

Opis choroby *

Definicja

Serine-deficiency syndrome is a very rare infantile-onset potentially treatable neurometabolic disorder characterized clinically by microcephaly, neurodevelopmental disorders and seizures. Three serine-deficiency syndromes have been described: 3-phosphoglycerate dehydrogenase (3-PGDH) deficiency, 3-phosphoserine phosphatase (3-PSP) deficiency, and phosphoserine aminotransferase deficiency (see these terms).

Dane

Klasyfikacja

Kategoria

Synonimy

Serine deficiency

Niedobór seryny

Kod ORPHA

35705

Kod OMIM

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Kod ICD10

E72.8

Kod ICD11

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*Źródło

orphanet