



FULL TEXT LINKS



Case Reports Eur J Paediatr Neurol. 2016 Jan;20(1):53-60. doi: 10.1016/j.ejpn.2015.10.007.

Epub 2015 Nov 5.

## Two new cases of serine deficiency disorders treated with l-serine

A Brassier <sup>1</sup>, V Valayannopoulos <sup>1</sup>, N Bahi-Buisson <sup>2</sup>, Elsa Wiame <sup>3</sup>, L Hubert <sup>1</sup>, N Boddaert <sup>4</sup>,  
A Kaminska <sup>2</sup>, F Habarou <sup>5</sup>, I Desguerre <sup>2</sup>, E Van Schaftingen <sup>3</sup>, C Ottolenghi <sup>5</sup>, P de Lonlay <sup>6</sup>

Affiliations

PMID: 26610677 DOI: 10.1016/j.ejpn.2015.10.007

### Abstract

**Objective and patients:** We report on two new cases of serine deficiency due respectively to 3-phosphoglycerate dehydrogenase (PHGDH) deficiency (Patient 1) and phosphoserine aminotransferase (PSAT1) deficiency (Patient 2), presenting with congenital microcephaly (<3rd centile at birth) and encephalopathy with spasticity. Patient 1 had also intractable seizures. A treatment with oral l-serine was started at age 4.5 years and 3 months respectively.

**Results:** Serine levels were low in plasma and CSF relative to the reference population, for which we confirm recently redefined intervals based on a larger number of samples. l-Serine treatment led in patient 1 to a significant reduction of seizures after one week of treatment and decrease of electroencephalographic abnormalities within one year. In patient 2 treatment with l-serine led to an improvement of spasticity. However for both patients, l-serine failed to improve substantially head circumference (HC) and neurocognitive development. In a couple related to patient's 2 family, dosage of serine was performed on fetal cord blood when the fetus presented severe microcephaly, showing reduced serine levels at 30 weeks of pregnancy.

**Conclusions:** l-Serine treatment in patients with 2 different serine synthesis defects, led to a significant reduction of seizures and an improvement of spasticity, but failed to improve substantially neurocognitive impairment. Therefore, CSF and plasma serine levels should be measured in all cases of severe microcephaly at birth to screen for serine deficiency, as prompt treatment with l-serine may significantly impact the outcome of the disease. Reduced serine levels in fetal cord blood may also be diagnostic as early as 30 weeks of pregnancy.

**Keywords:** Congenital microcephaly; Intractable epilepsy; Oral l-serine treatment; Serine deficiency.

Copyright © 2015 European Paediatric Neurology Society. Published by Elsevier Ltd. All rights reserved.

[PubMed Disclaimer](#)

### Supplementary concepts

[Phosphoglycerate Dehydrogenase Deficiency](#)

[Phosphoserine Aminotransferase Deficiency](#)

### Related information

[Cited in Books](#)

[ClinVar](#)

[MedGen](#)

[MedGen \(Bookshelf cited\)](#)

[OMIM \(cited\)](#)

[PubChem Compound \(MeSH Keyword\)](#)

## LinkOut – more resources

### Full Text Sources

[ClinicalKey](#)

[Elsevier Science](#)

### Other Literature Sources

[The Lens - Patent Citations Database](#)

[scite Smart Citations](#)

### Medical

[MedlinePlus Health Information](#)

### Molecular Biology Databases

[The Weizmann Institute of Science GeneCards and MalaCards databases](#)

### Research Materials

[NCI CPTC Antibody Characterization Program](#)

### Miscellaneous

[NCI CPTAC Assay Portal](#)