

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 variable 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 moczowopłciowych 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

*Źródło

orphanet