

## Alternate-Sided Homonymous Hemianopia as the Solitary Presentation of Mitochondrial Encephalomyopathy, Lactic Acidosis, Stroke-Like Episodes Syndrome

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Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS) syndrome has various presentations. We report on a case of MELAS in which alternate-sided homonymous hemianopia was the main symptom of recurrent neurological defects. A 19-year-old woman suffered from blurred vision, headaches, vomiting, and fever that subsided within days. The ophthalmic examination demonstrated right homonymous hemianopia. One month later a similar episode occurred again. Computed tomography (CT) and magnetic resonance imaging (MRI) of her brain revealed an infarct in the left temporo-occipital lobes. Exercise tests showed lactic acidosis, and a muscle biopsy confirmed the diagnosis of mitochondrial myopathy. Half a month later, a third episode occurred. Visual field examination demonstrated left homonymous hemianopia and partial recovery of the right visual field. The infarct in the brain, as revealed by CT and MRI, was compatible with the visual field changes. MELAS should be ruled out in young patients who present with homonymous hemianopia accompanied by recurrent headaches or other recurrent symptoms. (*Chang Gung Med J* 2003;26:199-203)

**Key words:** mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS) syndrome, homonymous hemianopia, alternating sides.

Mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes (MELAS) syndrome is a multisystem mitochondrial disease, which involves muscles and the central nervous system.<sup>(1)</sup> It has been defined on the basis of (1) stroke-like episodes (with computed tomographic or magnetic resonance imaging evidence of focal brain abnormalities); (2) lactic acidosis, ragged red fibers, or both; and (3) at least 2 of the following: focal or generalized seizures, dementia, or recurrent headaches and vomiting.<sup>(2)</sup>

The initial presentation of neurological defects includes homonymous hemianopia, cortical blind-

ness, hemiparesis, spasticity, and dystonia.<sup>(2-5)</sup> Although hemianopia and cortical blindness are the most common manifestations,<sup>(2,6)</sup> they seldom occur solitarily. We report a case of MELAS who presented with alternate-sided homonymous hemianopia as the only neurological defect.

### CASE REPORT

A 19-year-old woman, who had generally been well in the past, suffered from sudden-onset blurred vision in November 2001. Headaches, nausea, and vomiting associated with fever were noted initially,

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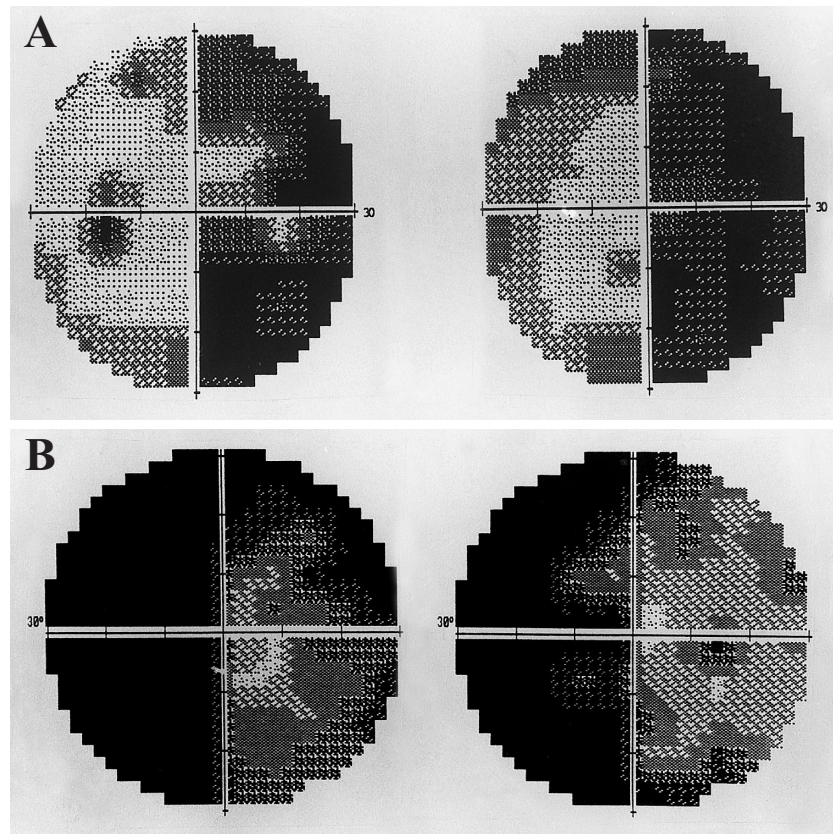
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but they subsided in a few days. Because of the persistence of the right hemianopia, she was referred for ocular examinations. Her visual acuity with her glasses was 20/20 in the right eye, and 20/50 in the left; these values were subjectively comparable with her vision before the attack. No limitations in extraocular muscle movements were found. The light reflex of both pupils was normal. No abnormality was found with slit-lamp and fundus examinations. Right homonymous hemianopia was revealed by examination of the visual field. The findings of visual evoked potential were normal.

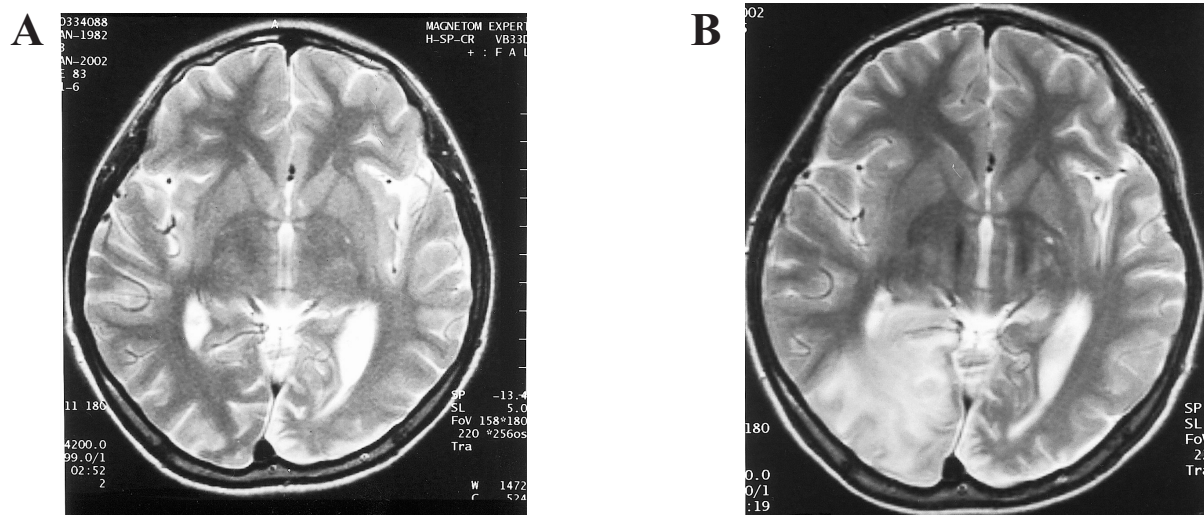
The visual field defect was stable for a month. However, a similar episode with headaches, vomiting, and blurred vision occurred in December 2001, but symptoms improved within 1 day. She went to the emergency department, and was admitted to the neurology ward for further evaluation. She had no

family history of stroke or seizures. Physical examinations revealed a short stature with a height of 154 cm and a body weight of 35 kg. No neurological motor defects were detected. Neither sign of dementia nor hearing loss was noted. Her visual acuity with her glasses was 20/30 in the right eye and 20/50 in the left. The results of a slit-lamp exam and direct ophthalmoscopy were unremarkable. Right homonymous hemianopia was found using a visual field examination (Fig. 1A). A computed tomographic (CT) scan of her brain showed encephalomalacic changes in the left occipital lobe. A magnetic resonance imaging (MRI) scan of the brain demonstrated a left posterior temporo-occipital lesion that was compatible with an infarction (Fig. 2A). Visual evoked potential performed later showed an abnormality in left visual pathway conduction.

To identify the cause of the recurrent stroke,



**Fig. 1** Visual field changes revealed by Humphrey 30-2 threshold tests. (A) Right homonymous hemianopia noted during the first admission. (B) Left homonymous hemianopia found with moderate depression of the right visual field which was also present after the third attack.



**Fig. 2** (A) MRI T2-weighted axial section showing an ill-defined hyperintense lesion involving the cortical region of the left posterior temporo-occipital lobe during the first admission. (B) After the third attack, the MRI T2-weighted axial section demonstrating high signal areas present in the right occipital, posterior temporal, and parietal lobes. Brain volume loss with scattered high-signal areas was found in the left occipital lobe.

hematological examinations, carotid artery echogram, and transcranial Doppler study were performed, but no abnormality was found. Under a suspicion of MELAS, an exercise test was performed which revealed lactic acidosis. The lactate level before exercise was 1.86 mmol/L (normal, 0.63-2.44 mmol/L), and the pyruvate level was 27.36  $\mu$ mol/L (normal, 34.2-102.6  $\mu$ mol/L). After exercise, the lactate level increased to 8.42 mmol/L, and the pyruvate level increased to 101.46  $\mu$ mol/L. Thus muscle biopsy was performed to confirm the diagnosis. Gomori-trichrome stain showed increased subsarcolemmal mitochondrial accumulation, which was further confirmed by reduced nicotinamide-adenine dinucleotide (NADH) staining. There were no apparent decreases in activities by cytochrome C oxidase stain. The pathologic changes in the muscle were compatible with mitochondrial myopathy.

Unfortunately another episode occurred in January 2002. She was re-admitted due to severe headaches and blurred vision. She felt that her vision had become worse. There was no other sensory loss or motor defect, except for left homonymous hemianopia. The best-corrected vision in the right eye was 20/50, and 20/100 in the left. The results of a visual field examination showed left homonymous

hemianopia, and moderate depression of the right-side visual field (Fig. 1B). Brain CT showed infarcts at the bilateral parieto-occipital lobes. An MRI of the brain revealed sequela of an infarct in the left occipitoparietal lobes, and subacute infarct in the right occipital, parietal, and temporal lobes (Fig. 2B). Diffuse cortical dysfunction with epileptic activities on the right side was found by electroencephalography. Visual evoked potential suggested a bilateral visual pathway conduction abnormality. Her condition stabilized, and she was followed up in the clinic after discharge from the hospital.

## DISCUSSION

The clinical presentation of MELAS syndrome is variable. It has been reported to mimic herpes encephalitis which presents with cognitive decline, hemiparesis, and hemianopia after herpes infection.<sup>(7)</sup> It could also be confused with a migrainous stroke, which presents as pulsatile headache and hemiparesis.<sup>(8)</sup> Although hemianopia and cortical blindness are the most common symptoms, they are usually accompanied by other neurological defects, such as hemiparesis, dementia, and seizures.<sup>(2,3,6)</sup> In addition to headaches and vomiting, our patient presented

with homonymous hemianopia as the only focal neurological sign in all 3 attacks. With CT and MRI scans, a parieto-occipital infarction was diagnosed. Without the associated neurological symptoms, MELAS was not initially suspected. It was the occurrence of a visual cortex infarction and the pathologic results of a muscle biopsy that drew us toward the final diagnosis.

The stroke-like episodes in MELAS have a propensity for the occipital area,<sup>(3,9)</sup> and about 10% of patients with mitochondrial disorders suffer from an occipital stroke before the age of 45 years.<sup>(10)</sup> The occipital cortex might be more susceptible to metabolic derangement secondary to mitochondrial dysfunction.<sup>(3)</sup> In the 3 episodes of our case, the hemianopia developed first on one side and then the other, representing alternate involvement of both occipital lobes. The visual field examination performed after the third episode revealed not only left homonymous hemianopia, but also the incomplete recovery from right hemianopia. This suggested that infarcts of each episode did not recover fully and left the sequela of moderate visual field depression.

MELAS syndrome may present in various forms, and should be suspected in patients with unusual strokes. Homonymous hemianopia may be the only focal neurological sign of MELAS syndrome, thus ophthalmologists should remain aware of this when managing patients with hemianopia accompanied by headaches, vomiting, or seizures.

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# 以交替性同側偏盲爲單獨表徵之粒腺體腦肌病伴乳酸性酸血症和中風狀發作症候群

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粒腺體腦肌病伴乳酸性酸血症和中風狀發作 (MELAS) 症候群有多樣化的表現，在本篇我們報告一個以交替性同側偏盲爲單獨表現的病例。一名19歲女性突發性視力模糊，併發頭痛、嘔吐、及高燒。在症狀穩定後，眼科檢查發現右側偏盲。一個月後，類似症狀再度發生。電腦斷層攝影及核磁共振造影發現腦部左顳葉及左枕葉有梗塞現象。運動測試則顯示有乳酸性酸血症 (lactic acidosis)。肌肉組織檢驗則證實爲粒腺體肌肉病變 (mitochondrial myopathy)。半個月後病症三度發作。視野檢查發現爲左側偏盲，右側視野部分恢復。而電腦斷層攝影及核磁共振造影則顯示腦部變化與視野相符。當病患患有同側偏盲徵兆，合併其他反覆發作的頭痛等症狀時，必須將MELAS症候群列入考慮。(長庚醫誌 2003;26:199-203)

**關鍵字：**粒腺體腦肌病伴乳酸性酸血症和中風狀發作 (MELAS) 症候群，同側偏盲，交替性。

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