

Niedobór transketolazy

Kod Orpha: 488618 Kod OMIM: 617044

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

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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