

## 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