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Cerebral atrophy is linked to clinical severity and worsens with aging in patients with Pelizaeus-Merzbacher disease and Spastic Paraplegia type 2

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Objective: This study aims to determine the natural history of genetic hypomyelinating disorders Pelizaeus–Merzbacher disease (PMD) and Spastic Paraplegia type 2 (SPG2) on magnetic resonance imaging (MRI) and to correlate MRI results with clinical severity.

Methods: A prospective study in 35 patients with variable forms of PMD or SPG2 was performed on a total of 66 cerebral MRI. A myelination score and a global atrophy score were determined for each MRI. We also performed image segmentation to calculate corpus callosum area, cerebellum, white matter and gray matter volumes.

Results: Multivariate analysis with adjustment to the age revealed a significant difference for global atrophy score and corpus callosum area in different subgroups of severity at the first MRI acquisition. Evolution of MRI over time showed an increasing score of myelination until the age of 12 years, followed by stabilization. On the contrary, global atrophy, atrophy of corpus callosum and cerebellum worsened in the majority of patients with aging. No change in gray matter and white matter proportions was noticed over time.

Discussion: This study points out a correlation between cerebral atrophy and clinical severity in patients with PMD and SPG2 whereas degree of myelination was not clearly discriminating. It also highlighted, for the first time in a large cohort of patients, a clear inter- and intra-individual cerebral atrophy over time involving white and gray matter.