Opis choroby *

Definicja

A rare syndromic form of lissencephaly characterized by severe microcephaly, agyria, agenesis of the corpus callosum, cerebellar hypoplasia, facial dysmorphology and epiphyseal stippling of the metacarpal bones. The syndrome may be an allelic variant of Neu-Laxova syndrome and Lissencephaly type III with cystic dilations of the cerebellum and foetal akinesia sequence.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
86822

Kod OMIM
601160

Kod ICD10
Q04.3

Kod ICD11
LD20.1

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*Źródło

orphanet