CASE REPORT

Paramyotonia Congenita without Cold Paralysis: A Case Report

Yoshio Izumi, Yasuo Fukuuchi¹, Atsuo Koto¹ and Shinji Nakajima²

Department of Neurology, School of Medicine, Tokai University, Kanagawa, ¹Department of Neurology, School of Medicine, Keio University, Tokyo and ²First Department of Internal Medicine, National Defence Medical College, Saitama, Japan

(Received for publication on February 2, 1994)

Abstract. A 27-year-old woman with paramyotonia congenita was reported. She began to suffer from myotonia since infancy. Myotonia was aggravated by cold, but with intense cooling myotonia did not change to flaccid paralysis. Four generations of her family showed the same symptoms that suggested autosomal dominant inheritance. Neurological examinations revealed no impairments of mental function, cranial nerves and sensory system. Muscular atrophy or hypertrophy was not observed. Percussion myotonia of the tongue and thenar muscles could be elicited at room temperature. Myotonia was aggravated by cold. An oral intake of potassium chloride (7 grammes) did not provoke any muscle weakness or flaccid paralysis. Routine laboratory data and findings of head CT scan, cerebrospinal fluid and electroencephalogram were normal. The nosological distinction between paramyotonia congenita and hyperkalemic periodic paralysis has been debated since 1956. Paralysis induced by cold is thought to be a feature of paramyotonia congenita, thus raising a possible relationships to hyperkalemic periodic paralysis. In our case cold paralysis never occurred spontaneously and could not be provoked by immersion in ice water or by potassium loading. This finding confirms the existence of paramyotonia congenita without cold paralysis and may provide a nosological distinction between paramyotonia congenita and hyperkalemic periodic paralysis. (Keio J Med 43 (2): 94–97, June 1994)

Key words: Eulenburg's syndrome, myotonic syndrome, myotonia, periodic paralysis, adynamia episodica hereditaria

Introduction

The classification of myotonic disorders has caused controversy. It is generally accepted that paramyotonia congenita (PMC) is an independent disease, which is not related to myotonia congenita (Thomsen's disease) or myotonic dystrophy. The most typical clinical presentation in PMC is the precipitation of active myotonia by exposure to cold temperature and prolonged attacks of muscle stiffness and weakness.

However, Gamstorp¹ described a similar disease which she named adynamia episodica hereditaria or as it came to be called hyperkalemic periodic paralysis (HPP). HPP may be associated with myotonia and in patients with HPP paralysis may occur with cold exposure. The nosological distinction between the two entities has been debated.

Recently besides clinical symptoms, the neurophysiological examination became important for the differential

diagnosis and has been proposed as specific to each entity.² Ricker *et al*³ reported the different effectiveness of tocainide and hydrochlorothiazide in PMC with HPP.

Paralysis induced by cold is thought to be an aspect of PMC that revealed relationships to HPP. We have observed a case of PMC whose myotonia did not change to paralysis in the cold. We discussed that the patient offer the suggestion of nosological distinction between PMC and HPP.

Case Report

The patient was a 27-year-old woman who began to suffer from a muscular dysfunction since infancy. Under the influence of cold, her face and then her hands would go into stiffness. The stiffness was painless but was associated with a feeling of tension. She found that walking against a cold would cause her face to feel stiff and tight, and she could not open her eyes. When she

泉 義雄, 福内靖男, 厚東篤生, 中島伸二

Reprint requests to: Dr Yoshio Izumi, Department of Neurology, School of Medicine, Tokai University, Boseidai, Isehara, Kanagawa 259-11, Japan

took ice cream, she could barely move the tongue and could not eat it. She had to rest the tongue for several minutes before it could be moved with strength. She stated that her tongue felt stiff but not weak. She did not experience any episodic weakness or paralysis in the cold or without any relation to cold in the past. These symptoms were stable and she did not feel any progression. She denied any influence on her symptoms by emotions, hunger, fatigue, pregnancy and menses. There was no prior history of endocrine or cardiac disorder, cataract or any other serious illness. Her father, daughter, brother, paternal grand mother, uncles, aunts and niece all had similar episodes (Fig 1).

General physical examination revealed a moderately nourished woman. There was no evidence of frontal baldness or struma. Neurological examinations revealed no impairment of mental function and cranial nerves. The sternocleidomastoid and trapezius muscles showed a normal bulk and strength. No muscle weakness, atrophy or fasciculation could be demonstrated in the extremities. Her gait was normal and she had no difficulty of standing or hopping on either foot unaided. Percussion myotonia of the tongue could be demonstrated at room temperature. Percussion myotonia of the thenar eminence was demonstrated. There were no other apparent abnormalities of muscle tone. Deep tendon and superficial reflexes were active and symmetrical. Plantar responses were flexor. No cerebellar incoordination or sensory deficit could be demonstrated.

Laboratory findings revealed no abnormalities in serum and urinary electrolytes, hormones and other blood chemistry. Head CT scan, electroencephalogram, cerebrospinal fluid and ECG were normal. An oral intake of potassium chloride (7 gramme load) and glucose (200 gramme load) did not provoke any muscle weakness or flaccid paralysis.

Fig 1 Pedigree of the family. Dark circles (women) and squares (men) represent clinically affected patients. Index case is marked by an arrow.

Five minutes after immersion of the face into cold water, marked myotonia of the orbicularis oculi developed following closure of the eyelid (Fig 2). The patient had to rest her eyes for several minutes at room temperature before the lids could be opened enough to see.

In the EMG (Fig 3) at a room temperature, insertion of the needle electrode into the thenar muscle or extensor group of forearm muscles was accompanied by typical "dive-bomber" burst of activity lasting several seconds. During the period after cooling, the amplitude was increased and the duration was elongated. There was no electrical silence.

Muscle biopsy from the biceps brachii muscle showed

Fig 2 Effect of cold on facial muscles. Five minutes after immersion of the face into cold water, marked myotonia of the orbicularis oculi developed following closure of the eyelid.

Fig 3 Electromyographic demonstration. In the EMG at a room temperature, insertion of the needle electrode into the thenar muscle was accompanied by typical "dive-bomber" burst of activity lasting several seconds. During the period after cooling, the amplitude was increased and the duration was elongated. There was no electrical silence.

Fig 4 Biopsy from the biceps brachii muscle. It showed a variation of muscle fiber diameters and internal nuclei, suggesting slight myopathic changes. Stain with hematoxylin and eosin.

Fig 5 Biopsy from the biceps brachii muscle. In the stain with NADH, there were no specific findings like ringed fibers, sarcoplasmic masses, type I fiber atrophy, moth-eaten fibers which are characteristic to myotonic dystrophy.

a variation of muscle fiber diameters and internal nuclei, suggesting slight myopathic changes (Fig 4). In the stain with NADH, there were no specific findings like ringed fibers, sarcoplasmic masses, type I fiber atrophy, moth-eaten fibers which are characteristic to myotonic dystrophy (Fig 5).

Discussion

The patient has noted myotonia since infancy, particularly when she was exposed to a cold environment. With intense cooling myotonia did not change to flaccid weakness. Nine members from four generations of her family showed the same symptoms, suggesting autosomal dominant inheritance. She was diagnosed as a typical

case of paramyotonia congenita (PMC).

Myotonia is characterized by delayed muscle relaxation after contraction and the phenomenon is usually best demonstrated in the tongue and thenar eminence, however most striated muscle shows this phenomenon. Clinically myotonia is a prominent sign in three hereditary muscular disorders: Myotonia congenita (Thomsen's disease), myotonic dystrophy and paramyotonia congenita (PMC).

Thomsen's disease is a rare hereditary disorder characterized by localized or generalized myotonia. Typically the disorder is present from birth and there is stiffness and difficulty in relaxation of the entire voluntary musculature. Generalized muscular hypertrophy is common.

Myotonic dystrophy is the best known syndrome in which myotonia is a prominent sign. The disorder usually first appears in the middle age and is slowly progressive. The facial appearance is characteristic. Wasting of the masseters, temporal muscles and sternocleidomastoids is almost invariable and in the extremities there is distal weakness and wasting. Frontal baldness, cataract and endocrine abnormalities are common associated findings.

PMC is a rare hereditary disorder originally described by Eulenburg in 1886.⁴ It is inherited as an autosomal dominant trait and characterized clinically by myotonia and weakness which are provoked or aggravated by cold. The cold-induced myotonia is paradoxical insofar as it is aggravated by repeated muscle contraction.⁵ During deep cooling the myotonia disappears, giving way to flaccid paralysis, which may outlast rewarming by many hours. The condition is nonprogressive. The patient actually improves with the progress of adulthood showing gradual adaptability. Muscle wasting is not present and muscular hypertrophy generally does not occur. Endocrine imbalance is absent.

It is generally accepted that myotonic dystrophy is a disease entity which is separate from other myotonic disease. The separation of PMC from Thomsen's disease has become more accepted recently. Many people believe that any kind of myotonia worsens with exposure to cold. However, profound and long-lasting muscular weakness evoked by local cooling is not observed in Thomsen's disease. An impressive dependency upon cold, the so-called paradoxical myotonia and the subsequent cold paralysis make, among other signs, a quite different disease entity from Thomsen's disease.⁵

In 1956, Gamstorp¹ described a familial disease with episodic attacks of muscular weakness associated with elevated serum potassium, which she named adynamia episodica hereditaria, or as it came to be called, hyperkalemic periodic paralysis (HPP). The nosological distinction between PMC and HPP has been debated. PMC and HPP have many features in common.⁶ In HPP

Table I Chronological List of Paramyotonia Congenita without Cold Paralysis Reported in the Literature

Year	Authors	No. of Cases	Generations	Genetics
1889	Martius and Hanseemann	11	4	dominant
1891	Friis			
1895	Hlawaczek	1	1	one case
1898	Funcke	1	1	one case
1917	Hübner	15	4	dominant
1930	Serog	9	4	dominant
1936	Schott	5	2	dominant
1938	Smitt			dominant
1955	De Jong	37	5	dominant
1966	Becker			dominant
1977	Brüngger	26	4	dominant
1985	present case	9	4	dominant

the occurrence of myotonia has shown considerable variation, ranging from absent, slight and moderate to severe.⁷ The patient reported by French and Kilpatrick⁸ and episodic attacks of flaccid paresis which were chiefly precipitated by exercise followed by rest, and the oral administration of 5 grammes of potassium produced generalized weakness. The patients described by Drager *et al*⁹ and Baxter and Dyck¹⁰ had episodes of flaccid paresis, but the effect of increasing the potassium level was not investigated.

What seems to be characteristic of PMC is the weakness brought out by cold and tending to outlast the myotonia.^{10–12} It is this aspect of PMC that reveals its relationships to HPP. However in the literature cases of Martius and Hanseemann, Serong, Schott, Smitt and Brüngger etc. did not have any evidence of paresis (Table I).^{13–17} In our case cold paralysis never occurred spontaneously and could not be provoked by immersion in ice water or by potassium loading. This finding confirms the existence of PMC without cold paralysis. According to Becker,¹³ “There is ample evidence to assure that Thomsen’s disease, PMC and adynamia episodica hereditaria are independent genetic types different from each other.” Our patient resembled the cases of PMC without cold paralysis described by De Jong.^{16,17} These cases

may provide nosological distinction between PMC, HPP and other variant of myotonia syndrome.

References

1. Gamstorp I: Adynamia episodica hereditaria. *Acta Paediatr Suppl* 1956, 108: 1–126
2. de Silva SM, Kuncel RW, Griffin JW, Cornblath DR, Chavoustie S: Paramyotonia congenita or hyperkalemic periodic paralysis? Clinical and electrophysiological features of each entity in one family. *Muscle Nerve* 1990, 13: 21–26
3. Ricker K, Böhlen R, Rohkamm R: Different effectiveness of tocainide and hydrochlorothiazide in paramyotonia congenita with hyperkalemic episodic paralysis. *Neurology* 1983, 33: 1615–1618
4. Eulenburg A: Über eine familiäre, durch 6 Generationen verfolgbare Form congenitaler Paramyotonie. *Neurol Centralbl* 1886, 5: 265–272
5. Magee KR: A study of paramyotonia congenita. *Arch Neurol* 1963, 8: 461–470
6. Layzer RB, Lovelace RE, Rowland LP: Hyperkalemic periodic paralysis. *Arch Neurol* 1967, 16: 455–472
7. Thrush DC, Morris CJ, Salmon MV: Paramyotonia congenita: a clinical, histochemical and pathological study. *Brain* 1972, 95: 537–552
8. French EB, Kilpatrick R: A variety of paramyotonia congenita. *J Neurol Neurosurg Psychiatr* 1957, 20: 40–46
9. Drager GA, Hammill JF, Shy GM: Paramyotonia congenita. *Arch Neurol Psychiatr* 1958, 80: 1–9
10. Baxter DW, Dyck PJ: Paramyotonia congenita. *Can Med Assoc J* 1961, 85: 113–118
11. Lundberg PO, Stålberg E, Thiele B: Paralysis periodica paramyotonica. A clinical and neurophysiological study. *J Neurol Sci* 1974, 21: 309–321
12. Subramony SH, Wee AS, Mishra SK: Lack of cold sensitivity in hyperkalemic periodic paralysis. *Muscle Nerve* 1986, 9: 700–703
13. Becker PE, ed: Paramyotonia congenita (Eulenburg). Stuttgart Georg Thieme Verlag, 1970
14. Becker PE: Genetic approaches to the nosology of muscular disease: myotonias and similar diseases. *Birth Defects* 1971, 7: 52–62
15. Brüngger U, Kaeser HE: Paramyotonia congenita without cold paralysis and myotonia levior. A genetic and clinical study. *Eur Neurol* 1977, 15: 2–4
16. De Jong JGY, ed: Dystrophia Myotonica, Paramyotonia and Myotonia Congenita. Van Gorcum Assen, 1955
17. Jong JG de, Slooff JL, Eerden AA vander: A family with paramyotonia congenita with the report of an autopsy. *Acta Neurol Scand* 1973, 49: 480–494