CACH syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. CACH syndrome. ORPHA:135

A new leukoencephalopathy, the CACH syndrome (Childhood Ataxia with Central nervous system Hypomyelination) or VWM (Vanishing White Matter) was identified on clinical and MRI criteria. Classically, this disease is characterized by (1) an onset between 2 and 5 years of age, with a cerebello-spastic syndrome exacerbated by episodes of fever or head trauma leading to death after 5 to 10 years of disease evolution, (2) a diffuse involvement of the white matter on cerebral MRI with a CSF-like signal intensity (cavitation), (3) a recessive autosomal mode of inheritance, (4) neuropathologic findings consistent with a cavitating orthochromatic leukodystrophy with increased number of oligodendrocytes with sometimes ``foamy'' aspect.