Sarcosinemia

Author: Professor Jaak Jaeken[1]
Creation Date: August 2001

Scientific Editor: Professor Jean-Marie Saudubray

[1]Director Centre for Metabolic Disease, University Hospital Gasthuisberg, Herestraat 49, B-3000 Leuven, Belgium. jaak.jaeken@uz.kuleuven.ac.be

Abstract
Sarcosinemia is an autosomal recessive condition characterized by an increased concentration of sarcosine in plasma and urine, due to sarcosine dehydrogenase deficiency. Sarcosinemia is most probably a benign condition without significant clinical problems. Mutations in the gene for sarcosine dehydrogenase, located on chromosome 9q34. Prevalence has been estimated to be 1:28,000 to 1:350,000 in newborn screening programs.

Keywords
sarcosinemia, sarcosine dehydrogenase

Disease name
Sarcosinemia

Excluded disease
Severe folate deficiency

Diagnosis criteria
Increase of sarcosine in plasma and urine; sarcosine dehydrogenase deficiency

Differential diagnosis
Severe folate deficiency

Prevalence
1/28,000 to 1/350,000 in newborn screening programs

Clinical description
A great variety of symptoms has been reported in sarcosinemia such as mental retardation, growth failure, hepatomegaly, craniosynostosis, syndactyly, and cardiomyopathy. On the other hand, a number of children with sarcosinemia detected by the neonatal metabolic screening program have remained completely symptom-free. Therefore it has been proposed that sarcosinemia is a benign condition and that the reported association with clinical symptoms is due to an ascertainment bias.

Management including treatment
On the assumption that this is a benign condition, treatment makes no sense.

Etiology
Mutations in the gene for sarcosine dehydrogenase, located on chromosome 9q34

Diagnostic methods
- amino acid analysis in plasma and/or urine
- enzymatic test

Genetic counseling
Autosomal recessive inheritance

Antenatal diagnosis
It is not relevant

References
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