CLİNİCAL CHARACTERİSTİCS OF CASES WİTH SPİNAL MUSCULAR ATROPHY

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INTRODUCTİON

Spinal muscular atrophy (SMA) is an autosomal recessive neurodegenerative disease and is one of the leading genetic causes of infantile hypotonia. This study aims to evaluate the clinical features of cases with a diagnosis of SMA.

OBJECTIVE

This study aims to evaluate the clinical features of cases with a diagnosis of SMA.

MATERİALS AND METHODS

Twenty-four pediatric patients followed in Eskişehir Osmangazi University Faculty of Medicine, pediatric neurology department were evaluated retrospectively. The diagnosis of SMA was confirmed by the detection of homozygous deletions in the Survival Motor Neuron 1 (SMN1) gene in the medical genetics laboratory. All patients' histories, physical examinations, pre-and post-treatment motor function measurements were investigated.

RESULTS

The study population consisted of 12 girls (50%) and 12 boys (50%). According to the SMA classification, 13 patients (54.16%) were type-1, 8 (33.33%) type-2 and 3 (12.5%) type-3. The mean age of type-1 patients was 53.1±23.4 months (range:6-89 months), and the mean age of type-2 and type-3 patients was 122.8±61.3 months (range:53-236 months) was. The mean age of type-1 patients at the start of treatment is 10.5±9.4 months (range:2-36 months), and type-2 and type-3 patients are 87.7±66.2 months (range:19-204 months) was. The mean CHOP-INTEND score of SMA type-1 patients before Nusinersen sodium treatment was 18±16.2, while it was 25.3±17.1 after the 4th dose. While the mean Hammersmith Functional Motor Scale was 25±16.0 in type-2 and type-3 patients before Nusinersen sodium treatment, it was 33.6±18.1 after the 4th dose.

CONCLUSİONS

SMA is an autosomal recessive neurodegenerative disease characterized by degeneration of alpha motor neurons in the brain stem and spinal cord. Muscle weakness is usually symmetrical and predominates proximally. The spectrum of severity of the disease can range from respiratory failure in the neonatal period to mild proximal muscle weakness recognized in adulthood. The lower extremities are more involved than the upper extremities. SMA is a disease that mostly occurs with the homozygous deletion of the SMN1 gene and presents with the deficiency of the survival motor neuron (SMN) protein, which plays a critical role in motor neuron development. The treatment of SMA patients has been in the form of supportive therapy, including respiratory functions and nutrition, until recently. However, the identification of SMN2 as a molecular target has made progress in therapy. Motor neuron dysfunctions may be reversible when SMN - dependent therapeutic approaches are applied presymptomatically . Induction of SMN levels in spinal cord motor neurons will not completely eliminate the progressive neurodegenerative process, but it is possible to slow it down. Nusinersen is an antisense oligonucleotide approved by the FDA in December 2016 for pediatric and adult SMA patients. In our patients, the mean CHOP-INTEND score of type-1 patients before Nusinersen sodium treatment was 18±16.2, while it was 25.3±17.1 after the 4th dose. While the mean Hammersmith Functional Motor Scale was 25±16.0 in type-2 and type-3 patients before Nusinersen sodium treatment, it was 33.6±18.1 after the 4th dose. SMN-dependent therapeutic approaches; When applied presymptomatically, motor neuron functions can be restored. Therefore, early diagnosis is very critical for SMA patients. Since SMA disease is frequently autosomal recessive and prenatal diagnosis is possible, prenatal genetic counseling was given to the families of all our cases for the next pregnancy. As a result; SMA is a neuromuscular disease that requires a multidisciplinary approach. The treatment of SMA disease is constantly evolving. In recent years, the medical approach has improved survival and quality of life by shifting from purely palliative therapy to targeted therapy.

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