

Opis choroby *

Definicja

12q14 microdeletion syndrome is characterised by mild intellectual deficit, failure to thrive, short stature and osteopoikilosis. It has been described in four unrelated patients. The syndrome appears to be caused by a heterozygous deletion at chromosome region 12q14, which was detected in three of the four patients. The deleted region contains the *LEMD3* gene: mutations in this gene have already been implicated in osteopoikilosis.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Del(12)(q14)

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Delecja 12q14

Monosomia 12q14

Osteopoikilia - niski wzrost - niepełnosprawność intelektualna

Deletion 12q14

Monosomy 12q14

Osteopoikilosis-short stature-intellectual disability syndrome

Kod ORPHA

94063

Kod OMIM

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Kod ICD10

Q93.5

Kod ICD11

LD2F.1Y

*Źródło

orphanet