

Opis choroby *

Definicja

3-Phosphoserine phosphatase deficiency is an extremely rare form of serine deficiency syndrome (see this term) characterized clinically by congenital microcephaly and severe psychomotor retardation in the single reported case to date, which was associated with Williams syndrome (see this term).

Dane

Klasyfikacja

Podtyp etiologiczny

Synonimy

PSPH deficiency, infantile/juvenile form

PSPH deficiency, infantile/juvenile form

Kod ORPHA

79350

Kod OMIM

614023

Kod ICD10

E72.8

Kod ICD11

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*Źródło

orphanet