

Distal Muscle Diseases

Abstract

Distal muscle diseases are primary genetic muscle disorders with progressive loss of muscle tissue causing at onset prominent weakness in hands and/or feet. Some may progress later to involve proximal muscles while others remain mainly restricted to distal limbs. The age of onset and the histological findings are extremely variable. High throughput sequencing has further expanded the long list of genes associated with a distal muscle disease. Currently, more than 20 genes have been associated with autosomal dominant forms and four genes are known causes of autosomal recessive forms. Disease-causing variants in five genes result in either dominant or recessive disease and we have recently identified the first X-linked form. And we have also reported an even more complex digenic mechanism as the cause of a late onset distal myopathy.