Opis choroby *

Definicja

A rare genetic disorder characterized by the association of total or partial aniridia, genitourinary anomalies (ranging from sexual ambiguity to ectopia testis), variable degrees of intellectual disability, and an increased risk of developing Wilms tumor. Glaucoma or cataract are also possible, and a minority of patients develop kidney failure. Other varaible findings may include obesity and duplicated halluces.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Del(11)(p13)
Del(11)(p13)
Delecja 11p13
Monosomia 11p13
Zespół guza Wilmsa, aniridii, wad moczowo-płciowych i niepełnosprawności intelektualnej
Deletion 11p13
Monosomy 11p13
Wilms tumor-aniridia-genitourinary anomalies-intellectual disability syndrome

Kod ORPHA
893

Kod OMIM
612469

Kod ICD10
C64

Kod ICD11
LD2A.Y

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*Źródło

orphanet