, and immune dysregulation. Behavioral disorder, joint contractures, and arthralgia have also been described.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
SMO deficiency
Niedobór sterol-C4-metylo oksydazy
Sterol-C4-methyl oxidase deficiency

Kod ORPHA
488168

Kod OMIM
616834

Kod ICD10
E88.8

Kod ICD11
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*Źródło

orphanet