multi-organ involvement in MCP; onset of MCP is quite variable, and patients with suspected MCP should be thoroughly investigated for clinical and subclinical affection of the CNS, PNS, endocrinologic system, heart, eyes, ears, guts, dermis, and bone-marrow.

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Diagnosis of mitochondrial diseases: Clinical and histological study of sixty patients with ragged red fibers: Authors’ Reply

Sir,

We thank Dr. Finsterer for his interest in our study. The aim of our study was to find the usefulness of the diagnostic criteria proposed by Bernie et al.² to diagnose mitochondrial diseases when the patients present to the clinician. The clinical characteristics present at the initial visit were considered while categorizing the patients into various clinical syndromes. We have taken the presence of ragged-red fibers (RRFs), > 2% in the skeletal muscle as one of the important criteria for the diagnosis as we have no facilities for studying defects in respiratory chain (RC) complex expression and molecular studies to identify nuclear or mtDNA mutation of probable pathogenicity. The following is the clarifications to the concerns raised by Dr Finsterer.

Presenting phenotypic syndromes was the indication for muscle biopsy in 26 (43%) patients. In the remaining 34 patients we considered the possibility of mitochondrial disease clinically as they had two of the three clinical criteria proposed by Bernie et al.¹

We categorized the patients into various clinical syndromes based on the presenting clinical characteristics at the initial visit. In mitochondrial diseases RRFs can be demonstrated in the clinically uninvolved muscles. Like in many center we do EMG in the right vastus lateralis and use left vastus lateralis biopsy.

We agree that multisystem involvement is a feature in a significant proportion of patients. As mentioned above we considered the presenting clinical characteristics at the first visit for the analysis. This may probably be one limitation for the low frequency of some of the systemic manifestations. It is quite possible that the patients would have developed some of these features in the course of the illness.

Partial abnormality in mitochondrial DNA may cause characteristic cardiac change in mitochondrial diseases and clinical features of cardiac involvement vary according to the different subgroups of mitochondrial disease: Kearns-Sayre syndrome - AV conduction disturbances in Kearns-Sayre syndrome, asymmetrical septal hypertropy progressing to dilated cardiac myopathy in MERRF, and symmetrical ventricular hypertropy with or without abnormal wall motion in MELAS. Cardiac involvement is atypical in ocular myopathy.² This may explain the low frequency of cardiac involvement in our series. In our series of the 60 patients studied progressive external ophthalmoplegia was the presenting clinical syndrome in 26 (43%) patients. Lack of comprehensive cardiac workup might have also been a factor. ECG showed evidence of complete block in 3 patients and one of them had dilated cardiac myopathy. All the three patients had permanent pace maker implantation.

In mitochondrial myopathies the serum CK is almost always normal or only mildly elevated (less than 3 times the upper limit of normal).³,⁴ Rather normal serum CK in patients with significant weakness raises the possibility of mitochondrial dysfunction.³,⁴ Electromyography (EMG) may be normal.³,⁴ The discrepancy between the mean age at onset, mean age at diagnosis and mean disease duration is probably related to wide age range. Of the 60 patients, 10 were in the pediatric age groups (one month to 15 years).

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References

Biomechanics responsible for effect of elbow position on biceps tendon reflex

Sir,

I read with interest the electrophysiological study published by Keles et al. quantifying the effect of elbow position on biceps tendon reflex in normal healthy volunteers. The amplitude and reflex onset latency were studied at 3 different elbow positions, at 90º, 120º, and 150º of joint angles. It was interesting to note in this study that the mean amplitude decreased progressively from 90º to 150º. The amplitude of the biceps tendon reflex is proportional to the net forces acting at the elbow in the direction of contraction of the biceps muscle during the biceps tendon reflex, which include the force of contraction (F) of the biceps muscle and the opposing force of the horizontal component of the ‘tension’ (T) in the forearm due to the weight of the forearm. The force of contraction of the biceps muscle during the reflex is again dependent on various factors such as the strength of tendon tap, influences of reciprocal inhibition etc. As mentioned in this study, if care is exerted to maintain the strength of the tendon taps similar, the amplitude may, hence depend primarily on the opposing force of the forearm tension.

When the elbow is placed at 90º, as seen in Figure 1, tension ‘T’ is equal to the component of the weight of the forearm in vertical direction i.e. ‘mg’ (‘mass of the forearm’ X ‘acceleration due to gravity’). The horizontal component of this force which opposes the force of contraction of muscle is mgCos 90º i.e. zero.

When the elbow is placed at 120º, as seen in Figure 2, tension ‘T’ along the direction of the forearm will be mgCos 30º. The horizontal component of this force opposing ‘F’ will be equal to (mgCos 30º) X 0.57 mg or approximately half the weight of the forearm.

When the elbow is placed at 150º, as seen in Figure 3, tension ‘T’ along the direction of the forearm will be mgCos 60º. The horizontal component of this force opposing ‘F’ will be equal to (mgCos 60º) X Cos 30º or 1.73 mg. Therefore at 150º, the opposing force to muscle contraction was almost equal to twice the weight of the forearm, which led to a statistically significant decrease in the amplitude at 150º as reported by Keles et al.1

Evidently the opposing force to the muscle contraction due to the horizontal component of the tension in the forearm increased with the increase in the elbow angle and thence led to the decrease in the amplitude of the muscle reflex with increasing angle. Clinically, most commonly appreciated character of a DTR is its amplitude and is thus accordingly noted as exaggerated, normal or diminished reflex. Hence to obtain the maximum amplitude in order to make an appropriate clinical judgment about the biceps tendon reflex, it is best to tap the biceps tendon with the elbow placed at an angle of 90º.