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Format: Abstract

Myoimaging in Congenital Myopathies.

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Author information

Abstract

There is a great clinical and genetic heterogeneity in congenital myopathies. Myo-MRI with pattern recognition has become a first-line complementary tool in clinical practice for this group of diseases. For diagnostic purposes, whole-body imaging techniques are preferred when involvement is axial or diffuse, as in most congenital myopathies, because of involvement of the tongue, masticator, neck or trunk muscles. Myo-MRI is widely used to identify abnormalities in muscle signal, volume or texture. Recognizable profiles or patterns have been identified in many of these genetic myopathies. The role of the radiologist is crucial in order to adapt the Myo-MRI protocols to the age of the patient and several clinical situations. Myo-MRI in children with congenital myopathies is a very demanding technique with a balance between acceptable time of examination and sufficient spatial resolution in order to detect subtle changes. Technical evolutions combining qualitative and quantitative analysis are useful to follow disease progression overtime. Outcome measures are expected to play a role in natural history description as well as in future therapeutic trials. Genetic diagnosis and interpretation of next generation sequencing results could be greatly influenced by statistical analysis with tools such as algorithms as well as graphical representations using heatmaps.

Publication type, MeSH terms

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