

Opis choroby *

Definicja

3-Phosphoglycerate dehydrogenase deficiency (3-PGDH deficiency) is an autosomal recessive form of serine deficiency syndrome (see this term) characterized clinically in the few reported cases by congenital microcephaly, psychomotor retardation and intractable seizures in the infantile form and by absence seizures, moderate developmental delay and behavioral disorders in the juvenile form

Dane

Klasyfikacja

Podtyp etiologiczny

Synonimy

PHGDH deficiency, infantile/juvenile form

PHGDH deficiency, infantile/juvenile form

Kod ORPHA

79351

Kod OMIM

601815

Kod ICD10

E72.8

Kod ICD11

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*Źródło

orphanet