CASE REPORT

Myotonia Congenita (Thomsen's Disease) Report of Five Cases in a Family

HARDIONO D. PUSPONEGORO, JAHJA ZACHARIA and JIMMY PASSAT

(From the Department of Child Health, Medical Faculty University of Indonesia, Cipto Mangunkusumo Hospital, Jakarta)

Abstract

This report describes 5 out of 8 siblings who were the first cases of myotonia congenita diagnosed in our department. The parents were first cousins. Neither the parents, nor the other family members have myotonia. The affected siblings 4 boys and 1 girl, all showed a very typical myotonia especially after prolonged rest, and it could be worked off with continuining activity. They had a muscular looking body or a herculean proportion. The diagnosis were based upon family history, clinical findings of percussion myotonia, had grip myotania, prominent muscular hypertrophy and confirmed by electromyographic examination revealing myotonic discharges. Since there were some functional impairments, these patients were treated with diphenylhydantoin and then with quinine sulphate, with good results.

The patients related parents were much likely to be heterozygous for the same harmful recessive genes, because they had common ancestor. The role of marriage counseling is important in this kind of inherited disease, to prevent the occurrence of this inherited disorder in the next generations.

Received November 8, 1990

Introduction

Myotonia is a clinical phenomenon of delayed relaxation of the skeletal muscle after contraction, both voluntarily following a stimulus. This phenomenon occurs in a number of disorders. Myotonia congenita is a rare inherited disorder of the skeletal muscle, it begins in early life and is characterized by myotonia and muscular hypertrophy (Adams and Victor, 1989).

In 1876, this congenital muscle disorder was first reported by a patient suffering from this disorder, that was Dr. Julius Thomsen himself. He described it as ataxia muscularis. In 1881 Strumpell assigned the name of myotonia congenita to this disease and later on in 1883 Westphall referred it as Thomsen's disease.

The purpose of this report is to demonstrate the clinical features of myotonia and as a reminder that a proper diagnosis and prompt approach could be very beneficial to patient with congenital muscle disorder. The functional impairments could be treated with good result by the administration of drugs.

Report of cases

N, a 10 years old girl, was brought by her mother to the outpatient clinic of the Department of Child Health, Dr. Cipto Mangunkusumo Hospital on 8th August, 1990. The chief complaints were muscle stiffness and difficulties in moving, especially after a period of inactivity. She had been suffering from these complaints 3 years. One year later her body became very muscled. Despite her large muscles she felt weak, and easily fatiqued. For example after a nap, sitting, sleeping or even after a fall, she found difficulty to resume an activity. Her gait was unusually stiff.

She was the 7th of siblings. Since the age of 7 years, 3 of her older brothers and the youngest family brother suffered from the same disease. The family pedigree is seen in figure 1.

On physical examination the girl was alert, with a peculiar stiff swadding gait, and a muscular looking body especially in her lower extremitas. The vital signs and the organs were within normal limits. The extremities showed a moderate hypertrophy of the muscles with a delayed relaxation of stretch reflexes (Figure 3A–B and 4). Most of the muscles had an increased tone.

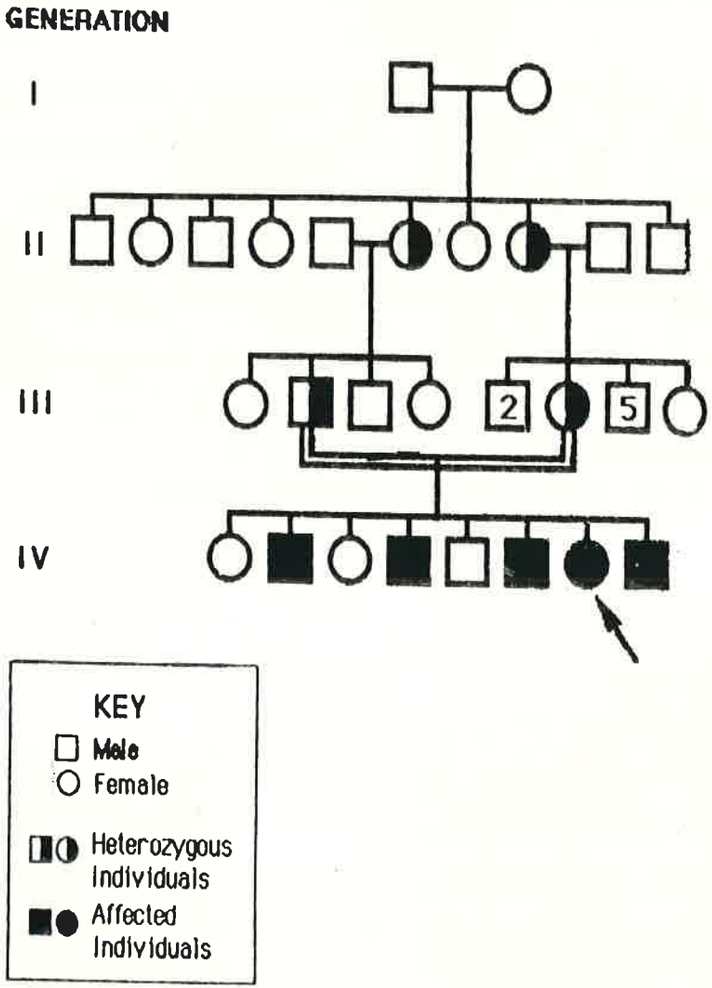


Figure 1 : *Pedigree of patient, showing recessive inheritance. Our first patient was the propositus of this pedigree.*

Based on the family history and clinical findings the diagnosis of myotonia congenita was made. To confirm the diagnosis an EMG was done. On muscle percussion there were myotonic dischargers and the characteristic dive bomber sound. Voluntary activity revealed a myotonic disorder, and the diagnosis of myotonia was disclosed. Electromyographic examination were also done on her brothers showing the characteristic myotonic discharges. Other laboratory findings were within normal limits. Except in one patient the creatine kinase levels were all elevated. **(Table 1)**

These patient were treated with diphenylhydantoin followed by quinine sulphate as an alternative. The response was very satisfying. The patient could then run around the house, chasing her brother.

Table 1 : *Clinical Features, EMG and Laboratory Findings in 5 Siblings Suffering Recessively Inherited Myotonia Congenita*

Patient	L	S	N	N	M
Sibling Number	2	4	6	7	8
Age (yrs)	20	16	12	10	8
Sex	M	M	M	F	M
Myotonia	+	+	+	+	+
Muscle Hypertrophy	+ + +	+ + +	+ +	+ +	+
EMG (myotonia)	+	+	+	+	+
CK (10-80 V/L)	296	178	233	45	93
Na (135-147 mEq/L)	142	144	136	140	140
K (3.5-5.5 mEq/L)	3.7	4.7	3.4	4.1	3.9
Cl (100-106 mEq/L	104	107	101	103	107

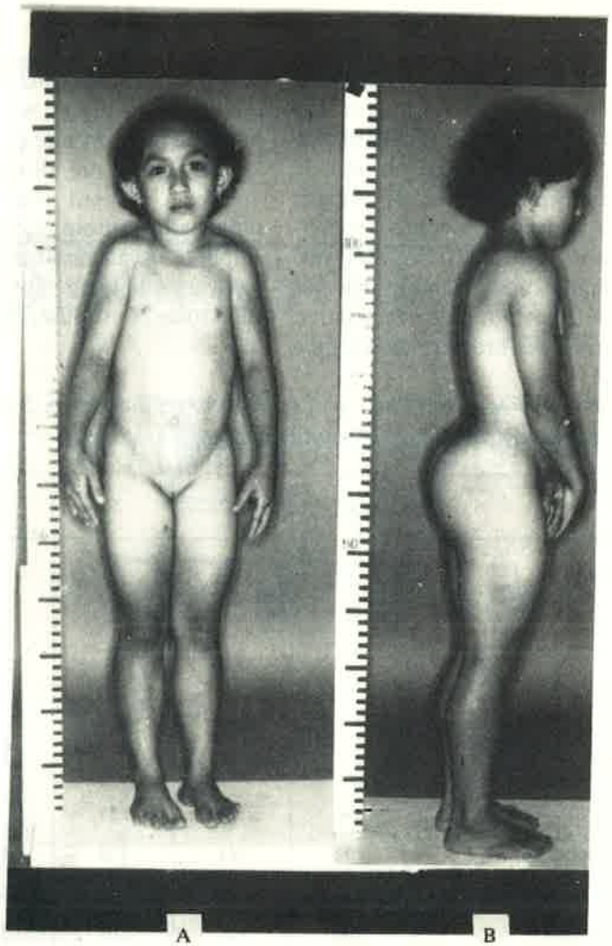
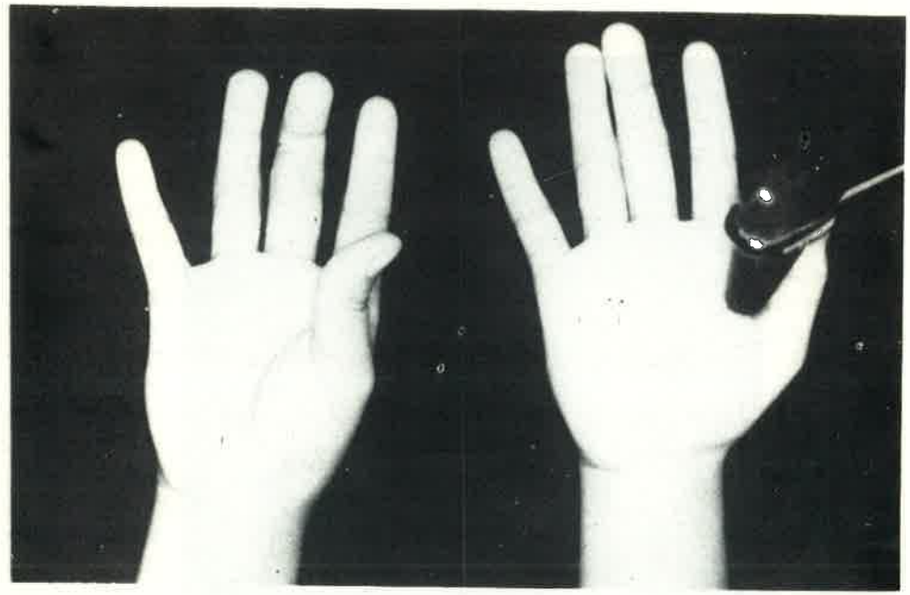


Figure 2 A - B *Muscle hypertrophy especially in the lower extremities and ncreased muscle tone in the trapezius muscles elevating the shoulders.*

The working diagnosis was congenital muscle disorder. The patient was then consulted to the pediatric neurology clinic. On further examination the patient showed percussion myotonia and hand grip myotonia. Later on all her brothers were examined and showed severe muscle hypertrophy in her elder brothers and myotonia in all of them.



A B

Figure 3 A-B : *Percussion myotonia on the thenar muscle.*

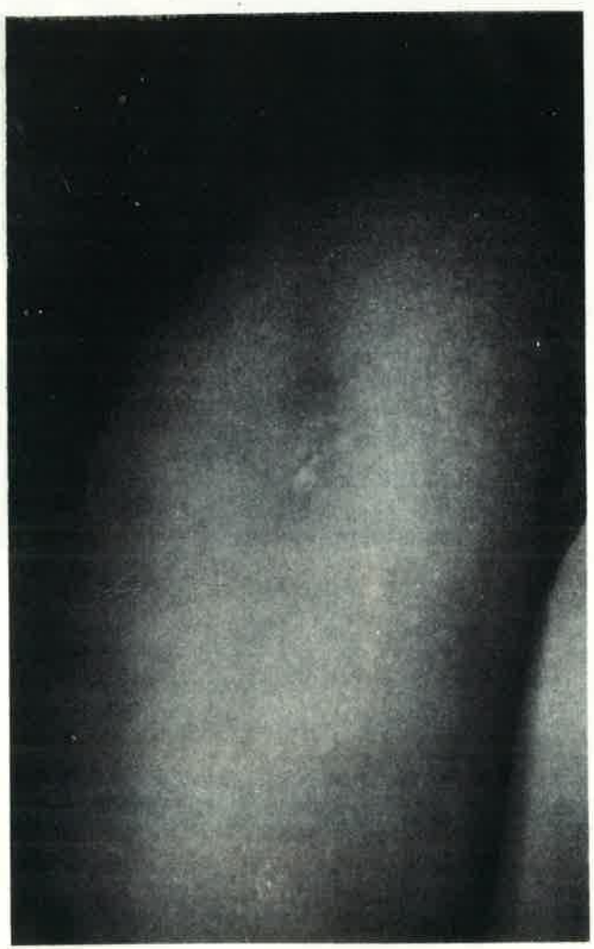


Figure 4 : *Percussion myotonia on the deltoid.*

Discussion

These rare cases of myotonia congenita might be the first cases recognized in our hospital. Before this patient came to Dr. Cipto Mangunkusumo General Hospital she had been examined by a doctor in another hospital and one of her brothers had also been examined in another department of our hospital. She was told that the disorder could not be treated. Her brother was diagnosed as having polyneuropathy, though he had been examined electromyographically. The first kind of information may arise from a wrong diagnosis or perception that every congenital disorder of the muscle can not be treated and are progressive, like progressive muscular dystrophy. The second improrer diagnosis was made because the family history was not taken.

The best approach to patients with familial, chronic, or disabling disorders is to have detailed family history and clinical features, so that they can receive the most cost effective treatment including prevention of further disabilities and rehabilitation thus leading a better life. A wrong information may bring some hazards to person's perception of his/her own future. Fortunately after a period of helplessness, the mother brought the child to the outpatient clinic of Child Health Department, Cipto Mangunkusumo General Hospital. The result was that our first patient and her brothers could benefit from a very simple and low cost treatment.

The diagnosis was based on clinical findings and electromyographic picture of myotonia. Serum enzymes are usually normal but may also be elevated (Huttenlocher, 1987; Adams and Victor, 1989). Neither the parents, nor other family members had myotonia. The patient and her affected brothers all showed a very typical myotonia especially after prolonged rest, and could be worked off with contuining activity.

They had a very muscular looking body also called the herculean proportion, especially in the older patients. Muscle hypertrophy is one the two unique features of myotonia congenita, described by Erb in 1886 i.e. muscular hypertrophy and muscular hyperexcitability. One of the possibilities that caused this muscular hypertrophy is prolonged contraction of the muscle fibers. On EMG examination the tension in contracting muscle fibers is slow to diminish, due to persistence of very fine electrical potentials. Their activity continues after the volley of nerve impulses that initiated the contraction (Adams and Victor, 1989). Consequently in older patients, the muscle hypertrophy is more prominent.

Reduction in chloride and potassium conductance has been found in myotonia congenita. Substances like quinine, procainamide and calcium, known as acting on the sarcelomma, lessen the duration of myotonia. Myotonia can be induced by a drug (diazocholesterol) presumably by altering the membrane resistance and decreasing chloride conductance. These suggest that myotonia depends on some basic alteration of the sarcolemma itself (Adams and Victors, 1989).

In this patient muscle biopsy was not done, as no abnormality other than enlargement of muscle fibers could be revealed. This minimal change occurs only in hypertrophied muscles (Adams and Victor, 1989). Absence of group IIB fibers

on strining for myosin ATPase reaction has been noted (Crews et al., 1976). There are no changes in the peripheral or central nervous system.

The cause of this disease is of genetic origin. From the studies of Becker in 1971 and 1977, myotonia congenita could be divided into two forms. The first is the autosomal dominant inherited type and the second the autosomal recessive type (Harper, 1988). The differing features of those types can be seen in table 2.

Table 2 : *Features of Two Types of Myotonia Congenita*

Type 1 (Dominant)	Type 2 (Recessive)
° Early Onset; Infancy	° Later Onset; Childhood
° Male - Female	° Male Predominance (3:1)
° Cold ---- Worse	° No other Factor
° No/Slight Hypertrophy	° Prominent Muscle Hypertrophy
° Slow Eye Opening	° Severe Myotonia
° Stiff Legs	

From family history/pedigree and the clinical findings, it was quite obvious that this patient and the siblings suffered from the recessively inherited form of myotonia congenita. The patient's related parents were much likely to be heterozygous for the same harmful recessive genes, because they had a common ancestor (Holmes, 1987).

Although the disorder affecting this family was not very disabling, the parents had some guilty feeling. They had this feeling because of their consanguinous marriage so that 5 (of 8) of their children had to suffer from this disorder. Nevertheless they were rather relieved to know that the disorder was benign and might improve with age (Huttenlocher, 1987). Not less important is that the functional impairment, could be treated by the administration of quinine sulphate (300-600 mg), procainamide (250-500 mg) and phenytoin (100 mg) tid (Adams and Victor, 1989).

The role of marriage counselling is important in this kind of inherited disease to prevent the occurrence of this disorder in the next generations.

REFERENCES

1. ADAMS, R.D., VICTOR, M. : Disorders of muscle characterized by cramp, spasm, pain, and localized masses; in Adams, Victor, Principles of neurology; 4th ed., pp. 1172-1174 (McGraw-Hill, New York, Singapore 1989).
2. HARPER, P.S. : The myotonic disorders; in Walton, Disorders of voluntary muscle; 5th ed., pp. 581-582 (Churchill Livingstone, Edinburgh, 1988).
3. HOLMES, L.B. : Prenatal factors in diseases of children. Genetic factors; in Behrman, Vaughn, Nelson Textbook of pediatrics; 13th ed., pp. 244 (Saunders 1987).
4. HUTTENLOCHER, P.R. : Neuromuscular diseases; in Behrman, Vaughn, Nelson Textbook of pediatrics; 13th ed., pp. 1339 (Saunders 1987).