CASE REPORT

A THAI PATIENT WITH LEBER’S HEREDITARY OPTIC NEUROPATHY LINKED TO MITOCHONDRIAL DNA 14484 MUTATION

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Abstract. A young Thai male presented with bilateral visual loss and disc pallor. The 14484 mutation responsible for Leber’s hereditary optic neuropathy (LHON) was identified on blood mitochondrial analysis. His visual loss was more severe than the visual loss described in Caucasian and Japanese patients and showed no improvement. He had no other identifiable mutations related to LHON nor any associated neurological disorder. This is the first case report of LHON with the 14484 mutation in a Thai patient.

Leber’s hereditary optic neuropathy (LHON) is a maternally inherited ocular disorder associated with a mutation in mitochondrial DNA (mtDNA). It is characterized by acute or subacute bilateral painless, simultaneous or sequential loss of vision. The most common primary mutations are identified at nucleotide position (np) 11778, 3460 and 14484. The 11778 mutation is the only mutation that has so far been reported in Thai LHON patients (Lertrit et al, 1999; Chuenkongkaew et al, 2001). We present the first case of LHON with the 14484 mutation in a Thai patient.

A healthy 33-year-old Thai man developed visual loss in both eyes over 18 months. He had a history of a fall from a building site 7 years ago and he was also a heavy smoker. He has a brother and a sister who are both healthy. His pedigree is shown in Fig 1. Eye examination revealed a visual acuity of 1/60 in both eyes. The optic discs were pale bilaterally. The rest of the neurological examination was normal. A central visual field defect was detected in both eyes using Goldmann perimetry.

A 14484 mutation was detected in his blood mtDNA by restriction enzyme analysis and sequencing methods (Fig 2). His visual acuity remained stable throughout the 8 months follow-up period.

The LHON mtDNA point mutation at np 14484 was first described in 1992 (Johns et al, 1992). Unlike the mutations at np11778 and 3460, this mutation is identified in the NADH dehydrogenase subunit 6 gene which changes methionine-64 to valine. In Caucasian patients with LHON, the 14484 mutation has been reported in 10-15%, while the 11778 and 3460 mutations have been documented in 31-90% and 8-15% respectively (Newman, 1995). Although this mutation has been documented as a primary mutation, the combination of 11778 and 14484 has recently been reported (Riodan-eva et al, 1995; Brown et al, 2001).

The vision loss in patients with the 14484 mutation is usually less severe than in those with the 11778 and 3460 mutation. Approximately a half
of these patients have a greater likelihood of spontaneous recovery, particularly in the patients with an early onset of loss of vision (Johns et al., 1992; Mackey and Howell, 1992; Oostra et al., 1994; Riodan-eva et al., 1995; Funalot et al., 1996; Yamada et al., 1997).

A 11778 mutation has been the only mutation which has so far been identified among Thais with LHON. However, this is the first reported case of a young Thai man with LHON with the 14484 mutation.

REFERENCES


