A CASE OF DYSTROPHIA MYOTONICA

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This hereditary disorder, characterized by muscular dystrophy, myotonia and other dystrophic disturbances, especially cataract and gonadal atrophy, was first described by Deleage (1890). There is usually a history of cataract in members of preceding generations of the same family.

The incidence of the disease throughout the world is difficult to assess from the literature available. Waring et al. (1940) point out that the condition is more common than is generally believed and frequently unrecognized clinically. Their view is supported by Caughey and Brown (1950).

It is certainly a condition rarely found in the British Army. Figures supplied by the Statistical Branch, Army Medical Department, War Office, show that only four cases have been recorded for the years 1947 to 1951. Of these two were reported in 1948, one from the Middle East and the other in the U.K. A third case was reported from B.A.O.R. in 1950, and the fourth in 1951 from U.K. The following are the number of cases recorded by the Royal Air Force: 1943, 2; 1946, 1; 1947, 1.

CASE NOTES

P. S., a regular senior non-commissioned officer with 16½ years’ service, age 31 years, unmarried, was admitted to hospital for investigation as he had difficulty in carrying out rifle drill. Some six months prior to admission to hospital on 29th February, 1952, patient noticed weakness in both wrists, which had been getting progressively worse.

Past History.—Patient was the elder of two sons. His father died in 1929 from the effects of being gassed in the 1914-18 war. His mother died from heart trouble in 1934. His brother is alive and well. No history could be elicited of mental illness, blindness, or any nervous disorder in the family. When the patient was three years old he had a severe attack of measles, and had thereafter difficulty in seeing with his right eye. He has worn glasses as long as he can remember. He suffered from occasional colds. At the age of 21 he cut his head when diving into a swimming bath. There was no history of possible venereal infection. He did well at school and won a scholarship when 13 years old. On leaving school he joined the Army and has had a distinguished and satisfactory service record.

Present History.—He alleges that about five years ago his friends drew attention to his peculiar stamping type of gait. He himself paid little attention
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to it, as it did not worry him. Some six months prior to admission he noticed weakness in both wrists. This weakness became more noticeable, and he then observed his right forearm getting thinner. When doing rifle drill a week prior to admission, he discovered that he could not lift his rifle, or carry out routine drill with it. It was for this reason that he reported to his medical officer. He has shaved daily since he was 18 years old. He has a high moral code and has never been troubled by sex, having no interest in women or any desire to get married.

Examination

A thin, dark-haired man of asthenic build and sallow complexion, wearing glasses and appearing older than his years. He exhibits a myopathic facies (Fig. 1). His hair is thin and there is frontal baldness. He has, however, strong facial hair and normal male distribution over the rest of the body. Ptosis is present in both upper eyelids. There is wasting and weakness of the facial muscles and of the sterno-cleido-mastoids in the neck. The temporal muscles and masseters appear in good condition. There is loss of tone, together with wasting and weakness in the muscles of the forearms, which is more marked in the dorsiflexors of the wrists. The ulnar and hypothenar eminences are wasted, and all the muscles of the hand are wasted and weak. The handclasp is definitely myotonic. In the lower limbs, there is less loss of tone with slight wasting and weakness of the quadriceps and marked wasting and weakness of the dorsiflexors of the feet and the right peroneal muscles.

Reflexes.—Upper limbs—triceps, biceps and supinator absent on both sides; abdomen—present; lower limbs—knee jerks and ankle jerks present and equal; Babinski—Flexor plantar response.

Sensation.—Nothing abnormal detected.

Gait.—The patient walks with a high steppage gait characterized by marked “slapping” of the feet on the ground.

Other than the above findings, nothing abnormal was detected in the central nervous system. Thyroid palpable but does not appear enlarged or nodular. Penis appears normal in size and scrotum well developed. The right testis is atrophied and hard, and testicular sensation diminished compared with left testis. The latter is normal. Nothing abnormal could be detected in the lungs, heart or abdomen.

Mental State.—His stream of talk is relevant, rational, logical and coherent. There is no evidence of disordered thinking. His mood is one of anxiety and apprehension, and congruous with his thought content. He is of average intelligence and there is no evidence of mental deterioration. Memory is intact. Concentration and attention unimpaired. There is no evidence of any psychosis.

Special Investigations

Eyes.—External examination of eyes shows some slight exophthalmos, probably due to some loss of orbital fat; otherwise normal. Slit lamp shows posterior polar cupuliform cataracts right and left, brown in colour but with no polychromatic lustre. Amblyopia of right eye since childhood.

X-Ray of Skull.—Small sella turcica and calcification of interclinoid ligaments.

Cerebrospinal Fluid.—Fluid clear and not under pressure; Gram’s stain, nothing abnormal seen; Ziehl-Neelsen, no acid-fast bacilli; culture, no growth; sugar, 65 mg. per cent.; chlorides, 725 mg. per cent.; proteins, 20 mg. per cent.; Pandy’s test, negative.

Electrical Reactions in Muscles (10th March, 1952).—These show a myotonic response in both upper and lower limbs. Both tibiales anteriores fail to react. Certain muscles responded normally to galvanic and faradic stimulation. Others showed reaction of degeneration, while isolated muscles, particularly of the peroneus longus, deltoid and triceps, showed a myotonic reaction, the galvanic contraction lasting for about five seconds after the current ceased to flow. The muscles of the thenar eminences in both hands and the left tibialis anterior gave no response to either galvanic or faradic stimulation.

Comments

Onset.—The onset of dystrophia myotonica is usually in the third or fourth decade. The symptoms and signs in this patient first presented themselves in the second decade.
There is usually a history of cataract in the preceding generation in the same family. No such history could be elicited in this case.

One of the most impressive features of the disease is the insidiousness of the onset and the slowness with which it progresses. This probably accounts for the apparent insensibility and indifference of the patients to their physical disability until the disease becomes well established.

The literature on the disease indicates that the symptoms vary between myotonia, muscular weakness and defective vision. The case here presented complained initially of muscular weakness.

Muscular Atrophy.—The muscular atrophy involved especially the facial muscles, the sterno-clido-mastoids, the muscles of the forearms and hands, the extensors of the legs and the dorsiflexors of the feet.

Myotonia.—The myotonia in this case was most marked in the handclasp. It was elicited also by percussion in the deltoids, triceps and peronei. One of the earliest groups of muscles to show this reaction is usually the tongue, but in this case no myotonia could be elicited there. In contrast to myotonia congenita (Thomsen’s disease), in which the myotonia is generalized, appears earlier and is the only disability, the myotonia in this disease is limited in distribution and overshadowed by the progressive muscular atrophy. The myotonia is, however, characteristic and diagnostic.

Extra-Muscular Dystrophic Signs.—These consisted mainly of frontal baldness, atrophy of the right testicle and opacities in the lenses. Although mental changes are described in textbooks, e.g., Brain (1951), this patient showed no evidence of any mental deterioration. Caughey and Brown (1950) describe endocrine disorders in this disease, including the presence of a small sella turcica, which in some cases was bridged by an area of calcification. The case here presented did show a small sella turcica with calcification of the interclinoid ligaments.

SUMMARY

A typical case of dystrophia myotonica in a regular N.C.O. is described.

REFERENCES