

## **Opis choroby \***

## Definicja

16p13.3 microduplication syndrome is a rare chromosomal anomaly syndrome resulting from a partial duplication of the short arm of chromosome 16 and manifesting with a variable phenotype which is mostly characterized by: mild to moderate intellectual deficit and developmental delay (particularly speech), normal growth, short, proximally implanted thumbs and other hand and feet malformations (such as camptodactyly, syndactyly, club feet), mild arthrogryposis and characteristic facies (upslanting, narrow palpebral fissures, hypertelorism, mid face hypoplasia, bulbous nasal tip and low set ears). Other reported manifestations include cryptorchidism, inguinal hernia and behavioral problems.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Distal duplication 16p
	Dup(16)(p13.3)
	Duplikacja dystalna 16p
	Duplikacja telomerowa 16p
	Trisomia 16pter
	Trisomia dystalna 16p
	Distal trisomy 16p
	Dup(16)(p13.3)
	Telomeric duplication 16p
	Trisomy 16pter

Kod ORPHA  
96078

Kod OMIM  
613458

## Kod ICD10

Ked ICD11

## \* Źródło

orphanet