عنوان مقاله:

Spinal Muscular Atrophy: A Short Review Article

محل انتشار:

مجله بین المللی کودکان, دوره 2, شماره 3 (سال: 1393)

تعداد صفحات اصل مقاله: 5

نویسندگان:

Farah Ashrafzadeh - *Professor of Pediatric Neurology Ward, Faculty of Medicine, Mashhad University of Medical Sciences, Mashhad, Iran.*

Ariane Sadr-Nabavi - *Assistant Professor of Human Genetic, Faculty of Medicine, Mashhad University of Medical Sciences, Mashhad, Iran.*

Nazanin Asadian - *Students Research Committee, Faculty of Medicine, Mashhad University of Medical Sciences, Mashhad, Iran.*

Javad Akhondian - *Professor of Pediatric Neurology Ward, Faculty of Medicine, Mashhad University of Medical Sciences, Mashhad, Iran.*

خلاصه مقاله:

Spinal muscular atrophy (SMA) is a genetic disorder which affect nervous system and is characterized with progressive distal motor neuron weakness. The survival motor neuron (SMN) protein level reduces in patients with SMA. Two different genes code survival motor neuron protein in human genome. Skeletal and intercostal muscles denervation lead to weakness, hypotony, hyporeflexia, respiratory failure, symmetric muscle atrophy and paralysis in patients with SMA. Manifestations are prominent in proximal muscle of lower extremities. There is no curative treatment for spinal muscular atrophy, and supportive treatment should be considered to improve patients' quality of life and independency. New treatment strategies focus on gene therapy or invent method to increase survival motor neuron protein level. The aim of this study is to review Spinal muscular atrophy (SMA) clinical and molecular manifestations.

کلمات کلیدی:

Molecular analysis, Spinal muscular atrophy, Survival motor neuron protein