Nusinersen

Disease progression was halted in 60% of cases associated with mutation of the SMN1 gene on chromosome 5 (which codes for survival motor neuron (SMN) protein) with intrathecal administration of SpinrazaTM [1] an orphan drug that costs $125,000 U.S. per injection or $750,000 per year.

Nusinersen,marketed as Spinraza, is a medication used in treating spinal muscular atrophy (SMA), a rare neuromuscular disorder.

In December 2016, nusinersen became the first approved drug to treat SMA while several other compounds remain in clinical trials.

In 2016 and 2017, the Federal Drug Administration and the European Medical Agency approved nusinersen for all types of SMA. It is a splicing modifier that enhances the expression of survival motor neuron and it has to be administered intrathecally. In patients with profound scoliosis, intrathecal administration can be challenging.

Case series

Iannaccone et al. published a retrospective chart review of all SMA patients seen at a single site between 2016 and 2020 for treatment with nusinersen.

They reported 8 patients who underwent placement of an Ommaya reservoir and lumbosacral catheter for drug delivery. Complications included infection and revisions due to catheter separation. One patient required fluoroscopy for injections because of the location of the port site.

They conclude that placement of an Ommaya port is a viable option for patients who have challenges with access to intrathecal space. Practical innovations have the potential to control administration costs, achieve therapeutic value, and promote patient safety [2].

Flotats-Bastardas presented the experience with the implantation of an intrathecal port in a patient with SMA type 2 [3]

1)
FDA. Highlights of prescribing information (SpinrazaTM). 2016. https://www.accessdata.fda.gov/drugsatfda_docs/label/2016/209531lbl.pdf
2)
Iannaccone ST, Paul D, Castro D, Weprin B, Swift D. Delivery of Nusinersen Through an Ommaya Reservoir in Spinal Muscular Atrophy. J Clin Neuromuscul Dis. 2021 Mar 1;22(3):129-134. doi: 10.1097/CND.0000000000000333. PMID: 33595996.
3)
Flotats-Bastardas M, Linsler S, Zemlin M, Meyer S. Nusinersen Administration Via an Intrathecal Port in a 16-Year-Old Spinal Muscular Atrophy Patient with Profound Scoliosis. Pediatr Neurosurg. 2020;55(1):54-57. doi: 10.1159/000504058. Epub 2019 Nov 13. PMID: 31722365.