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

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### 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.

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