Autosomal recessive cerebelloparenchymal disorder type 3

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive cerebelloparenchymal disorder type 3. ORPHA:1170

The disorders involving primarily the cerebellar parenchyma have been classified into six forms. In cerebelloparenchymal disorder III, cerebellar ataxia is congenital (non-progressive) and characterized by cerebellar symptoms such as incoordination of gait often associated with poor coordination of hands, speech and eye movements. The other features are congenital mental retardation and hypotonia, in addition to other neurological and non-neurological features. MRI or CT scan show marked atrophy of the vermis and hemispheres. A severe loss of granule cells with heterotopic Purkinje cells is observed. The mode of inheritance in the few reported families is autosomal recessive. In one family, cerebellar ataxia was associated to albinism.: In a large inbred Lebanese family the disease locus was assigned to a 12.1-cM interval on chromosome 9q34-qter between markers D9S67 and D9S312. The primary biochemical defect remains unknown. Up to now, the only treatment has consisted in early interventional therapies including intensive speech therapy and adequate stimulation and/or training.