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3-phosphoserine phosphatase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. 3-phosphoserine phosphatase deficiency. ORPHA:79350

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).