

## Opis choroby \*

### Definicja

A rare X-linked genomic disorder associated with interstitial chromosomal duplications at Xq28 encompassing the *MECP2* gene. It is characterized in males by infantile onset hypotonia, severe global developmental delay, intellectual disability, progressive spasticity, seizures, gastrointestinal symptoms and recurrent respiratory infections. In females, the phenotype is more variable.

### Dane

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	MECP2 duplication syndrome
	Dystalna duplikacja Xq
	Telomerowa duplikacja Xq
	X-linked intellectual disability syndrome, Lubs type

<b>Kod ORPHA</b>	<b>Kod OMIM</b>	<b>Kod ICD10</b>
1762	300260	Q99.8

### Kod ICD11

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### \*Źródło

orphanet