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# Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency. ORPHA:308410*

Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency is a rare disorder of branched-chain amino acid metabolism characterized by childhood-onset epilepsy, autism and intellectual disability with reduced levels of plasma branched chain aminoacids.