Open Peer Review on Qeios

Adult-Onset Leukoencephalopathy with Axonal Spheroids and Pigmented Glia

National Cancer Institute

Source

National Cancer Institute. <u>Adult-Onset Leukoencephalopathy with Axonal Spheroids and</u> <u>Pigmented Glia</u>. NCI Thesaurus. Code C153289.

A rapidly progressive neurodegenerative disorder, caused by mutations in the colonystimulating factor 1 receptor (CSF1R) gene, that presents in adulthood with a variety of neuropsychiatric and motor disturbances. Hallmark features include diffuse myelin loss and axonal destruction, neuroaxonal spheroids, and pigmented macrophages and other glia.