

## Opis choroby \*

### Definicja

A rare non-syndromic central nervous system malformation characterized by complete or near-complete absence of the cerebellum with a normal sized posterior fossa, possibly accompanied by hypoplasia of the brainstem. The clinical picture is highly variable, but typically includes ataxia, dysarthria, tremor, dysmetria, dysdiadochokinesia, and oculomotor abnormalities, in addition to impaired mental, motor, and language development and intellectual disability.

### Dane

|                     |  |                  |
|---------------------|--|------------------|
| <b>Klasyfikacja</b> | <b>Synonimy</b>  |                  |
| Wada morfologiczna  | Near total absence of cerebellum<br>Subtotal absence of cerebellum |                  |
| <b>Kod ORPHA</b>    | <b>Kod OMIM</b>  | <b>Kod ICD10</b> |
| 1398                | -  | Q04.3            |
| <b>Kod ICD11</b>    |  |                  |
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### \*Źródło

orphanet