

## Opis choroby \*

### Definicja

A rare, severe form of mevalonate kinase deficiency (MKD) characterized by dysmorphic features, failure to thrive, psychomotor delay, ocular involvement, hypotonia, progressive ataxia, myopathy, and recurrent inflammatory episodes.

### Dane

<b>Klasyfikacja</b>	<b>Synonimy</b>
Podtyp kliniczny	Complete mevalonate kinase deficiency Całkowity Niedobór kinazy mewalonianowej MVA MVA

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
29	610377	E88.8

**Kod ICD11**  
5C52.10

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### \*Źródło

orphanet