Syndrome of Progressive Ataxia and Palatal Myoclonus : A Case Report

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Abstract

A 46-year old man presented with progressive cerebellar ataxia for 5 years. Physical examination revealed palatal and tongue myoclonus, cerebellar gait, limb ataxia and spasticity of the lower extremities. The imaging studies including CT-scan and MRI of the brain revealed progressive pancerebellar atrophy and bilateral hypertrophic degeneration of inferior olives. The clinical course was slowly progressive. Various medications included anticonvulsants, benzodiazepines and antispasticity failed to abolish the abnormal palatal movement and ataxic syndrome. The syndrome of progressive ataxia and palatal myoclonus is a rare and unique neurodegenerative syndrome. The pathogenesis and treatment are still unknown.

Key word : Palatal Tremor, Palatal Myoclonus, Cerebellar Ataxia

Palatal myoclonus or palatal tremor is a rare condition and is characterized by brief, rhythmic involuntary movement of the soft palate and is often accompanied by synchronous contractions of muscles derived from the branchial arches, the tongue, diaphragm or limbs. The condition is usually a delayed clinical manifestation of an acute or subacute lesion in the brainstem or cerebellum[1,2]. Palatal myoclonus or tremor which is caused by an indentified lesion is classified as symptomatic palatal tremor (SPT)[1]. However, the abnormal movement may occur without any accompanying neurological manifestations or pathology of the nervous system. In this case, it is classified as an essential palatal tremor (EPT)[1]. The syndrome of patatal myoclonus and progressive ataxia is an extremly rare neurodegenerative syndrome and only 9 cases have been reported in the English literature[1,3-8]. This entity may be one of the multisystem degenerative disorders or spinocerebellar degenerative disorders. In order to stimulate further study of this entity worldwide, we herein report a case of this syndrome in Thailand.

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CASE REPORT

In 1992, at age 41, the patient noted progressively unsteady gait for one year. He was a motorcycle driver. He had first notified that he had difficulty in balancing during his driving and accidents frequently occurred. Later on he also had gait imbalance. He had no history of head injury, alcoholic drinking or drug addiction. He had no systemic illness and took no medication. In 1993, he was admitted to King Chulalongkorn Memorial Hospital for an evaluation of this symptom. The vital signs and general physical examination were unremarkable. Mental status examination was normal. Cranial nerve evaluation revealed an irregular continuous movement of the soft palate at a rate of 100 per minute. The palatal myoclonus was symmetrical and characterized by bilateral upward and backward movement of the soft palate and uvula before returning to the resting position. No fasciculation, atrophy or other abnormal movements of the tongue and other bulbar, facial or ocular musculature were detected. The limb strength and muscular tone were normal. Deep tendon reflexes were +2 all. Plantar responses were flexor. He had mild cerebellar standing balance impairment characterized by increased sway and could barely stand with his feet together. He also had mild cerebellar ataxic gait. No limb ataxia, postural tremor, slurred speech or ocular movement impairment were observed. The sensory system and the rest of the neurological examinations were normal. No Keyser-Fleicher ring was detected under slit lamp. He had one elder brother, 2 sons and 1 daughter. There was no neurological disorder in his family.

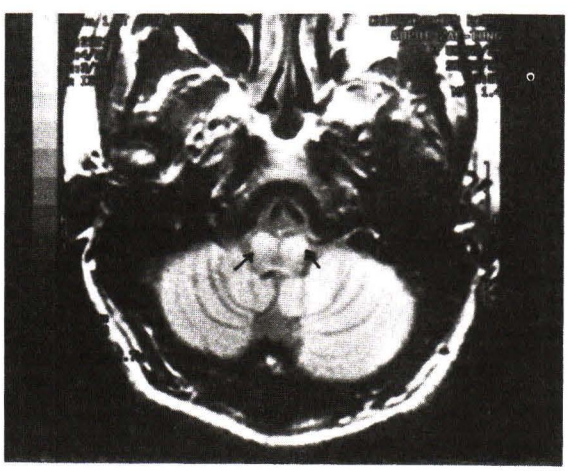

Fig. 1. MRI (PD) section at the level of upper medulla revealed increased signal intensity and bilateral enlargement of inferior olives (arrow).

Laboratory results including complete blood count, urinalysis, erythrocyte sedimentation rate, plasma glucose, blood urea nitrogen, lipid profiles, creatinine, liver function tests, thyroid function tests, blood ceruloplasmin, anti-HIV antibody, cerebrospinal fluid analysis, serum VDRL, CSF VDRL were within normal limit. CT-scan of the brain was unremarkable. MRI showed a bilateral increased signal intensity and enlargement of the inferior olives (Fig. 1). There was no evidence of cerebral infarction, infiltrative lesion, inflammatory or demyelinating processes. Electroencephalography and electrocardiography were normal. Treatment with clonazepam, trihexyphenidyl, baclofen, carbamazepine, valproic acid was of no benefit.

During the four years of regular follow-up, he had no concurrent systemic illness, but the ataxic syndrome was progressive. He could not perform his job as a driver. The repeated general physical examinations were normal. Neurological examination in 1997 revealed the previous neurological deficits plus myoclonus of the tongue, spasticity with no definite weakness of both legs, and bilateral extensor plantar responses. The deep tendon reflexes were +2 on the upper extremities and +4 on the lower extremities. No sensory deficits or autonomic disturbances were observed. The standing balance impairment had increased and there was mild gait and limb ataxia. Neurophthalmologic and neurootologic examinations were unremarkable. The follow-up CT-scan revealed evidence of progressive pancerebellar atrophy, but follow-up MRI was not performed (Fig. 2, 3).

DISCUSSION

Palatal myoclonus has many synonyms e.g. rhythmic palatal myoclonus, oculopalatal myoclonus, palatal nystagmus, brainstem or palatal myorhythmia[1-3]. It is a rare movement disorder[1-3]. Since the movement fulfills the criteria of a tremor, it was reclassificaled among tremor i.e. palatal tremor[1]. The palatal myoclonus is caused by a lesion in the central tegmental tract, red nucleus to inferior olive and occasionaly of dentate nucleus [1-3]. The pathological finding which is associated

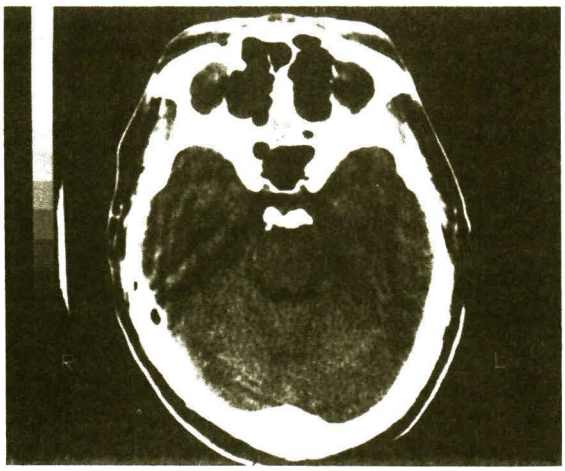

Fig. 2. CT-scan of the brain in 1993 revealed normal cerebellum.

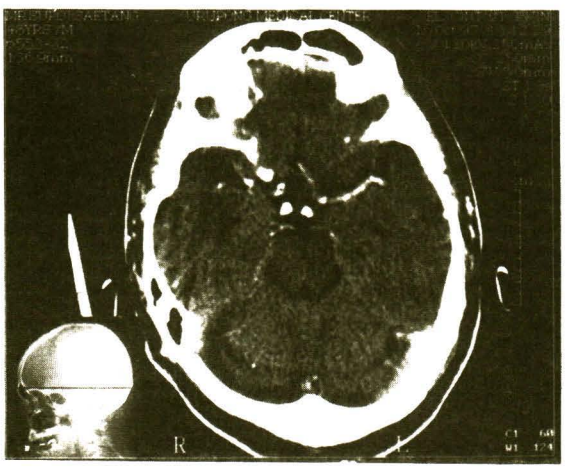

Fig. 3. CT-scan of the brain in 1997 revealed prominent cerebellar folia which indicated a cerebellar degenerative process.

with palatal myoclonus is an enlargement of inferior olives attributed to pseudohypertrophic transneural degenerations from interuption of dentato-olivary pathways[1-3]. Microscopical features are swollen olivary neurons and astrocyte proliferation[9]. In cases with a long clinical course pseudohypertrophy may result in atrophic changes[9]. The etiology of the lesion which causes the interruption is often an infarction but syndrome has been reported in inflammatory, neoplastic, traumatic and paraneoplastic processes[1-3]. The degenerative process as a cause of palatal tremor is rare[1-3]. Among the degenerative processes, a distinct syndrome of progressive ataxia and palatal myoclonus has been described[1,3-8]. There have been only 9 cases of this entity reported in the English literature[1,3-8]. These cases had an unique clincal syndrome of palatal myoclonus plus cerebellar ataxia. The myoclonus in these cases included palatal, buccal, ocular, eyelid, chin, laryngeal, pharyngeal and lip myclonus[1,3-8]. The ataxic syndrome consisted of gait ataxia, slurred speech, intention tremor of limbs, head and neck tremor, nystagmus[1,3-8]. Other associated neurological abnormalities included hyperreflexia, Babinski signs, dysarthria, limitation of lateral gaze, nasal speech, sluggish pupils, limb amyotrophy, impaired visual acuity and dementia[1, 3-8]. The imaging findings were bilateral hypertrophic degeneration of inferior olivary nuclei[6]. However, the pathological data were not available for a definite etiological conclusion. This entity was suggested to be a multisystem atrophy[6] or spinocerebellar degenerative disorder[8]. Several substances such as anticholinergic drugs, anticonvulsants, benzodiazepines, 5-hydroxytryptophen have been tried to improve the palatal myoclonus but have rarely been successful especially in the symptomatic form[3].

The present case had a progressive ataxia and palatolingual myoclonus. He had no stroke risk factors. Imaging findings revealed no evidence of a vascular lesion, demyelinating disease, inflammatory precess or tumor infiltration. The MRI revealed degenerative hypertrophy of inferior olives. The MRI finding was identical to Sparling and Herrmann's case[6]. The long term follow-up in this case revealed progression of the clinical manifestations of cerebellar ataxia, palatal myoclonus and spasticity of both lower extremities. The CT-scan of the brain demonstrated progressive pancerebellar atrophic changes. The clinical profiles and imaging changes in this patient favoured the diagnosis of spinocerebellar degenerative disorder. However, this syndrome has not been mentioned among the classical spinocerebellar degenerative disorders[10]. Further studies for data concerning the clinical pattern, genetic aspects, neuropathology, biochemistry and molecular biological aspects of this syndrome is necessary inorder to shed light on this rare but interesting syndrome.

SUMMARY

A case of progressive ataxia and palatal myoclonus was studied. The clinical profiles consisted of progressive cerebellar ataxia, palatolingual myoclonus and spasticity of the lower extremities. Structural studies using CT-scan and MRI of the brain indicated a progressive pancerebellar atrophy and degenerative hypertrophy of inferior olives. No associated systemic disease was observed after a long term follow-up.

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กลุ่มอาการเดินโซเซและเพดานปากกระตุก : รายงานผู้ป่วย

*กัมมันต์ พันธุมจินดา, พ.บ.**

ผู้ป่วยชายไทย อายุ 46 ปี มาด้วยอาการเดินโซเซเหตุโรคสมองน้อยฝ่อมาเป็นเวลา 5 ปี การตรวจร่างกายพบอาการกระตุกของเพดานปากและลิ้นร่วมกับอาการเดินโซเซ และแขนขาเซจากรอยโรคของสมองน้อยและอาการเกร็งของขาสองข้าง. การตรวจร่างกายและการบันทึกภาพด้วยคอมพิวเตอร์สมองและการตรวจสมองด้วยเอ็มอาร์ไอ พบสมองน้อยฝ่อที่เป็นมากขึ้น, ร่วมกับการเสื่อมที่ทำให้อินฟีเรียโอลิฟทั้ง 2 ข้างโตขึ้น. การดำเนินโรคในผู้ป่วยรายนี้เป็นมากขึ้นเรื่อย ๆ อย่างช้า ๆ. การรักษาอาการต่าง ๆ ด้วยยากันชัก, ยาในกลุ่มเบนโซไดอาเซปีนและยาลดเกร็งต่าง ๆ พบว่าไม่ได้ผลในเรื่องการลดอาการกระตุกของเพดานปากและอาการเซ. กลุ่มอาการนี้ เป็นกลุ่มอาการที่พบน้อยและต้องแยกจากโรคเสื่อมชนิดอื่น ๆ ของทางระบบประสาท. พยาธิกำเนิดและการรักษากลุ่มอาการนี้ยังคงไม่ทราบในปัจจุบัน

คำสำคัญ : เดินโซเซ, เพดานปากกระตุก

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