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Visual Diagnosis

Rapidly Progressing Brain Atrophy in a Child With Developmental Regression

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This 17-month-old boy was admitted due to delayed psychomotor development. His prenatal and perinatal histories were uncomplicated. His psychomotor development was normal until the end of his first year of life when a delay in speech development and motor deficits appeared. At admission, he was unable to turn onto his back or sit and he stood on his toes with assistance. Neurological examination revealed microcephaly, axial hypotonia, and hyperactive tendon reflexes in the legs. His family history was unremarkable.

His first brain magnetic resonance imaging at age 17 months was normal (Fig A,B), but subsequent scans at ages 21 and 29 months demonstrated rapidly progressive brain atrophy (Fig C-F, respectively). Further decline in motor and intellectual function was observed. He could not speak, raise his head from a lying position, or follow a light. He developed Babinski signs, muscle twitches (myoclonus), and bilateral ankle clonus.

Low activity of palmitoyl-protein thioesterase 1 (PPT1) and a mutation c.364A>T (p.Arg122Trp) in *PPT1* gene confirmed the diagnosis of ceroid lipofuscinosis type 1 (CLN1).

The occurrence of motor disorders, drug-resistant epilepsy, developmental regression, and worsening of eyesight should arouse the suspicion of CLN. These diseases represent one of the most common causes of progressive encephalopathy in children.[1,2] CLN2 is the most frequent type, with an incidence of 1:100 000 children.[1] Type CLN1 is less common, but it is characterized by a more progressive course with rapidly evolving brain atrophy and loss of acquired skills, leading to a vegetative state and death by about age four years.[3,4]

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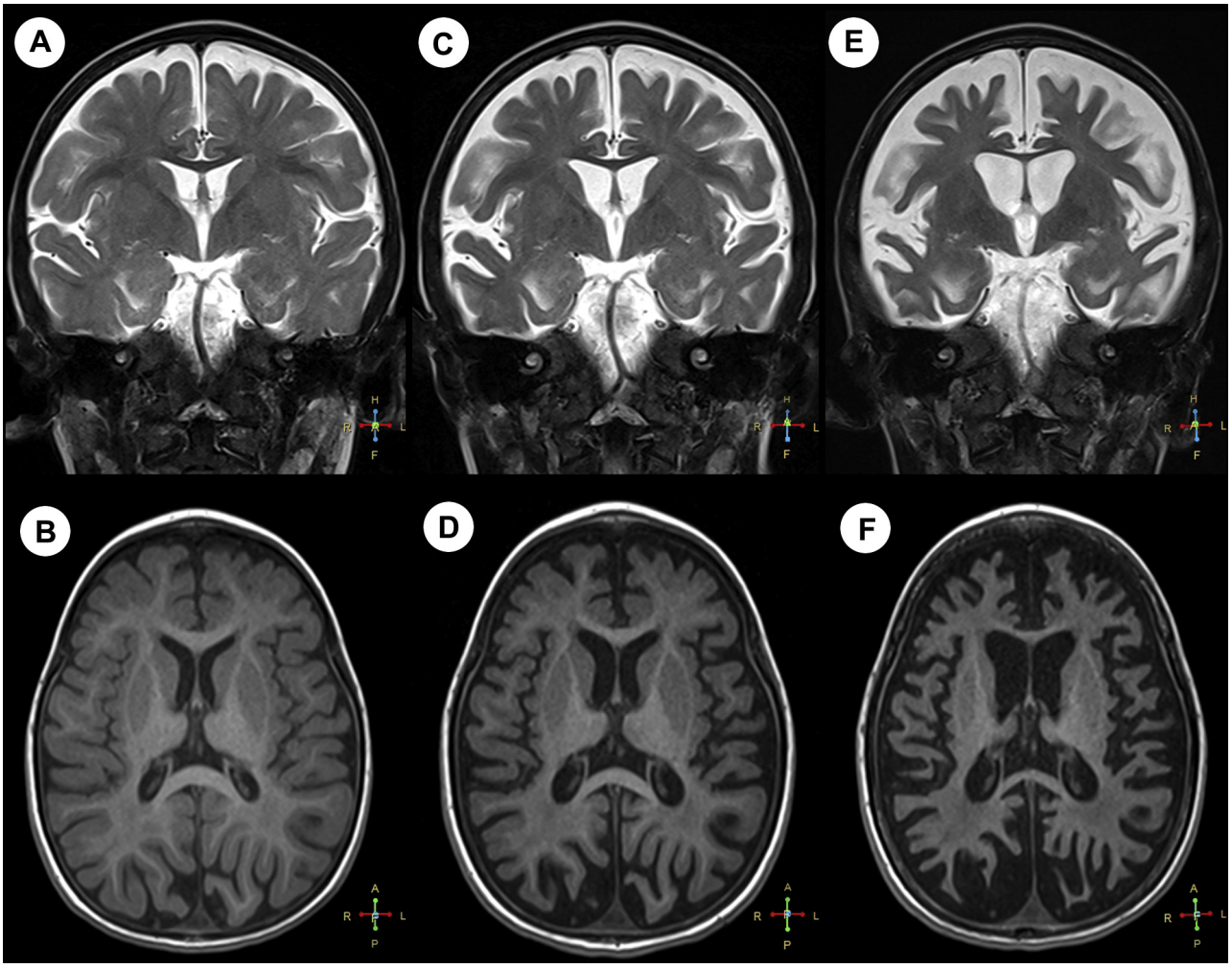

FIGURE. Sequential brain magnetic resonance imaging studies at ages 17 months (A and B), 21 months (C and D), and 29 months (E and F). Panels A, C, and E depict T2-weighted coronal images, and panels B, D, and F show T1-weighted axial images. The scans demonstrate rapid progression of brain atrophy from normal to severe generalized symmetric atrophy of both cortex and deep structures, with loss of white-gray matter differentiation and cerebellar atrophy.