

Opis choroby *

Definicja

Microcephalic primordial dwarfism due to ZNF335 deficiency is characterized by severe antenatal microencephaly, simplified gyration, agenesis of the corpus callosum, absence of basal ganglia (very rare), pontocerebellar atrophy and involvement of the white matter with secondary cerebral atrophy. Congenital cataract, choanal atresia, multiple arthrogyrosis and spastic tetraparesis can occur.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Microcephalic primordial dwarfism, Walsh type
Pierwotna karłowatość mikrocefaliczna, typ
Walsha

Kod ORPHA

329228

Kod OMIM

615095

Kod ICD10

Q87.1

Kod ICD11

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*Źródło

orphanet