

Niedobór fosfatazy 3-fosfoseryny

Kod Orpha: 79350 Kod OMIM: 614023

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	Synonimy
Podtyp etiologiczny	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

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.