

## Opis choroby \*

### Definicja

X-linked intellectual disability, Snyder type is a rare X-linked intellectual disability syndrome characterized by hypotonia, asthenic build with diminished muscle mass, severe generalized psychomotor delay, unsteady gait and moderate to severe intellectual disability, as well as a long, thin, asymmetrical face with prominent lower lip, long fingers and toes and nasal, dysarthric or absent speech. Bone abnormalities (e.g., osteoporosis, kyphoscoliosis, fractures, joint contractures) are also characteristic. Myoclonic, or myoclonic-like, seizures and renal abnormalities have been associated in some patients.

### Dane

<b>Klasyfikacja</b> Choroba	<b>Synonimy</b> Snyder-Robinson syndrome Zespół Snydera i Robinsona	
<b>Kod ORPHA</b> 3063	<b>Kod OMIM</b> 309583	<b>Kod ICD10</b> Q87.8
<b>Kod ICD11</b> LD90		

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### \*Źródło

orphanet