Spinal Muscular Atrophy: Diagnosis and Management in the New Therapeutic Era

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Spinal muscular atrophy (SMA) is an autosomal recessive, progressive motor neuron disease mainly manifested with muscle atrophy and limb weakness in childhood, which caused by deletions or mutations in the survival motor neuron 1 (*SMN1*) gene with retained at least 1 *SMN2* gene, and lead to deficiency of full-length SMN protein. To date, SMA is the leading genetic cause of infant mortality in the world.

There is no current active therapy other than supportive standard of care (SOC) for SMA until 2007. Even so the clinical care of infants and children with SMA has advanced significantly over the past two decades. Newer technologies, such as cough assist machines, non-invasive positive pressure ventilation, and gastrostomy tube feeding now offer home-based pulmonary and nutritional management, and have prolonged the survival of severe SMA infants. In 2016, there is a revisiting the consensus statement for SOC in SMA in 8 care areas: diagnostic and genetic counseling, pulmonary, acute care management in the hospital setting, orthopaedics (spinal curvature, joint contractures, fractures), physical therapy and rehabilitation, gastrointestinal and nutrition, other organ systems involved in SMA, ethical considerations and palliative care.

In the past decade, there has made a significant progress in understanding of both SMA molecular genetics and pathomechanisms. Multidisciplinary investigators have identified different SMN-dependent therapeutic approaches including *SMN2* ISS-N1 targeting antisense oligonucleotides, *SMN2* targeting small molecules, and *SMN1* gene therapy that show promise in treating SMA. Until recently FDA has approved nusinersen, the first treatment drug for children and adults with SMA in US (December, 2016) and marketing authorization in Europe (June, 2017). Numbers of disease modifying interventions are rapidly bridging the translational gap from the bench to clinical trials. In this presentation, we will outline the most interesting therapeutic strategies that are currently developing, which are represented by multidisciplinary ways for the treatment and a changing landscape in this new therapeutic era of SMA.