

## 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