Lek. Karolina Aragon-Gawińska

Tytuł : "Ocena skuteczności i ograniczeń terapii nusinersenem pacjentów z rdzeniowym zanikiem mięśni - dane z rzeczywistej praktyki (real-world experience)"

Rozprawa na stopień doktora nauk medycznych i nauk o zdrowiu w dyscyplinie nauki medyczne.

Promotor: dr hab. n. med. Anna Potulska-Chromik

KLINIKA NEUROLOGII

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Streszczenie w języku angielskim

“Evaluation of efficacy and limitations of nusinersen therapy in patients with spinal muscular atrophy - data from real-world experience”

Spinal muscular atrophy (SMA) is a severe, genetically determined disease leading to progressive motor disability, as well as respiratory failure and death in the first years of life in the early-onset form. SMA occurs in about 1:9,000 births and is associated with the SMN1 gene mutation, inherited in an autosomal recessive manner. It is characterized by a wide phenotypic spectrum, which depends on the age of onset of symptoms. One of the factors determining the clinical form of SMA is the number of copies of the *SMN2* gene, which produces the SMN protein, but in much smaller amounts than the SMN1 gene.

Nusinersen was the first drug registered for the treatment of SMA. Its mechanism of action is based on increasing the production of functional SMN protein from the *SMN2* gene by correcting splicing. The series of publications analyzed the course of treatment with nusinersen in the "real-world" population. It has been demonstrated that treatment with nusinersen in patients with SMA type 1 over 7 months of age is effective and safe, and that better motor status at baseline and a higher response to treatment after 6 months are positive prognostic factors of the acquisition of sitting position in patients. In addition, it has been shown that introduction of appropriate organizational measures and adjustment of the treatment schedule, and above all, mobilization of the medical community and the priority given to the treatment of SMA during COVID-19 pandemic ensured that patients did not experience significant delays in drug administration during this period.