A Rare Case of Severe Congenital RYR1-Associated Myopathy.

Laforgia N, Capozza M, De Cosmo L, Di Mauro A, Baldassarre ME, Mercadante F, Torella AL, Nigro V, Resta N.

Abstract

Congenital myopathies are a group of rare inherited diseases, defined by hypotonia and muscle weakness. We report clinical and genetic characteristics of a male preterm newborn, whose phenotype was characterized by severe hypotonia and hyporeactivity, serious respiratory distress syndrome that required mechanical ventilation, clubfoot, and other dysmorphic features. The diagnostic procedure was completed with the complete exome sequencing of the proband and of his parents and his sister, which showed new mutations in the ryanodine receptor gene (RYR1), which maps to chromosome 19q13.2 and encodes the skeletal muscle isoform of a calcium-release channel in the sarcoplasmic reticulum (RyR1). This report confirms that early diagnosis and accurate study of genomic disorders are very important, enabling proper genetic counselling of the reproductive risk, as well as disease prognosis and patient management.