Metastatic breast cancer in a man with nonprogressive ataxia and epilepsy

Figure 1 Brain MRI findings in Bannayan-Riley-Ruvalcaba syndrome

(A) Sagittal T1-weighted, (B) axial T2-weighted, and (C) fluid-attenuated inversion recovery (FLAIR) sequences show marked global cerebellar atrophy. (D) Axial T1-weighted and (E) FLAIR sequence show left cerebral hemiatrophy and left frontal sinus hyperpneumatization.

Figure 2 Spine MRI findings in metastatic breast cancer

(A) Sagittal T1-weighted, (B) T2-weighted, (C) short tau inversion recovery sequence, and (D) postgadolinium-enhanced T1-weighted images show a heterogeneous invasive mass at T10-T11 level with severe spinal cord compression (white arrows).

A 44-year-old man presented with subacute-onset paraparesis and urinary incontinence. Medical history disclosed macrocephaly, neurodevelopmental delay, epilepsy, and nonprogressive ataxia. Examination disclosed a complete spinal cord syndrome without clear sensory level. Neuroimaging disclosed marked brain hemiatrophy (figure 1) and a compressive mass at T10-T11 level (figure 2). Lesion biopsy revealed a breast adenocarcinoma. Genetic analysis confirmed mutations in the *PTEN* gene, diagnosing Bannayan-Riley-Ruvalcaba syndrome (BRRS).

PTEN gene is a tumor suppressor gene involved in *PTEN* hamartoma tumor syndromes that include Cowden syndrome, BRRS, and Proteus-like syndrome with familial cancer predisposition and autism spectrum disorders or developmental delay with macrocephaly.^{1,2}

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