Opis choroby *

Definicja

A rare disorder of pentose phosphate metabolism characterized by developmental delay and intellectual disability, delayed or absent speech, short stature, and congenital heart defects (such as ventricular septal defect, atrial septal defect, and patent foramen ovale). Additional reported features include hypotonia, hyperactivity, stereotypic behavior, ophthalmologic abnormalities (bilateral cataract, uveitis, strabismus), hearing impairment, and variable facial dysmorphism, among others. Laboratory analysis shows elevated plasma and urinary polyols (erythritol, arabitol, and ribitol) and urinary sugar-phosphates (ribose-5-phosphate and xylulose/ribulose-5-phosphate).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Short stature-developmental delay-congenital heart defect syndrome
TKT deficiency
Short stature-developmental delay-congenital heart defect syndrome
TKT deficiency

Kod ORPHA
488618

Kod OMIM
617044

Kod ICD10
E88.8

Kod ICD11
5C51.0

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*Źródło

orphanet