Paroxysmal Kinesigenic Choreoathetosis

Il Saing Choi, Jin Ho Kim*, and Won Young Jung

Paroxysmal kinesigenic choreoathetosis (PKC) is characterized by short paroxysms of focal or generalized involuntary movement induced by sudden movements, and is a well-known disease in the neurologic literature, but only 4 cases have been reported in Korea. The purpose of the presentation is to clarify the clinical features of PKC in Korea. We clinically analyzed 20 patients with PKC between 1986 and 1994 at Yongdong Severance Hospital, Yonsei Medical Center, with a minimum of a 1 to 2 year follow-up period. There were 14 men and 6 women. The age at onset of the condition ranged from 8 to 17 years (mean, 13.1 years). Six patients (30%) had a family history of the condition and the mode of inheritance was suggestive of an autosomal recessive pattern. The involuntary movements seemed to be dystonic rather than choreoathetotic upon a mild attack, and the paroxysms were precipitated by sudden movements. The attacks occurred on one or both sides, and were often associated with dysarthria, upward gaze and sensory aura. Consciousness was never lost. Their duration were usually 10 to 30 seconds, and never more than two minutes. All laboratory tests including electroencephalographic and neuroimaging studies showed no abnormality. All patients responded well to diphenylhydantoin. PKC is not rare in Korea and has a benign course.

Key Words: Paroxysmal kinesigenic choreoathetosis, autosomal recessive inheritance, diphenylhydantoin

Paroxysmal kinesigenic choreoathetosis (PKC) is characterized by short paroxysms of unilateral or bilateral involuntary movement and posturing without loss of consciousness which are usually precipitated by sudden movement (Kertesz, 1967; Lance, 1977; Plant, 1983).

Since Mount and Reback (1940) first presented a case entitled “familial paroxysmal choreoathetosis”, it has been well documented in the neurologic literature, but that only 4 cases have been reported in Korea (Shin et al. 1985).

The purpose of this presentation is to clarify the clinical features of PKC in Korea by analyzing 20 patients and reviewing the literature.

MATERIALS AND METHODS

Between 1986 and 1994, we clinically examined 20 patients with PKC at Yongdong Severance Hospital, Yonsei Medical Center.

Routine laboratory tests including a CBC, urinalysis, SMA-12, chest x-ray and ECG, electroencephalography (EEG) and brain computed tomographic (CT) scan were performed in all patients, and a brain magnetic resonance imaging (MRI) study was done in 2 patients. An anticonvulsant, diphenylhydantoin (200 to 300 mg daily) was used in attempts to treat PKC.

Twenty patients were classed according to age and gender, inheritance, clinical symptoms,
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duration of attack, provocation, drug response and outcome.
All completed the one to two-year follow-up period.

RESULTS

The clinical summary is seen in the Table 1.

Of the 20 patients with PKC, there were 14 men and 6 women. Age at the onset of the condition ranged from 8 to 17 years (mean, 13.1 years) and the duration of illness varied from 1 month to 13 years (mean, 5.1 years). Six patients (30%) had a family history of PKC and the mode of inheritance was suggestive of an autosomal recessive pattern.

The attacks were all provoked by sudden movements such as quickly standing from sitting, hopping and running. During an attack, the fingers flexed in a spasm and the arm flexed at the elbow and abducted at the shoulder. The leg extended, and the foot flexed plantarly and inverted. In addition, 2 patients were associated with dysarthria and upward gaze, and 4 had a sensory aura immediately before the attack.

Consciousness was never lost. The paroxysms occurred on one and/or both sides respectively. Of the 20 patients with PKC, 10 had unilateral attacks (6 right sided attacks and 4 left sided attacks), 6 unilateral and bilateral attacks, and 4 bilateral attacks. The duration of paroxysms was usually less than 30 seconds, and never more than two minutes.

All laboratory tests including EEG and neuroimaging studies showed no abnormality.

All patients responded well to diphenylhydantoin. Two patients had a family history whereby their parents or relatives had complete remission after the age of 23.

DISCUSSION

Records of movement-induced attacks similar to PKC can be found scattered in the literature before the report of Mount and Reback (1940). A year after Mount and Reback's article, Smith and Heersema (1941) reported 3 cases of "periodic dystonia", and Lance (1963) presented a family with attacks very much like the case of Mount and Reback under the title "sporadic and familial variety of tonic seizures". Since the term PKC was introduced by Kertesz (1967), it has generally become accepted to describe PKC as an easily recognizable disorder in which brief paroxysms are precipitated by sudden movement. PKC is seen in children and young adults (Kertesz, 1967; Lance, 1977; Plant, 1983). The onset age reported herein was between the ages of 8 and 17, which was comparable to

| Table 1. Clinical summary of 20 patients with paroxysmal kinesigenic choreoathetosis |
|--------------------------------------|----------------------------------|
| Age at onset                         | 8 to 17 years (mean, 13.1 years) |
| Sex ratio, M:F                       | 14:6                             |
| Family history                       | 6 patients (30%) (autosomal recessive) |
| Provocation                          | Sudden movements such as standing from sitting, hopping and running |
| Affected side                        | 6 patients 10 patients |
| 1. Unilateral                        | 4 patients |
| 2. Unilateral and bilateral          | 6 patients |
| 3. Bilateral                         | 4 patients |
| Duration of paroxysm                 | less than 30 seconds |
| Loss of consciousness                | Never |
| Drug response                        | Good |
| Outcome                              | Benign course |

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those of most reports except the reports of Mount and Reback (1940) and Lance (1963). The gender ratio reported favors boys (Kertesz, 1967; Lance, 1977; Gooenough et al. 1978; Plant, 1983), as 1 found. The familial occurrence is undoubtedly a significant feature. The mode of inheritance is either an autosomal recessive (Mount and Reback, 1940; Kertesz, 1967) or autosomal dominant pattern (Lance, 1963; Hudgins and Corbin, 1966). The study herein was suggestive of an autosomal recessive pattern.

One of the most remarkable and consistent features of the disease is the precipitation of attacks by sudden movement (Kertesz, 1967; Plant, 1983). The most commonly cited example is where the patient is called and rises quickly from a sitting position. During an attack, the fingers flex in a spasm, and the arm flexes at the elbow and abducts at the shoulder. The leg extends, and the foot flexes plantarily and inverts. In addition, dysarthria, an upward gaze and grimacing face are often associated, but consciousness is never lost. Some patients have a sensory aura immediately before attack. The paroxysms occur on one and / or both sides (Plant, 1983), and the duration of paroxysms is usually less than 30 seconds, never more than 2 minutes (Kertesz, 1967; Lance, 1977). All laboratory tests show no abnormality except acquired PKC (Lishman et al. 1962; Kertesz, 1967; Gooenough et al. 1978; Suber and Riley, 1980; Plant, 1983). Diagnosis can be easily made by a clinical history and features. The differential diagnosis would include reflex epilepsy (Lishman et al. 1962; Whitty et al. 1964), metabolic diseases such as tetany (Tabae-Zadeh et al. 1972; Soffer et al. 1977; Neman and Kinkel, 1984), acquired PKC, hysteria and etc (Rosen, 1964; Lance, 1977; Gooenough et al. 1978; Watson and Scott, 1979). The patients respond well to anticonvulsants such as diphenylhydantoin (Kertesz, 1967; Homan et al. 1980; Kinast et al. 1980), carbamazepin (Kato and Araki, 1969), phenobarbital, primidone (Garello et al. 1983) and valproic acid (Suber and Riley, 1980), and L-dopa (Loong and Ong, 1973). All patients herein responded well to diphenylhydantoin.

Mount and Reback (1940), Rosen (1964), and Kertesz (1967) described cases which terminated after the age of 20, and 2 patients reported herein had a family history where their relatives had complete remission after the age of 23.

The pathogenesis of PKC is unknown, but there are several hypothesis.

Lishman et al. (1962), Hudgins and Corbin (1966), and Stevens (1966) discussed the mechanism of paroxysms and concluded that it should be considered as a variety of reflex epilepsy because of the paroxysms nature of the attacks and the response to anticonvulsants. However, the essentially normal EEG in all patients, the lack of family or past history of epilepsy, and the preservation of consciousness would tend to set this entity apart from convulsive disorders.

Calcium metabolism was investigated by Lishman et al. (1962) because the movement during some of attacks resembled a carpopedal spasm. No abnormality was found in the electrolytes studied.

Most recent authors have considered PKC to be a disorder of basal ganglia function rather than a form of epilepsy (Loong and Ong, 1973; Homan et al. 1980; Plant, 1983).

The evidence for this arises from the character of the attacks, the lack of EEG abnormalities, the absence of impairment of consciousness, and the response to L-dopa. One can only speculate as to the possibility of a paroxysmal release phenomenon from striatal control on the analogy of the pathological and physiological evidence involving the caudate nucleus and putamen in chorea and dystonia and the pallidum in athetosis. This may be related to insufficient maturation of some of these control pathways with simultaneous activation of other basal ganglia circuits concerned with movement, as indicated by the young age of onset and the later disappearance of the attacks in some (Kertesz, 1967; Homan et al. 1980).

The results of this clinical study show that PKC is not rare in Korea and has characteristic clinical features which are comparable to those in the literature. The prognosis of PKC is excellent.
REFERENCES


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