Characteristic MRI pattern of LBSL

(A-D) Axial T2-weighted images through the brain of patient LBSL16 at the age of 16 years. (A) Inhomogeneous signal abnormalities are seen in the periventricular and deep cerebral white matter, (B) the splenium of the corpus callosum (indicated by s), and posterior limb of the internal capsule (pl). (C) At the level of the pons, the pyramidal tracts (pt), medial lemniscus (ml), mesencephalic trigeminal tracts (mtt), intraparenchymal parts of the trigeminal nerves (tn) and superior cerebellar peduncles (scp) are affected. (D) At the level of the medulla, the pyramids (p), decussation of the medial lemniscus (dml), inferior cerebellar peduncles (icp) and anterior spinocerebellar tracts (act) are involved. (E-G) T2-weighted images of the spinal cord. (E) The sagittal image shows signal abnormality over the entire length of the spinal cord (indicated by arrowheads). The axial images at the level of the cervical (F) and thoracic (G) spinal cord demonstrate that the signal abnormalities are confined to the dorsal columns (dc) and lateral corticospinal tracts (lct). (H) Proton MRS of white matter of patient LBSL16. The two abnormal peaks centered at 1.33 ppm represent lactate (Lac).