

## Opis choroby \*

### Definicja

Pelizaeus-Merzbacher disease (PMD) is an X-linked leukodystrophy characterized by developmental delay, nystagmus, hypotonia, spasticity, and variable intellectual deficit. It is classified into three sub-forms based on the age of onset and severity: connatal, transitional, and classic PMD (see these terms).

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Diffuse familial brain sclerosis

PMD

PMD

Pelizaeus-Merzbacher brain sclerosis

Sudanophilic leukodystrophy, Paelizeus-

Merzbacher type

#### Kod ORPHA

702

#### Kod OMIM

213900

#### Kod ICD10

E75.2

#### Kod ICD11

8A44.0

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#### \*Źródło

orphanet