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# Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.*

*Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency.*

*ORPHA:88618*

Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency is characterised by psychomotor delay and severe myopathy (hypotonia, absent tendon reflexes and delayed myelination) from birth, associated with hypermethioninaemia and elevated serum creatine kinase levels.