

DISEASE:**3-phosphoserine phosphatase deficiency, infantile/juvenile form**

NAME:	3-phosphoserine phosphatase deficiency, infantile/juvenile form
DESCRIPTION:	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).
ORPHACODE:	79350
SYNONYMS:	PSPH deficiency, infantile/juvenile form
XREF(S):	<u>Orphanet</u> <u>OMIM</u> <u>ICD-10</u>
ANALYTE(S):	<u>PSPH</u>
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