MYOTONIC DYSTROPHY AND HYPERTHYROIDISM — A CASE REPORT

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SYNOPSIS

The association of dystrophia myotonica and thyrotoxicosis is very rare. This is the first reported case in Singapore. We describe a family with familial thyroid disorders occurring in association with dystrophia myotonica. The onset of thyrotoxicosis in the patient described resulted in severe muscle weakness which recovered when euthyroid status was achieved.

CASE REPORT

A 22 year old Chinese girl presented with a history of severe weight loss of 16 pounds, progressive difficulty in swallowing liquids and solids and a weak voice over a period of 3 months. She had also noticed increased sweating and palpitations for the past year. Her appetite has remained normal. She was not aware of any previous illness before.

She is second in a family of five children. Her father aged 58, brother aged 25 and younger sister aged 18, were diagnosed to have dystrophia myotonica 3 paternal uncles afflicted with the disease has since passed away (Fig. I). Her father had a thyroid nodule removed 10 years ago. Her younger sister was treated for thyrotoxicosis several years ago.

Clinical examination revealed an emaciated young woman. She weighed 35 Kg. Her thyroid was diffusely enlarged, no bruits were heard over the thyroid gland. Her blood pressure was 90/70 mmHg and pulse rate was 108/min. The heart was not enlarged and a soft systolic murmur was heard over the left sternal edge. No masses were felt in the abdomen.

Examination of the nervous system revealed severe bilateral facial weakness and palatal weakness. There was generalised muscle weakness which was predominantly proximal. She had neck flexor weakness and a waddling gait was noted. She was completely areflexic.

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Percussion myotonia was demonstrated in the thenar muscles, extensor digitorum communis and tongue. Slit lamp examination revealed no cataracts. IV Edrophonium 10 mg was given with no positive results. Investigations done, confirmed a hyperthyroid state. T3 Resin uptake 112% (N 72-118%), T4 22.7 ug/dl (N 5.6-11.5 ug/dl) FTI 25.4 (N 4-13.5). A persistent hyperkalemia was noted during the first 3 days of admission, serum potassium being 6, 6.5 6.4 meq/L. The serum sodium and chlorides were normal. The serum creatinine phosphokinase activity was normal.

Electromyography showed frequent myotonic discharges. The motor unit potentials were of hormal amplitude and duration. Polyphasic units were not exceptionally profuse. The inferference pattern was complete. Muscle biopsy (light microscopy) showed atrophy of skeletal muscle fibres with increased internal nuclei and nuclear chains. Few basophilic regenerating fibres were also seen. No vacuolation was noted. There was normal fibre type distribution.

Treatment with Neomercazole 15 mg tid. p.o. was started immediately on admission to hospital. Clinical euthyroid status was achieved within six weeks. Muscle power improved but residual weakness was still present. Percussion myotonia persisted. She was biochemically euthyroid in 4/52. Her weight 6 months later was 45 Kg.

DISCUSSION

Dystrophia myotonica is a diffuse systemic disorder in which myotonia and muscle atrophy may be accompanied by cataracts, frontal baldness (in males), gonadal atrophy, heart disease, impaired pulmonary ventilation, bone changes, mental defects or dementia, abnormalities in serum immunoglobulins and mild endocrine abnormalities. It is inherited as an autosomal dominant trait and the pattern of manifestation and severity of disease shows great variation within families.

Endocrine abnormalities include pituitary disorders (1) disorders in carbohydrate metabolism (2) and rarely thyroid disorders (3, 4, 5) has been described. With particular reference to this article, thyrotoxicosis in association with myotonic dystrophy is very rare. Four other cases have been described before (3, 4, 5). Our patient has additional unusual features. Two out of three family members affected with myotonic dystrophy had concomitant thyroid disorders.

Her younger sister (age 18) also had thyrotoxicosis and had an acute exacerbation of weakness followed by some degree of residual weakness.

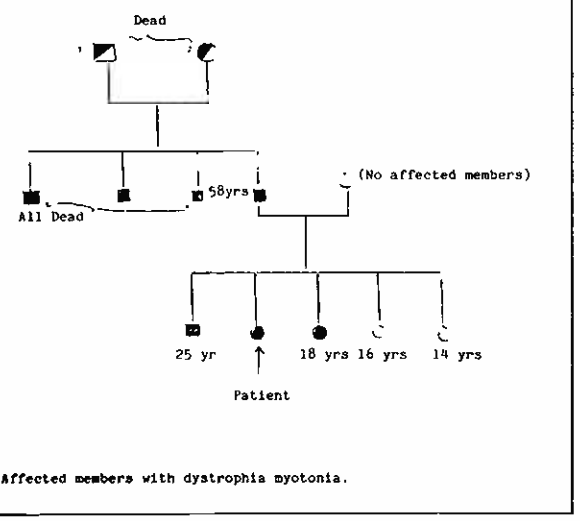
Her father presented with a gradual progressive distal weakness of the limbs and was found to have a coincidental thyroid nodule. His thyroid function was normal. The nodule was removed and histology showed a nontoxic adenoma.

The patient's elder brother has all of the above mentioned features of dystrophia myotonica. He has demonstrable weakness of the distal muscle groups. His thyroid function is normal.

At the time of publication, the HLA status of the affected members of this family was not yet known. The autosomal dominant trait has affected 50% of family members in 2 generations (see Fig I). It can be postulated that this family also carries a specific HLA typing linked to familial thyroid disorders. In previous reports (3, 4, 5) HLA typing of patients were not carried out.

FIG. I

PATIENT'S FAMILY TREE



Our patient was apparently well prior to the onset of thyrotoxicosis, in spite of having features of dystrophia myotonica. The sudden development of severe muscle weakness, dysphagia and dysarthria is postulated to be due mainly to excess thyroid hormone. However on regaining euthyroid status, residual muscle weakness could still be demonstrated.

Severe muscle weakness in thyrotoxicosis was first described by Graves & Von Basedow. Serum levels of creatinine phosphokinase are usually normal (6). The term 'acute thyrotoxic myopathy' (7) has been used to describe the clinical picture of a sudden onset of dysphagia, dysphonia and generalised weakness in a thyrotoxic patient. We feel that although above mentioned events has occurred in our patient, the severity of weakness was aggravated by the concomitant myotonia.

The muscle biopsy in this patient has features of myotonic dystrophy viz. increase in internal nuclei and nuclear chains. Vacuolation which is usually present in thyrotoxic periodic paralysis (8) and was absent. Light microsocpy in thyrotoxic myopathy is usually nonspecific (9).

The danger of thyrotoxicosis occurring in a patient with dystrophia myotonica cannot be further stressed as in 1 case report (3) death from aspiration pneumonia occurred during the acute phase of the illness.

This patient had persistent hyperkalemia initially. Hyperkalemia per se gives rise to transient weakness which reverses when the electrolyte imbalance is corrected. It is unlikely that the patient had a concomitant hyperkalemic periodic paralysis (Gamstop) (10) as when the electrolyte imbalance was corrected there was no improvement in muscle weakness.

The defect in myotonic dystrophy is postulated to be due to an abnormality of the muscle fibre itself, for it persists after section or blocking of the motor nerve and after curarisation (11).

CONCLUSION

In this brief communication we note that thyroid hor-

mone and hyperkalemia has an important role in muscle function. However the actual mechanism of action is still unknown and subject to postulation.

It is of paramount importance to ascertain the thyroid status in all patients with dystrophia myotonica as the presence of both diseases can result in life threatening complications.

REFERENCES

1. Marshall J: Observations in endocrine function in myotonic dystrophy Brain 1959; 82:221-2.
2. Huff T A, Horton E S, Lebovitz H E: Abnormall insulin secretion in myotonic dystrophy. New Eng J Med 277: 837-41.
3. Peterson D M, Bounds J V, Karnes W E: Clinical observations on thyrotoxicosis coexisting with myotonic dystrophy. Mayo Clinic Proceedings 1976; 51:176-9.
4. Okuno T, Kosuke M, Koichi F et al: Myotonic dystrophy and hyperthyroidism. Neurology 1981; 31:91-3.
5. Berkman J M: Hyperthyroidism associated with myotonic dystrophy. Mayo Clinic Proceedings 1935; 10:273-5.
6. Graig F A, Ross G: Serum Creatine phosphakinase in thyroid disease. Metabolism 1963; 12:57-9.
7. Joasoo K, Murray I P C: Involvement of bulbar muscles in thyrotoxic myopathy. Aust Ann Med 1970; 19:338-40.
8. Puvanendran K, Naganathan N, Cheah J.S.: Thyrotoxic myopathy. Journal of Neurological Sciences 1979; 42: 441-51.
9. Engel A G: Electron microscopic observation in thyrotoxic and corticosteroid induced myopathies. Mayo Clinic Proceedings 1966; 41:785-95.
10. Gamstorp I: Adynamia episodica hereditaria and myotonia Acta Neurol Scan 1963; 39:41-58.
11. Denny Brown D, Nevin S: The phenomenon of myotonia Brain 1941; 64:1-18.
12. Walton J N: Disorders of voluntary muscles (4th edition) Little Brown Co, Boston, Mass. 1981; 508-514, 666-670.