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## Characterization of Congenital Myopathies at a Korean Neuromuscular Center.

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### Abstract

**INTRODUCTION:** Congenital myopathies are muscle diseases characterized by specific histopathological features, generalized hypotonia from birth, and perinatal complications, although some cases develop during childhood or rarely even in adulthood. We undertook the study to characterize congenital myopathies among patients registered at our institution.

**METHODS:** Clinical, histopathological and genetic features were evaluated in 34 patients recruited for this study.

**RESULTS:** The majority of patients experienced a childhood onset, and no disease-related mortality was recorded during follow-up. Functional outcomes were no better for those with late-onset disease, indicating later disease progression can be significant. Nemaline myopathy was the most frequent pathology, followed by central core disease, and centronuclear myopathy. Of the 18 (54.5%) genetically confirmed patients NEB and RYR1 mutations were the most common followed by DNM2 mutations .

**DISCUSSION:** This study shows features not previously reported, and suggests congenital myopathy should be considered an important issue among adult patients. This article is protected by copyright. All rights reserved.

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**KEYWORDS:** central nuclei; congenital myopathy; core; fiber type disproportion; molecular genetics; nemaline rods

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