Familial Paroxysmal Dyskinesia

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Abstract

Familial paroxysmal dyskinesia is characterized by recurrent episodic dystonia and/or choreoathetosis with totally quiescent intervening periods. It is an autosomal dominant with variable penetrance basal ganglia disorder. An 11 year old girl who presented with brief kinesigenic paroxysmal dyskinesia is reported. The abnormal movements were dramatically controlled by diphenylhydantoin. Spontaneous remission was seen in the elder sister of this family.

Familial paroxysmal dyskinesia (FPD) is a rare and bizarre neurological disorder first described by Mount and Reback[1]. It is an autosomal dominant with variable penetrance basal ganglia disorder[2-4]. The movement disorder is stereotyped and easily recognizable. However, it is still frequently misdiagnosed as epilepsy or a psychogenic disorder. FDP is characterized by either brief (less than 5 minutes) or prolonged (more than 5 minutes up to a day) attacks of dystonia and or choreoathetosis[2]. The attacks may occur during the day or night[2]. FPD has been described in many parts of the world[2]. We report a patient with FPD who had brief and kinesigenic attacks. To our knowledge, this may be the first report of its occurrence among the Thais.

CASE REPORT

A girl, aged 11 complained of transient episodes of a movement disorder which began at the age of 8 years. During the first two years, the attacks occurred once a week. The girl was able to suppress or sometimes abort the attack by stopping the ongoing activity. She was able to prevent an attack by starting to move slowly or by forcefully holding the affected arms together for a moment (Fig. 1). No definite diagnosis was given and her attacks did not respond to tranquilizers. One year before admission, the attacks became more troublesome with a frequency of 5-10 episodes a day. She had two elder sisters. The eldest sister aged 14 years was symptom-free. The elder sister aged 13 years had the same invo-

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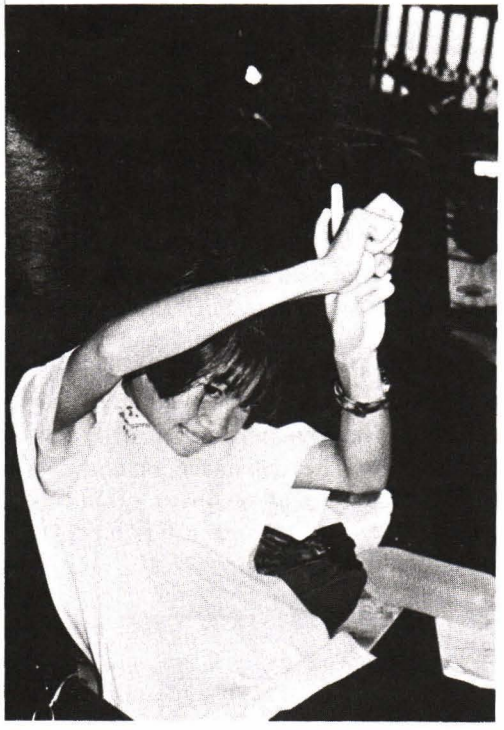

Fig. 1 The girl during an attack at home, facial grimacing, dystonic posture and athetotic movement of arms were demonstrated.

luntary movements which began at the age of 10 years. The frequency of attacks was about once a month. Her abnormal movements had a spontaneous remission after 2 years of attacks. Our patient and her affected elder sister had a normal delivery, growth and development. They had good learning records and had no systemic illness. The parents were not consanguineous and were free from neurological or systemic diseases.

During the attacks, observed at Chulalongkorn Hospital, the patient showed facial grimacing, dystonic posturing of the head, body and extremities. The dystonia affected the arms more than legs. During the spells, the arms were in supinated position and there was dystonic arm abduction, elbow flexion, wrist extension, and athetotic movement of fingers. Leg extension with equinovarous was observed. The episodes lasted 10 seconds and were unaccompanied by altered consciousness, incontinence, postictal amnesia, Todd paralysis, or confusion. The dystonic spasms were painless but impaired her posture and gait. Her dyskinesic episodes occurred when she had quick unintentional movements with a change of posture after a period of rest. The involuntory movements were not preceeded by a sensory aura such as tingling, numbness, pins and needles sensation. However, she did have a prodromal sensation of tightness in her extremities just before the abnormal movements. Her mental state was normal. Neurological examination revealed no abnormality between attacks. Systemic examination revealed no K-F ring or Trousseau sign. A complete blood count, blood chemistries including BUN, creatinine, thyroid function tests, liver function tests, calcium, phosphate, serum ceruloplasmin were within normal limits. Studies to exclude autoimmune diseases and rheumatic fever included LE preparation, ANA, complement levels, rheumatoid factors, ASO titer, and ESR were all unremarkable. Electroencephalogram and MRI of the brain were normal.

A diagnosis of brief familial paroxysmal dyskinesia (BFPD) was made and diphenylhydantoin 50 mg twice a day was prescribed. The attacks were completely controlled with this medication. The attempt to withdraw diphenylhydantoin and a trial of placebo brought back attacks. During one and a half years of follow-up, she still needed anticonvulsant in order to control her abnormal movements.

DISCUSSION

Paroxysmal dyskinesia is characterized by recurrent episodic dystonia and/or choreoathetosis with totally quiescent intervening periods[2,3]. This type of abnormal movement may be classified by familial history, diurnal variation, duration of the attacks and known or unknown causes[2,3]. FPD is separated into brief (BFPD) or prolonged (PFPD) forms according to the duration of paroxysms being less or more than 5 minutes[2,3]. FPD may be further categorized as daytime or nocturnal paroxysmal dyskinesia[2,3]. The BFPD is triggered by brief sudden movement after a period of sitting or lying at rest or kinesigenic paroxysmal dyskinesia[2-4]. The attacks may occur daily and are successfully treated with anticonvulsants[5]. The character of abnormal movement is usually choreoathetotic or athetotic[2-4]. Dystonic posturing or a mixture of choreoathetotic and dystonic posturing as occurred in this case is less common[3]. Occasionally the abnormal movement is characterized

by tonic spasm[3]. The frequency of attacks ranges from 1 to more than 100 times per day and there is usually 15-20 minute-refractory period between the attacks[6]. Anxiety, excitement and tension may increase the paroxysms[2-4,7,8]. Most patients have a prodrome before the attacks in the form of parethesias or a sensation of tightness[2-4,7,8]. The PFPD is characterized by choreoathetosis or tonic flexor spasms[2]. It is usually nonkinesigenic and is precipitated by alcohol, physical exhaustion, coffee, stress, exposure to cold and anxiety[2]. The prolonged type occurs less frequently and the paroxysms last more than 5 minutes and may last more than one hour[2]. They are not influenced by anticonvulsants but may respond to benzodiazepines or acetazolamide[5]. Familial paroxysmal hynogenic dystonia is characterized by flexion spasms or dystonia during sleep and the attack may be brief or prolonged[9]. A brief attack may respond to anticonvulsants but the prolonged attacks may resist pharmacotherapy[5]. Our patient had the classical picture of BFPD. The differential diagnosis includes: reflex epilepsy, partial seizure, structural lesions in basal ganglia, cerebral palsy, multiple sclerosis, drug-induced dyskinesia and metabolic disease such as idiopathic hypoparathyroidism[10,11]. These disorders were excluded by clinical profiles and appropriate investigations. The frequency and severity of the dyskinesias in BFPD tend to vary within the family[2]. The elder sister in this family had rather mild and infrequent attacks. The age of onset of BFPD usually begins between 8-12 years and three-quarters of cases are male[2]. The onset of dyskinesia in this family was 8 and 11 years. However, both of the patients were female. The clinical course of BFPD is usually benign. The frequency of the attacks usually increases during aldolesence[2-4]. The attacks decrease in adulthood, but continue throughout life[2-4]. Spontaneous remission, as seen in the elder sister of this family, is uncommon[3]. In BFPD the patern of inheritance is autosomal dominant[2-4]. In the present family, two siblings but no other family members were affected. This suggests a new mutation or autosomal dominant gene with incomplete penetrance. No conclusive pathological data exists in FPD[4]. Physical examination and extensive systemic and neurological investigations are usually normal. However, seizure disorders may be associated with BFPD[2,12,13]. The pathophysiology of paroxysmal dyskinesia is still unknown. The imbalance in corticostriatopallidothalamocortical feedback circuit is suggested to play a major role in this bizzare movement disorder[5,14,15].

SUMMARY

An 11 year-old girl presented with brief kinesigenic paroxysmal dyskinesia. The attacks were characterized by facial grimacing, neck, extremities and body dystonic posturing, and athetotic movement of the fingers. No alteration of consciousness, incontinence, postictal paralysis or amnesia were observed during the attacks. The attacks lasted about 10 seconds. The girl was able to suppress or abort the attacks by stopping ongoing activities or starting to move slowly. The abnormal movements were dramatically controlled by diphenylhydantoin. One of her elder sisters also suffered from identical but mild and infrequent similar symptoms. Her sister had spontaneous remission after two years of attacks. Extensive investigations failed to disclose any associated systemic or neurological abnormalities.

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การเคลื่อนไหวผิดปกติที่เป็น ๆ หาย ๆ ชนิดที่พบเป็นในครอบครัว

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ผู้ป่วยเด็กหญิงอายุ 11 ปี มาโรงพยาบาลด้วยอาการการเคลื่อนผิดปกติที่เป็น ๆ หาย ๆ ในระยะสั้น และสัมพันธ์กับการเคลื่อนไหว ขณะเกิดอาการจะพบลักษณะบิดเบี้ยวของใบหน้าคอแขนขาและลำตัวแบบดิสโทเนีย และการเคลื่อนผิดปกติแบบปัดไปมาแบบอะทีโตซีสของนิ้วมือ ระหว่างเกิดอาการเคลื่อนไหวผิดปกติดังกล่าว ผู้ป่วยยังมีสติสัมปชัญญะปกติ ไม่มีปัสสาวะราดและไม่พบอาการอ่อนแรงหรือหลงลืมภายหลังเกิดอาการ อาการเป็นนานครั้งละ 10 นาที การหยุดการเคลื่อนไหวขณะเริ่มมีอาการสามารถหยุดหรือทำให้อาการดังกล่าวลดน้อยลง และการเริ่มเคลื่อนไหวช้าจะป้องกันอาการดังกล่าวได้ การเคลื่อนไหวดังกล่าวตอบสนองอย่างดีกับการรักษาด้วยยากันชักไดเฟนนิไฮแดนโตอิน ผู้ป่วยมีพี่สาวหนึ่งคนซึ่งมีอาการเช่นเดียวกับผู้ป่วย แต่มีอาการรุนแรงน้อยกว่าและอาการดังกล่าวไม่เป็นถี่มาก พี่สาวของผู้ป่วยหายเองหลังจากมีอาการดังกล่าวเป็นเวลา 2 ปี ไม่พบความผิดปกติจากการตรวจค้นหาโรคทางกายและโรคทางระบบประสาทอื่น ๆ ในผู้ป่วยรายนี้

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