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# Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency. ORPHA:363432*

Autosomal recessive congenital cerebellar ataxia due to GRID2 deficiency is a rare, genetic, slowly progressive neurodegenerative disease resulting from GRID2 deficiency characterized by motor, speech and cognitive delay, hypotonia, truncal and appendicular ataxia, and eye movement abnormalities (tonic upgaze, nystagmus, oculomotor apraxia). Intention tremor may also be associated. Brain imaging reveals progressive cerebellar atrophy with cerebellar flocculus particularly affected.