Opis choroby *

Definicja

A partial deletion of the short arm of chromosome 16 characterized by developmental delay, intellectual disability, speech delay, autism spectrum disorder, epilepsy, hypogonadism, and hypotonia. The behavioral profile includes impulsivity, compulsivity, stubbornness, manipulative behaviors, temper tantrums, and aggressive behaviors.

Dane

Klasyfikacja
Podtyp etiologiczny

Synonimy
Del(16)(p13.2)
Del(16)(p13.2)
Monosomia 16p13.2
Monosomy 16p13.2

Kod ORPHA
500055

Kod OMIM
616863

Kod ICD10
Q93.5

Kod ICD11
-

---

*Źródło

orphanet