SUMMARY

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Metabolic myopathies are a group of hereditary disorders which are relatively common and often treatable group of muscle disease. However, recognition of these conditions is often difficult owing to their clinical heterogenicity. We aim in this study to highlight the clinical presentation of this group of muscle disorder and to evaluate the use of the available clinical investigations including CPK, lactate and L-carnitine as well as histopathological analysis of muscle biopsy specimens in their diagnosis.

This study included 34 myopathic patients with an age ranging from 1-18 years. Patients were classified into two groups; group (1) comprising 24 patients clinically suspected as having metabolic myopathies and group (2) including 10 patients with clinical and histopathological criteria of muscular dystrophy.

Patients with metabolic myopathies were further categorised into four subgroups based on the electron microscopic study of their muscle biopsy specimens; group I (a) included 5 patients with mitochondrial myopathies; group I (b) 7 patients with glycogen storage myopathies, group I (c) 4 patients with lipid myopathies and group I (d), atypical group comprised 8 patients in whom the clinical picture was highly suggestive of a metabolic aetiology, however electron microscopic study of their muscle biopsies revealed non specific myopathic changes with no definite metabolic abnormalities.

Concerning the clinical presentation, certain clinical features were peculiar to the metabolic group as the presence of deafness, external ophthalmoplegia, seizures, encephalopathy and ataxia in addition to generalised hypotonia or proximal myopathy.

Biochemical data revealed a significantly higher CPK in the dystrophic group so that a normal or mildly elevated CPK in the presence of a clinically evident myopathy should raise the suspicion concerning the presence of a metabolic disorder. Lactate estimation was not a useful marker, whereas a significant hypocarnitinaemia was found in group (1) and hence serum L-carnitine could be a useful biochemical marker in suspecting a metabolic muscle disorder.

Histopathological analysis of muscle was the single most useful investigation being diagnostic in 16/24 patients with suspected metabolic myopathies. Further processing of the muscle specimen to enzyme histochemistry should be performed especially in suspected cases with normal or non specific myopathic changes.

Metabolic myopathies are a potentially treatable group of muscular disorders, so a specific diagnosis is mandatory for early and prompt institution of treatment, and for prognostic and genetic implications.