Myotonia Congenita (Thomsen's Disease)

(A Case Report)

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Myotonia congenita was first described by Asmus Julius Thomsen in 1876, the disease having appeared in seven generations of his family including himself. The condition is heredofamilial, becoming apparent very early in life and characterized by a tendency to a delayed relaxation of voluntary muscular contractions or myotonia, often accompanied by a bulky musculature. This rare disorder is transmitted by autosomal genes, usually as a dominant characteristic, although recessive inheritance has been reported.[1-3]

No metabolic changes have been demonstrated in the tissues and the involuntary muscles are spared. The myotonia is usually accentuated by cold and tends to improve after repetitive voluntary movement. The cause of the contractions may be voluntary effort, direct percussion, electrical stimulation or emotional reactions. Myotonia may be found in disorders such as dystrophia myotonica although there has been a great deal of controversy as to whether these should be regarded as separate entities.

The purpose of this paper is to describe a case of myotonia congenita as a first report from Hacettepe Medical Center.

Case Report

M. B., a nine-year-old boy, was seen in the Outpatient Department of Hacettepe Children's Hospital because of difficulty in climbing stairs. For at least four years the patient's mother had noted some stiffness in his relaxing his grasp and initiating movements. The stiffness could be «worked out» with repetitive movement. His gait was rigid and much difficulty was encountered in climbing stairs. He could not relax his grip suddenly after shaking hands. He was

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unable to stand without holding onto something. Initiation of movements such as writing, eating and walking was always slow. He would then «loosen up» and his performance would improve. There was no myotonia following exposure to cold. The family history was negative for neuromuscular disease.

Physical Examination : The general physical, respiratory, cardiovascular and abdominal examinations revealed no abnormalities. Neurologic examination revealed the pupils to be normal to direct examination and there were no lenticular opacities. The eyegrounds were normal. Facial muscles were normal and neck flexors and extensors were strong. There was no fasciculation. There was voluntary myotonia on gripping which disappeared after repetitive grasping. No weakness or wasting was noted and there was widespread muscular hypertrophy giving him a Herculean appearance. On palpation the muscles were firm. Muscular strength was moderately good and equal, but not in keeping with the size of the muscles. There was no rigidity on passive movements, no sensory abnormalities, no testicular atrophy and no baldness.

Laboratory examination : Serum creatine phosphokinase was 17 u (within normal limits for our laboratory). Electromyographic examination revealed typical myotonic bursts and fibrillations.[4] A muscle biopsy was obtained from the gastrocnemius muscle and histologically the muscle fibers showed some hypertrophy.

After the diagnosis was established, the patient was placed on quinine and followed in the Outpatient Department.

Discussion

The early stage of dystrophia myotonica may be diagnosed as Thomsen's disease.[5] Bell has outlined the main differences between these two diseases (Table 1). The absence of atrophy in our case should rule out dystrophia myotonica, although some cases indicate that children of parents with dystrophia myotonica develop myotonic symptoms at an early age and that myotonia may appear before any wasting is observed.[5] Myotonia congenita and cases of dystrophia myotonica may occur in the same family, for example, the Thomsen family.[5,6] Cases of transition from one syndrome to the other are not uncommon.[5]

This condition rarely causes severe incapacity and does not progress. Wolf has shown that quinine will abolish the symtoms.[2] The

effect of this drug is entirely symptomatic. It is claimed that procainamid will also reduce the voluntary contraction myotonia, but not the percussion myotonia. Other drugs tried are cortisone, potassium binding ion exchange and serin.

The so-called hypertrophia musculorum vera might simulate Thomsen's disease though myotonïa is not present in this condition. The effect of quinine in reducing the myotonia is of value in the diagnosis.

Summary

A case of myotonia congenita is presented. The picture was that of myotonia congenita with widespread muscle hypertrophy and widespread myotonia. There was no wasting or weakness and no gonadal atrophy. There were no lenticular opacities, the skull was normal and the E.M.G. showed typical myotonic bursts.

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TABLE 1

BELL'S COMPARISON BETWEEN THOMSEN'S DISEASE AND DYSTROPHIA MYOTONICA [5]

	Thomsen's Disease	Dystrophia Myotonica
Age of onset	Usually congenital	75 per cent after 15 years
Myotonia	Widespread	Usually over restricted areas
Muscular signs	Hypertrophy common	Marked weakness and wasting in characteristic areas
Cataract	No association	Frequent association
Baldness	Not a characteristic	Characteristic
Testicular atrophy	Not a characteristic	To be expected
Tendon reflexes	Usually normal	Usually lost early in disease
Mental condition	Associated psychosis	Deterioration common
Genetic basis	Dominant, suppression rare	Dominant, suppression often demonstrable

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