Hypomyelinating leukodystrophy-ataxia-hypodontia-hypomyelination syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hypomyelinating leukodystrophy-ataxia-hypodontia-hypomyelination syndrome. ORPHA:137639

Hypomyelinating leukodystrophy-ataxia-hypodontia-hypomyelination syndrome is a rare, genetic, neurological disorder characterized by early-onset, progressive ataxia, white matter hypomyelination and cerebellar atrophy on brain MRI imaging, and various dental abnormalities, including hypodontia, delayed primary tooth eruption, complete retention of the primary maxillary central incisors and abnormal shape of the permanent maxillary incisors.