

Open Peer Review on Qeios

## Neurometabolic disorder due to serine deficiency

INSFRM

## Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

<u>Neurometabolic disorder due to serine deficiency</u>. ORPHA:35705

Serine-deficiency syndrome is a very rare infantile-onset potentially treatable neurometabolic disorder characterized clinically by microcephaly, neurodevelopmental disorders and seizures. Three serine-deficiency syndromes have been described: 3-phosphoglycerate dehydrogenase (3-PGDH) deficiency, 3-phosphoserine phosphatase (3-PSP) deficiency, and phosphoserine aminotransferase deficiency (see these terms).

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