

# Abstract

Curr Opin Neurol. 2004 Apr;17(2):197-204.

## Serine-deficiency syndromes.

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**PURPOSE OF REVIEW:** Serine-deficiency disorders comprise a new group of neurometabolic diseases and are caused by defects in the biosynthesis of the amino acid L-serine. In contrast to most neurometabolic disorders, serine-deficiency disorders are potentially treatable. Furthermore, the severe neurological symptoms observed in patients underscore the important roles of the serine biosynthetic pathway in brain tissue. An overview of the patients with serine-deficiency disorders reported to date, the biochemical findings and the results of treatment with amino acids is presented.

**RECENT FINDINGS:** L-Serine biosynthesis plays an important role in multiple cellular reactions, particularly in the brain, as L-serine is a precursor of important metabolites such as nucleotides, phospholipids and the neurotransmitters glycine and D-serine. Disturbances of serine-glycine metabolism in relation to N-methyl-D-aspartate-receptor activation are supposed to play a role in psychiatric disease as well. Recent findings concerning these roles of L-serine-derived phospholipids and neurotransmitters are presented.

**SUMMARY:** Congenital microcephaly, seizures and severe psychomotor retardation are symptoms of serine deficiency and can be treated with supplementation of L-serine, sometimes combined with glycine. The symptoms observed in serine deficiency confirm that L-serine and L-serine-derived metabolites play important roles in the central nervous system.

PMID: 15021249

