## Opis choroby *

Definicja

A rare chromosomal anomaly clinically characterized by mild to severe intellectual disability, severe developmental delay (psychomotor and speech development), hypotonia with tendency to later develop progressive hypertonia, and characteristic facial features. The main congenital anomalies associated include central nervous system (CNS) malformations such as hypoplasia/agenesis of the corpus callosum (80%), skeletal abnormalities such as scoliosis/kyphosis or dislocated hips (60%), and congenital heart defects (25%).

Dane

**Klasyfikacja**
Zespół wad wrodzonych

Synonimy
Invdupdel(8p)
Invdupdel(8p)
Zespół odwróconej duplikacji/delecji 8p
Inverted 8p duplication/deletion syndrome

**Kod ORPHA**
96092

**Kod OMIM**
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**Kod ICD10**
Q99.8

**Kod ICD11**
LD41.P

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*Źródło

orphanet