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## Succinic semialdehyde dehydrogenase deficiency

**INSERM** 

## Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>Succinic</u> <u>semialdehyde dehydrogenase deficiency</u>. ORPHA:22

Succinic semialdehyde dehydrogenase (SSADH) deficiency is a rare neurometabolic disorder of gamma-aminobutyric acid (GABA) metabolism with a nonspecific clinical presentation (ranging from mild to severe) with the most frequent symptoms being cognitive impairment with prominent deficit in expressive language, hypotonia, ataxia, epilepsy, and behavioral dysregulation.

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