عنوان فارسی مقاله:
اختلالات (بیماری های) متابولیسم لیپید ماهیچه:
مشکلات درمانی و تشخیص

عنوان انگلیسی مقاله:
Disorders of muscle lipid metabolism: Diagnostic and therapeutic challenges

توجه!
این فایل تنها قسمتی از ترجمه میباشد. برای تهیه مقاله ترجمه شده کامل با فرمت ورد (قابل ویرایش) همراه با نسخه انگلیسی مقاله اینجا کلیک کنید.
6. Conclusion and perspectives

Many patients in whom muscle biopsy shows lipidosis remain without diagnosis despite thorough investigations [9]. This low rate of diagnosis of muscle lipidosis could be explained by the following possibilities: (1) the physiological and inter-individual variability of lipid accumulation within muscle fibres limiting the accuracy of the pathological diagnosis; (2) the possibility of still unknown metabolic diseases; and (3) secondary increase of lipid in muscle due to other diseases without primary enzymatic defect.

Diagnosis of a metabolic myopathy can be difficult, particularly in late-onset case, as there may be high residual enzyme activity with few detectable biological abnormalities at rest or at distance of acute manifestations. In addition, some of these biochemical defects, such as NLSD, are only expressed in muscle without possibility to detect biochemical anomaly in blood analysis. Nevertheless, many of patients with a metabolic disorder show abnormal blood acylcarnitines when analysed by tandem mass spectrometry which should be undertaken in all case of unexplained muscle lipidosis. For many disorders it is also now possible to identify the causative genes, thus improving the diagnosis and genetic counselling.