Serial MRI in a child with PEHO syndrome

A 5-year-old girl was admitted to our hospital suffering from seizures and unable to walk. Neurological examination showed hypotonia, reduced spontaneous movements and lack of visual fixation. Her past medical history was also remarkable, as she had not started talking until she was 3 years old. At that age, she had non-febrile seizures and a walking disability. Initial electroencephalography (EEG) and metabolic screening tests were normal at this time. After 1 year, EEG showed bilateral occipital spike waves with abnormal background activity. Hypsarrhythmia developed during the following weeks. Antiepileptic drugs were started. At 4 years of age, bilateral optic-nerve atrophy was seen on eye examination. Serial MRI was carried out in March 2005, April 2006 and March 2007 (Figs. 1–3). Initial MRI showed moderate cerebral and cerebellar atrophy; the second MRI showed marked cerebral and cerebellar atrophy and the final MRI showed major progressive cerebral and cerebellar cortical atrophy.

Progressive encephalopathy with edema, hypsarrhythmia and optic-nerve atrophy (PEHO) syndrome is rare. Clinical features include hypotonia, convulsions with myoclonic jerking, infantile spasms and/or hypsarrhythmia, absence of motor milestones, poor or absent visual fixation, progressive brain atrophy, mental retardation, and edema of the face and limbs [5,6]. Additional features include subcutaneous peripheral and facial edema, microcephaly and facial dysmorphism [4].

Our case has the typical features of PEHO syndrome, with progressive cerebral and cerebellar cortical atrophy, convulsions, hypotonia, absent visual fixation and walking disability, but none of the additional features, such as subcutaneous peripheral and facial edema, microcephaly and dysmorphic features described by Riikonen. In PEHO syndrome, levels of insulin-like growth factor 1 were signifi-
significantly lower, and levels of nitric oxide were markedly elevated, compared with controls [4].

Disease onset is during early infancy and most children appear healthy at birth [2]. The hypsarrhythmic EEG pattern is usually evident by 1 year of age. Vision failure with abnormal eye movements usually develops early in infancy [1]. Our case had hypsarrhythmia and bilateral optical atrophy. Also, the rate of cerebellar atrophy in our case was less prominent than in earlier reported cases [2,3]. However, follow-up MR imaging is expected to reveal more cerebellar atrophy.

Our case has the typical features of PEHO syndrome, such as progressive cerebral and cerebellar cortical atrophy, convulsions, hypotonia, absent visual fixation and...
walking disability, which developed within a 2-year period, and is worthy of attention because of the rarity of the condition in this age group.

References