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Myotonic dystrophy: relative sensitivity of symptoms signs and abnormal investigations

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SUMMARY

Twenty-five symptoms, signs, and abnormal investigations were looked for in 20 patients with clinically-definite myotonic dystrophy. Weakness of facial muscles, neck flexors, and arm external rotators was found in all patients (sensitivity=100%). Arm external rotation has not been reported as a frequently involved muscle in previous clinical studies on myotonic dystrophy. Careful examination of muscle strength may therefore predict which patients may or may not carry the abnormal gene for myotonic dystrophy.

INTRODUCTION

Myotonic dystrophy is the commonest inherited neuromuscular disease as well as the one most varied in its clinical expression.[1] The myotonic dystrophy gene has recently been localised to the q13.3 region on the long arm of chromosome 19 where there are increased numbers of cytidine-thymidine-guanidine trinucleotide repeats.[2] This has enabled the diagnosis to be made on DNA extracted from peripheral blood.

Before the availability of a blood test there were numerous clinical evaluations to determine the sensitivity and specificity of various symptoms, signs, and investigations in myotonic dystrophy. These have become no less relevant now since not everyone with weakness or cataract is likely to have a gene test without some preliminary screening. Furthermore, with the existence of a gene test, there is now a "gold-standard" against which other measures can be judged. We were impressed that the frequency of muscle weakness on careful neuromuscular examination of patients with myotonic dystrophy was higher than that reported in the literature. We carried out the following study to test this hypothesis before the gene test was available.

PATIENTS AND METHODS

Twenty patients attending the Northern Ireland Muscle Clinic were studied. All patients had weakness, myotonia on either clinical examination or electromyography, and a family history compatible with autosomal dominant inheritance. Males and females were represented equally and ages ranged from 17 to 61 years with a mean of 34 years. Each patient was examined by M A who

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was experienced in the clinical evaluation of patients with neuromuscular disease. The symptoms, signs, and investigations set out in the table were recorded as present or absent; the investigations were generally available from the patients' records, but symptoms and signs were determined by clinical evaluation.

Symptoms	Falls	Hypersomnolence
	Dysphagia	
Signs	Cataracts	Testicular atrophy
Weakness	Eyelids	Proximal arms
	Eye muscles	Finger abduction
	Dysarthria	Hip flexion
	Face	Knee flexion
	Neck flexion	Knee extension
	Neck extension	Ankle dorsiflexion
	Arm external rotation	Ankle plantarflexion
Myotonia	Action	Percussion
Investigations	Raised CK	Myopathic EMG
	Abnormal ECG	Myotonic EMG
	Abnormal biopsy	

Muscle strength was recorded manually using the MRC scale and classified as either normal (grade 5) or abnormal (grade<5). Action myotonia was assessed by handgrip and percussion myotonia from thenar eminence and finger extensors. Electromyographic studies were carried out with concentric needle electrodes usually from deltoid, biceps, abductor digiti minimi, quadriceps and tibialis anterior. Muscle biopsies fom quadriceps were taken using a UCH needle and frozen in isopentane cooled in liquid nitrogen. Cryostat sections were stained with haematoxylin and eosin, Gomori's trichrome, acid phosphatase, NADH-tetrazolium reductase and ATP-ase pre-incubated at pH 9.4, 4.6, and 4.3.

RESULTS

The frequency of weakness in each muscle group is shown in figure 1 and that of the remaining symptoms, signs, and investigations in figure 2. Face, neck flexors, and arm external rotators were universally weak whereas knee flexion and neck extension were rarely involved. Myotonia was by definition present but was more reliably obtained by percussion than by handgrip. Cataract was the commonest somatic feature but was much less prevalent than weakness and myotonia.

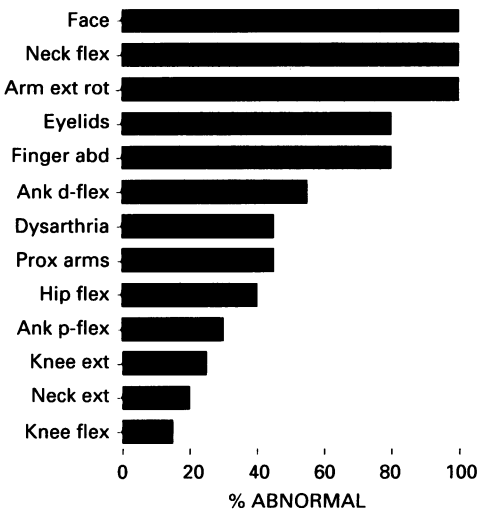

Fig. 1 Frequency of muscle weakness.

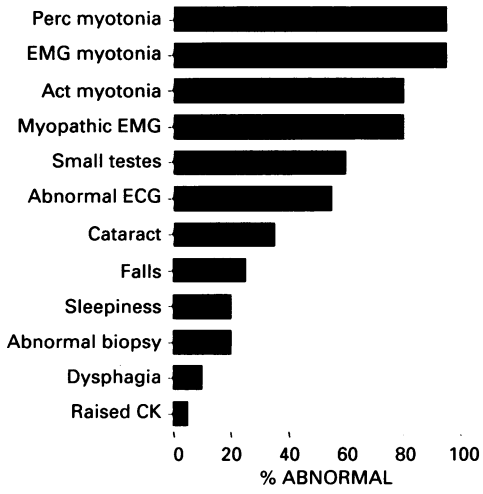

Fig. 2 Frequency of other abnormalities.

DISCUSSION

The measures which we studied differed greatly in their frequency of occurrence in patients with myotonic dystrophy. Weakness of the face, neck flexors, and arm external rotators achieved a sensitivity of 100%. Whereas the first two are well-known to be involved in this disease, weakness of the external rotators of the arm does not seem to have been commented on elsewhere as a frequent occurrence. This movement is carried out by relatively small muscles, principally infraspinatus, and the examiner tests these using pressure on the wrist with the elbow at 90°. The forearm then acts as a lever which generates a considerable torque. Lesser degrees of weakness may therefore be apparent than when a larger muscle with a smaller moment such as deltoid, is tested.

Apart from myotonia which was a criterion for inclusion the other variables showed relatively low sensitivity. The low frequency of cataracts was probably related to use of an ophthalmoscope with a +20D lens rather than a slit-lamp. Results on sensitivity of cataracts in myotonic dystrophy have varied but in a recent study this was 86% for all types of cataract compared with 61% for orbicularis oculi weakness.[3] Muscle biopsy was relatively insensitive with only 20% of biopsies showing the so-called "characteristic" appearances of myotonic dystrophy – increased central nuclei and type 1 fibre atrophy.

The finding of a group of abnormalities with 100% sensitivity would enable the diagnosis to be excluded in their absence and thus would be a useful screening procedure prior to gene testing. This would still be necessary as weakness of face, neck flexion, and arm external rotation is certainly not specific to myotonic dystrophy. This high sensitivity was obtained on an obviously affected group of patients, albeit relatively young. It remains to be seen whether they will be as sensitive in at-risk first degree relatives of myotonic dystrophy patients. The advent of a specific gene test will thus enable the value of careful clinical examination to be tested definitively.

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