

Open Peer Review on Qeios

## **CADDS**

**INSERM** 

## Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. <u>CADDS</u>. ORPHA:369942

CADDS is a rare, genetic, neurometabolic disease characterized by severe intrauterine growth retardation, failure to thrive, profound neonatal hypotonia, severe global development delay, elevated very long chain fatty acids in plasma, and neonatal cholestasis leading to hepatic failure and death. Other features include ocular abnormalities (e.g. blindness and cataracts), sensorineural deafness, seizures, and abnormal brain morphology (notably delayed CNS myelination and ventriculomegaly).

Qeios ID: RX5GN2 · https://doi.org/10.32388/RX5GN2