

## Unilateral tonic pupil in spinocerebellar ataxia without brainstem atrophy

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### Abstract

We report a case of unilateral tonic pupil in spinocerebellar ataxia without brainstem atrophy in a 42-year-old man. On neurological examination, he showed cerebellar symptoms and unilateral tonic pupil. Deep tendon reflexes were normal except for brisk patellar tendon reflexes. Brain MRI demonstrated cerebellar atrophy only. There was neither orthostatic hypotension nor bowel and bladder failure. The right pupil constricted from 5.0 mm to 1.7 mm 60 minutes after 0.125% pilocarpine administration, whereas the left pupil did not change, remaining at 3.7 mm. Although it is not proven that tonic pupil is causally related to spinocerebellar ataxia, physicians must remain aware of spinocerebellar ataxia as a disease that can demonstrate tonic pupil.

### INTRODUCTION

Tonic pupil is due to postganglionic parasympathetic denervation of the iris sphincter associated with a variety of neurological diseases, and is usually associated with areflexia or hyporeflexia. Idiopathic cerebellar ataxia (IDCA) with additional extracerebellar features not corresponding to multiple system atrophy (IDCA-P) includes the autonomic disorder such as bladder dysfunction or constipation.<sup>1</sup> To date, there are no reports of tonic pupil associated with IDCA-P. We describe here the first patient of spinocerebellar ataxia without brainstem atrophy who showed a unilateral tonic pupil without hyporeflexia.

### CASE REPORT

A 40-year-old man, who had no family history for spinocerebellar ataxia or tonic pupil, noticed gait disturbance and dysarthria at age 39. He had a history of membranous glomerulonephritis from age 36, and there was no anisocoria at that time. The patient was an occupational therapist. On neurological examination, he showed saccadic eye movement without nystagmus, moderately scanning speech, and mild limb and truncal ataxia. Pupillary examination demonstrated 1 mm of anisocoria, with the right pupil (about 4 mm) being larger than the left (about 3 mm). Both pupils reacted promptly to light

stimulation. Enophthalmos and anhidrosis were not observed. Limb muscle tone, muscle strength and sensation were intact. Deep tendon reflexes were normal except for brisk patellar tendon reflexes. Pathological reflexes were not shown. Brain MRI demonstrated cerebellar atrophy only. His only medication was taltirelin hydrate (10 mg/day). Two years later, at age 42, he showed horizontal gaze-evoked nystagmus, anisocoria was accentuated and light reaction of the right pupil became sluggish. The right pupil did not constrict in response to convergence. Funduscopy remained normal bilaterally. Wechsler Adult Intelligence Scale-Revised (WAIS-R) scores were VIQ 86, PIQ 72, FIQ 78. There was neither orthostatic hypotension nor bowel and bladder failure. The results of <sup>123</sup>I-metaiodobenzylguanidine (<sup>123</sup>I-MIBG) myocardial scintigraphy, thermography, cystometry, and nerve conduction study were normal. Laboratory studies including erythrocyte sedimentation rate (ESR) and Vit. E level were also normal. The antinuclear antibodies (ANA), SS-A, SS-B, P-ANCA, and C-ANCA were negative. The cerebrospinal fluid examinations were normal. Ga-67 scintigraphy did not show any abnormal accumulations. Genetic studies excluded SCA 1 (spinocerebellar ataxia type 1), SCA3, SCA6, SCA7, SCA8, SCA12, SCA17, and 16q-linked ADCA (autosomal dominant cerebellar ataxia). Brain MRI showed cerebellar

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atrophy without brainstem involvement, which was the same as previously (Figure 1A,B). MR angiography (MRA) and cervical MRI did not demonstrate any abnormalities. The right pupil constricted from 5.0 mm to 1.7 mm 60 minutes after 0.125% pilocarpine administration, whereas the left pupil did not change, remaining at 3.7 mm (Figure 1C, D).

**DISCUSSION**

We considered his anisocoria as a clinical sign of Horner syndrome at first. However, enophthalmos and anhidrosis were not observed. Therefore we concluded his anisocoria was not due to Horner syndrome. His clinical course and dilute pilocarpine test revealed his anisocoria was due to tonic pupil.

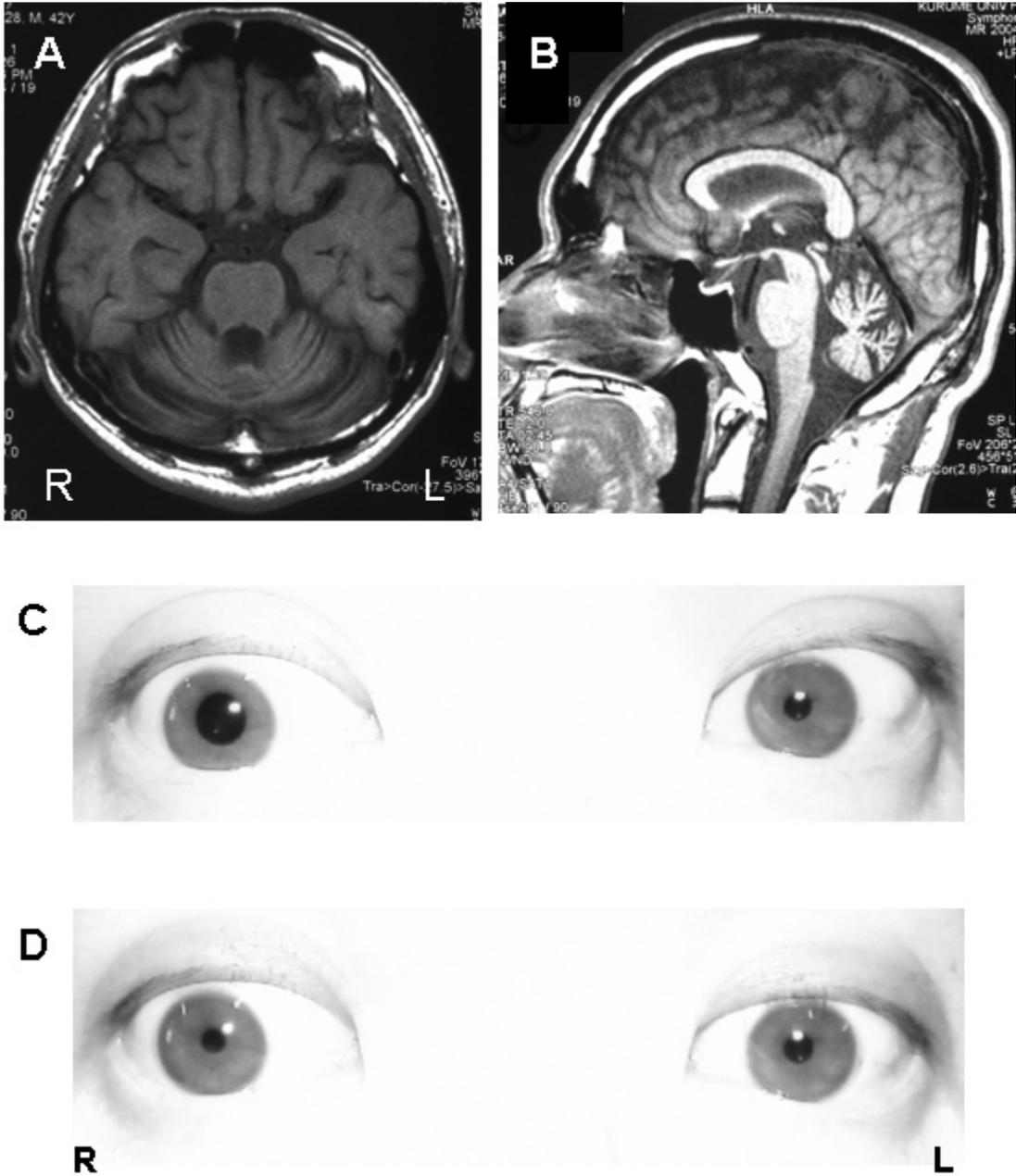


Figure 1 A,B: Brain MRI at presentation demonstrating loss of volume of the cerebellum. Brainstem and cerebral cortex show normal volumes. C,D: The right pupil constricted 60 minutes after instilling topical 0.125% pilocarpine, whereas the size of left pupil did not change.

This case demonstrated unilateral tonic pupil without hyporeflexia. Tonic pupil is commonly seen in patients with Holmes-Adie syndrome and Ross syndrome, which are characterized by hyporeflexia.<sup>2</sup> Recent case reports have described tonic pupils associated with giant cell arteritis<sup>3</sup>, Vogt-Koyanagi-Harada syndrome<sup>4</sup>, amyloidosis<sup>5</sup>, sicca syndrome<sup>6</sup>, polyarthritis nodosa<sup>7</sup>, paraneoplastic ganglionopathy with type-1 antineuronal nuclear autoantibodies<sup>8</sup>, and multiple system atrophy<sup>9</sup>, which usually show hyporeflexia or areflexia. However, serologic examination, MRI, ophthalmoscopy, Ga-67 scintigraphy, and electrophysiologic examination were not supportive of these diseases. In addition, our case did not show hyporeflexia but brisk patellar tendon reflexes and showed no autonomic failure except tonic pupil. Tonic pupils associated with flea spray and botulism have been reported<sup>10, 11</sup>; however the present case had no history of exposure to these toxins. Tonic pupil can result from supranuclear third nerve paresis<sup>12</sup> or an endodermal cyst of the third cranial nerve.<sup>13</sup> Brain MRI of our case demonstrated cerebellar atrophy only and neurologic examination did not show any abnormality of third nerve function. Thus, in our case, a unilateral tonic pupil was not due to supranuclear or preganglionic third nerve palsy, but due to postganglionic parasympathetic denervation of the iris sphincter according to cholinergic supersensitivity on dilute pilocarpine test.

There are no previous reports of tonic pupil associated with spinocerebellar ataxia in patient whose brain MRI showed only cerebellar atrophy without brainstem atrophy. In the present case, according to the patient's medical record, anisocoria and cerebellar ataxia are thought to have appeared at the same time. Therefore it is reasonable to think that tonic pupil was not a congenital state but an associated manifestation of spinocerebellar ataxia.

Idiopathic cerebellar ataxia (IDCA) include the patients with additional extracerebellar features (IDCA-P), such as bladder dysfunction or constipation, not corresponding to multiple system atrophy with prominent cerebellar symptoms.<sup>1</sup> Our case can be categorized under IDCA-P.

Although it is not proven that the observed tonic pupil is causally related to spinocerebellar ataxia, our patient demonstrated that physicians should be aware of spinocerebellar ataxia as a disease that can be associated with tonic pupil.

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