number of induction cycles allowed in this study was six whereas in our study the maximum number of induction cycles was eight and the median number of cycles to achieve CR was six. Thus, more than half of the CRs were achieved after receiving a seventh or eighth induction cycle in our study and it is possible that the CR proportion would have exceeded 29.7% in the other study if more induction cycles had been delivered. Further consideration of the optimal number of induction cycles is warranted.

Progression-free survival in our study was shorter than that reported for some combination regimens in patients with newly diagnosed PCNSL.[4] Moreover, the proportion of patients who experienced progression outside the CNS was higher than anticipated. However, salvage therapy was successful in many of our patients as demonstrated by the prolonged median overall survival which compares favorably to combination and WBI-containing regimens. High-dose methotrexate alone or in combination with other therapies is the most effective treatment available for PCNSL. Future studies are necessary to identify the optimal methotrexate-based combination regimen that will produce maximal efficacy and acceptable toxicity.

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THE FIRST CEREBROTENDINOUS XANTHOMATOSIS FAMILY FROM ARGENTINA: A NEW MUTATION IN *CYP27A1* GENE

Cerebrotendinous xanthomatosis (CTX) is a treatable, autosomal recessive, lipid storage disorder characterized by diarrhea and cataracts, usually appearing in the first decade of life, followed by growth of tendon xanthomas and progressive neurologic disability.[1] The typical brain findings include MRI signal abnormalities mainly in the globus pallidus and dentate nuclei.[2]

The disease is caused by deficiency of the mitochondrial enzyme, sterol 27-hydroxylase, resulting in impaired primary bile acid synthesis, decreased chenodeoxycholic acid production, and cholestanol accumulation in virtually every tissue.[3] The main blood chemistry abnormalities include high plasma levels of cholestanol with normal to low cholesterol. Different mutations of the *CYP27A1* gene have been described.[4] Early diagnosis is crucial as treatment with chenodeoxycholic acid (CDCA) may improve symptoms.[5]

Supplemental data at www.neurology.org

Here we report the cases of two Argentinian siblings with a novel mutation of the *CYP27A1* gene associated with clinical variants, including absence of tendon xanthomas.

Case reports. ***Case 1 (male, 17 years).*** The first clinical manifestation was chronic diarrhea from birth and growth retardation. Episodes of febrile seizures occurred at 1 and 3 years, followed by photosensitive epileptic syndrome. Posterior subcapsular cataracts were found at 3 years and removed at 8 years of age. Mild mental retardation, attention deficit, and hyperactivity were reported at school age. At 17 years of age physical examination showed low height and weight, brownish dental enamel, long upper extremities, scoliosis, and lumbar hyperlordosis. Neurologic examination showed palatal myoclonus, dystonic posture, distal tremor, and uncoordinated gait. Tendon reflexes were diffusely hyperactive. No tendon xanthomas were found. EEG showed abnormal response to photostimulation. T2-W MRI revealed a very small hyperintense area near the