Opis choroby *

Definicja

A rare genetic disease characterized by the presence of multiple café-au-lait macules and elevated rates of sister chromatid exchange demonstrated on cytogenetic testing. Pre- and postnatal growth deficiency with short stature, microcephaly, mild developmental delay, cardiomyopathy, and symptomatic gastro-esophageal reflux have also been described, while malar rash is typically absent.

Dane

Klasyfikacja
Choroba

Kod ORPHA
508512

Kod OMIM
618097

Kod ICD10
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Kod ICD11
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*Źródło

orphanet