

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	Synonimy
Podtyp etiologiczny	PHGDH deficiency, infantile/juvenile form PHGDH deficiency, infantile/juvenile form

Kod ORPHA	Kod OMIM	Kod ICD10
79351	601815	E72.8

Kod ICD11

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*Źródło

orphanet