Diagnosis of muscle diseases in the era of next generation sequencing.

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Muscle diseases comprise a large and heterogeneous group of acquired and inherited diseases. For many decades, muscle biopsy was considered as the gold standard laboratory examination for diagnosis of both acquired and inherited muscle diseases. Next generation sequencing (NGS) technologies provide us with the possibility to map entire genomes or exomes at affordable costs and though influence of our daily clinical practice in terms of diagnosis. The application of NGS technologies is transforming the practice of clinical neurogenetics and revolutionizing the approach to heterogeneous hereditary conditions, including muscle diseases. In recent years, cohort studies showed that the overall diagnostic rate of NGS strategies for patients with inherited muscle diseases is higher than the success rate obtained using the traditional approach gene-to-gene approach. Moreover, many experts pointed to the expansion of clinical phenotypes associated with already known disease genes. In the light of the aforementioned progress muscle biopsy is still a gold standard laboratory examination for acquired muscle diseases and for those where NGS failed to identify a causative gene.