

# 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

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

### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.