Opis choroby *

Definicja

8p23.1 deletion involves a partial deletion of the short arm of chromosome 8 characterized by low birth weight, postnatal growth deficiency, mild intellectual deficit, hyperactivity, craniofacial abnormalities, and congenital heart defects.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(8)(p23.1)
Del(8)(p23.1)
Monosomia 8p23.1
Monosomy 8p23.1

Kod ORPHA
251071

Kod OMIM
-

Kod ICD10
Q93.5

Kod ICD11
LD44.81

---

*Źródło

orphanet