Insight Medical Publishing

Pediatrics & Health Research

ISSN 2574-2817

2020

Vol.5 No.2

Spinal Muscular Atrophy- Emerging therapies and future directions

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Abstract

Spinal Muscular Atrophy (SMA) is a very severe neurodegenerative disease which causes progressive muscle atrophy and weakness due to loss of the anterior horn cells in the spinal cord and the lower brain stem nuclei. SMA is second most common autosomal recessive disorder and the most common genetic cause of death in infancy. It is caused by deficiency of survival of motor neuron (SMN) protein due to pathogenic variant in the SMN1 gene. SMA is classified clinically into four subtypes based on the age of onset and maximum motor function achieved. SMA type 1 (most severe form) is present with weakness and hypotonia before six months of age. Type II manifest between ages six and 12 months. Type III manifests between ages 18 to 36 months. Type IV (adult onset) develops muscle weakness in second or third decade. There is no cure for SMA and until recently only supportive treatment was available but the approval of Nusinersen and Zolgesma, has revolutionized the outcome of SMA. Nusinersen (Spinraza) is an antisense oligonucleotide for the treatment of SMA that increases the production of full-length SMN protein. Zolgesma is a gene therapy that restores the deleted SMN1 gene. Both Nusinersen and Zolgesma have demonstrated significant and clinically meaningful efficacy on the achievement of motor milestones and measures of motor function, as well as favorable safety across all types of SMA and significantly greater event-free survival in infants with infantile-onset SMA. It's important to recognize some of the early signs and symptoms of SMA and diagnose presymptomatic patients by Newborn screen for better outcome of this devastating disease.

Biography:

Farida Abid serves as an Assistant Professor in the Department of Pediatric Neurology at Baylor College of Medicine. She is a graduate of Dow Medical College in Karachi, Pakistan. She then moved to the USA. She completed her Pediatric Residency at the University of Medicine and Dentistry at New Jersey (UMDNJ), followed by a Pediatric Neurology fellowship at Baylor College of Medicine. She has worked at University of Oklahoma (OUHSC) as an assistant Professor (2006-2011). Also, she is a graduate of the OUHSC Faculty Leadership Program. She joined Baylor College of Medicine (BCM) in 2011. She is board certified in Neurology with a special qualification in Child Neurology. Dr. Abid is an active member of both community and medical societies. She is a fellow of American Academy of Pediatrics, Member of American academy of Neurology, Child Neurology Society. She has served as CME chair for several meetings in Houston, Texas and a featured speaker on muscular dystrophy and spinal muscular atrophy at conferences both in the US and internationally. She is an active member of Muscular Dystrophy Association and volunteers for MDA camp every year.

Speaker Publications:

1. "Cranial nerve palsies in renal osteodystrophy"; Pediatr Neuro. / 2007 / 36(1) /pp 64-65
2. "Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies"; Genet Med / Vol 21 (3) /pp 663-675

32nd European Pediatrics Congress; Webinar; July 22-23, 2020.

Abstract Citation:

Farida Abid, Spinal Muscular Atrophy- Emerging therapies and future directions, Euro Pediatrics 2020, 32nd European Pediatrics Congress; Webinar; July 22-23, 2020 (https://europediatrics.pediatricsconferences.com/abstract/2020/spinal-muscular-atrophy-emerging-therapies-and-future-directions)