Adult Onset Autosomal Dominant Leukodystrophy

- Caused by a duplication of the lamin B1 gene (LMNB1)

- Rare and slowly progressive neurologic disease characterized by the onset of autonomic dysfunction, pyramidal signs, and cerebellar ataxia in the fourth or fifth decade.

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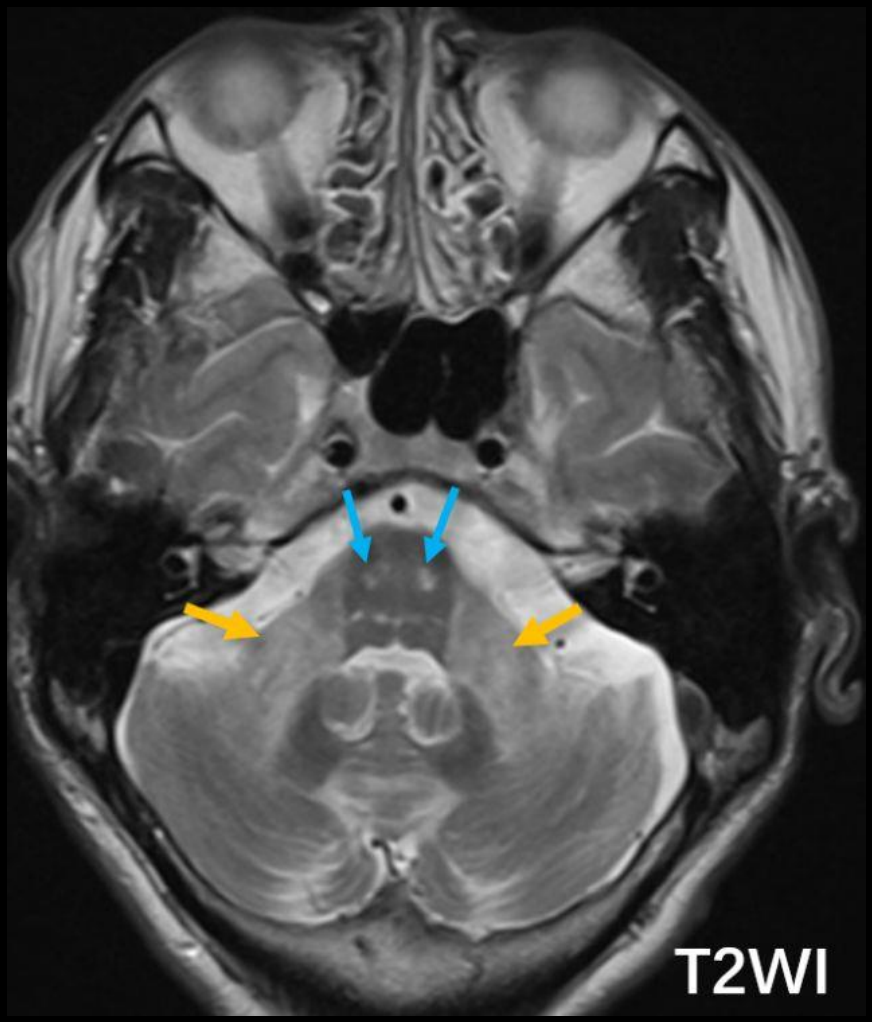
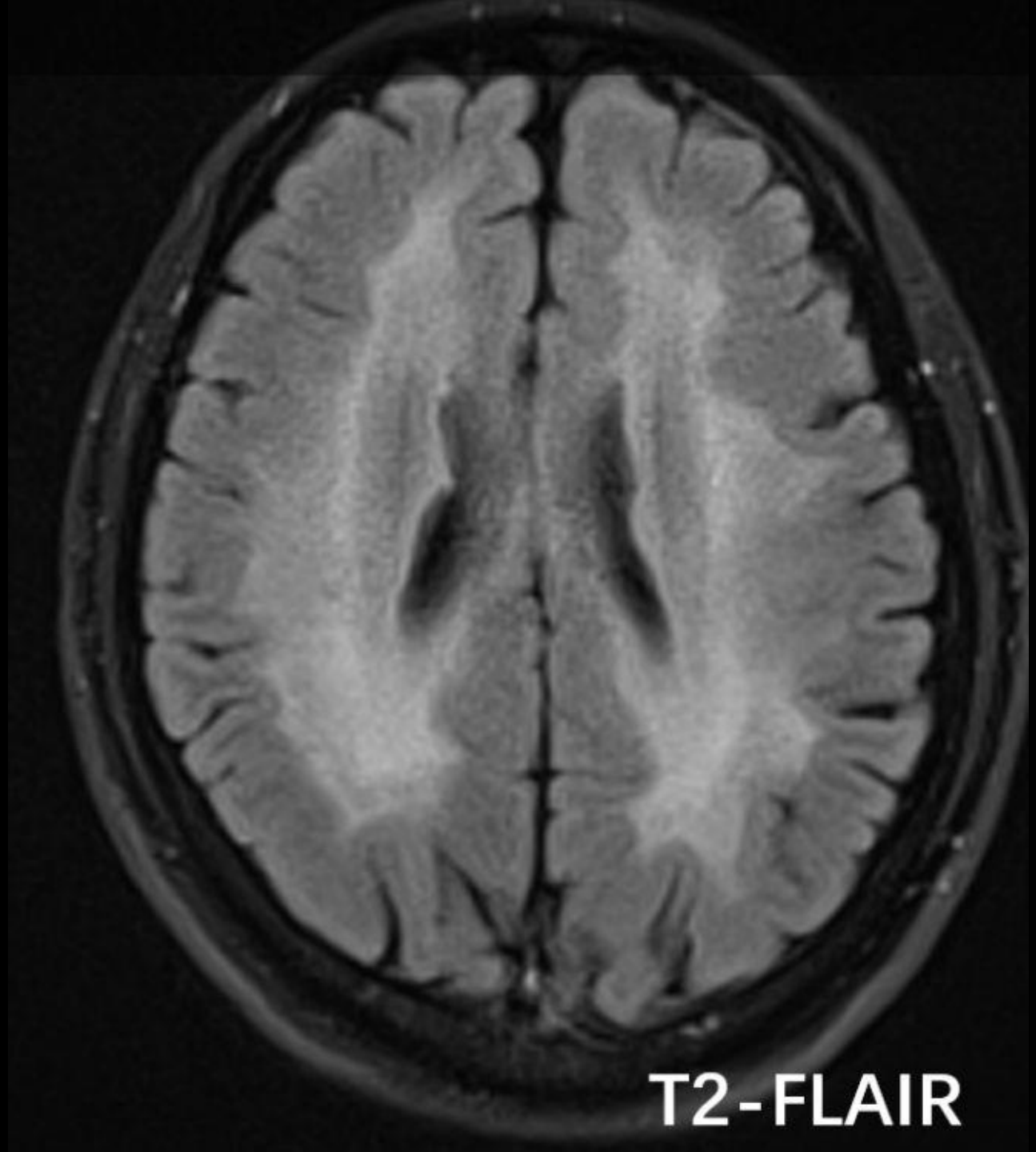
- MRI scans display diffuse, confluent and symmetric white matter involvement starting in the frontoparietal region, extending to the brainstem and cerebellar white matter
- Periventricular white matter appears less affected than the adjacent white matter
- The diameter of the medulla oblongata is reduced in the coronal plane and the corpus callosum is atrophic
- Although spinal cord involvement has been reported in some cases, white matter changes are most significant in the brain

The classic imaging findings are symmetric diffuse white matter hyperintensity on T2-FLAIR, mainly in the periventricular white matter of the frontal-parietal lobes at the level of the centrum semiovale (*A*), and bilateral middle cerebellar peduncles (*B, yellow arrows*).

Atrophy of the brainstem and spinal cord is also present, along with increased signal intensity of the corticospinal tracts (*B* and *C, blue arrows*), and pyramids (*C, yellow arrow*) on T2WI, which are other features of ADLD.




T2-FLAIR


T2WI

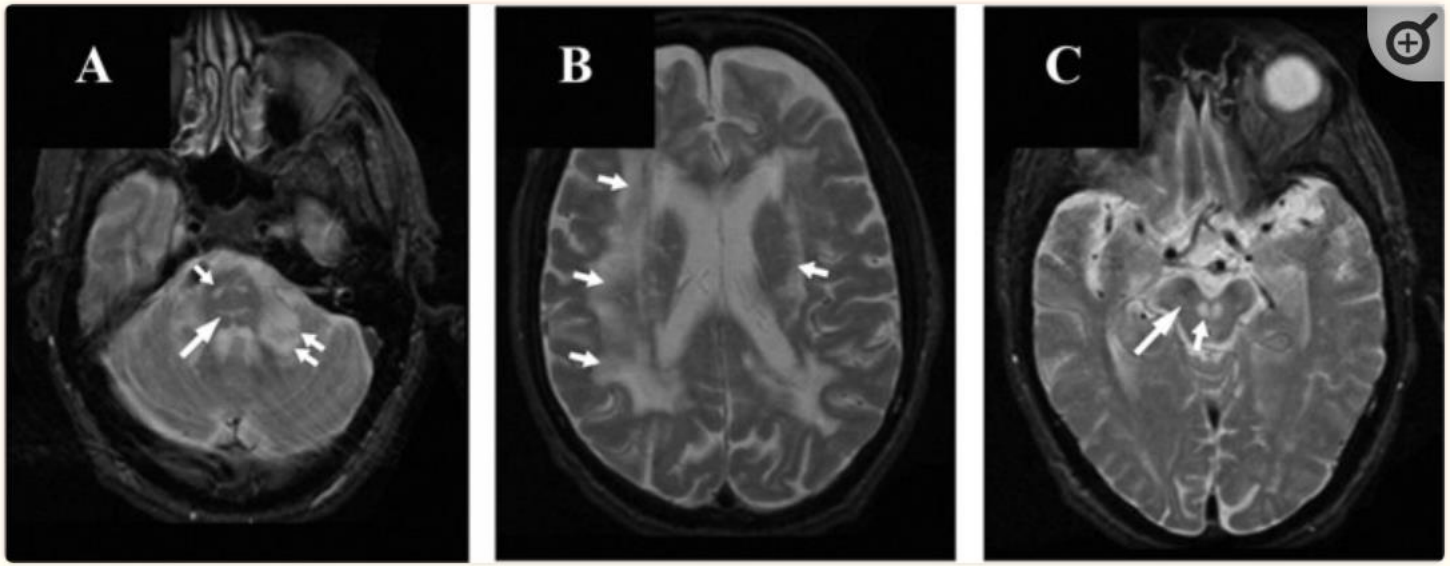


Figure 1.

T2 weighted MRI showing characteristic white matter hyperintensities identified in patients with Lamin B1 mutations. These MRI abnormalities appear to be restricted to specific white matter tracts and accumulate through the course of disease. ***A***, Transverse (axial) cross section through mid-pons at level of facial colliculus. Short arrow, Corticospinal and corticobulbar tracts (may include crossing pontine tracts); long arrow, medial lemniscus; double arrow, middle cerebellar peduncle (brachium pontis). ***B***, Transverse (axial) cross section through hemispheres at level of lateral ventricles. Three left side arrows, Superior longitudinal fasciculus; short arrow, cingulum. ***C***, Transverse (axial) cross section through caudal midbrain below level of red nucleus. Short arrow, Crossing fibers of superior cerebellar peduncle (brachium conjunctivum); long arrow, medial lemniscus.