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