EFFİCACY AND SAFETY OF CERLİPONASE ALPHA THERAPY İN NEURONAL CEROİD LİPOFUSCİNOSİS TYPE 2

Salih Akbaş[1], Tuğba Hirfanoğlu[1], Alp Özgün Börcek[2], Tuğba Atalay[3], Sebla Endürlük[4], Nazlı Balcan Karaca[1], İlknur Cankurt[1], Recep Kamil Kılıç[1], Deniz Menderes[1], Ayşe Serdaroğlu[1] , Esra Serdaroğlu[1], Kıvılcım Gücüyener[1], Ercan Demir[1], Ebru Arhan[1]

[1.] Gazi University Medical Faculty, Department of Pediatric Neurology, Ankara [2.] Gazi University Medical Faculty, Department of Neurosurgery, Ankara [3.] Gazi University Medical Faculty, Department of Ophthalmology, Ankara [4.] Gazi University Medical Faculty Department of Child Metabolism, Ankara

OBJECTİVE:

Neuronal ceroid lipofuscinosis type 2 (CLN2) is a rare neurodegenerative disease characterized by epileptic seizures, loss of motor and language skills, vision loss and death between the ages of 7-10 years.1 The only approved treatment option is Cerliponase alpha. We report our treatment experience in two CLN2 patients receiving Intraventricular Cerliponase alpha therapy every two week for 18 months.

Material and Methods:

We assessed the clinical, motor, cognitive function of our patients with CLN2 by Clinical Rating Scale (motor and language), Gross Motor Functional Scale (GMFS), Ataxia Evaluation and Grading Scale (SARA), Bayley III developmental screening scale before and every 3 months of the treatment. We also evaluated the retinal degeneration by optic coherence tomography before and every six months of the treatment.

RESULTS

The CLN2 Clinical Rating Scale ,GMFS, SARA and Bayley III Developmental Screening Scale scores remained stable during the treatment period. We had to interrupt the treatment for 3 months in one of our patients .Test scores worsened during that time period. She kept stable after the reinjection. No treatment related serious side effect was recorded. OCT didn't show any progression in retinal degeneration.

CONCLUSION

Regularly administered Cerliponase alpha delays the progression of motor, language, cognitive decline and inhibits retinal lipofuscin deposition in NCL2 patients.

REFERENCES

1-Fietz M, AlSayed M, Burke D, et al. Diagnosis of neuronal ceroid lipofuscinosis type 2 (CLN2 disease): expert recommendations for early detection and laboratory diagnosis. Mol Genet Metab. 2016;119(1-2):160-167.

2-Apak S, Tatlı B. pediatrik Epiloptoloji. 2019;2.

3-Specchio N, Kasteleijn-Nolst Trenite DG, et al. Diag-nosing photosensitive epilepsy: fancy new versus oldfashioned techniques in patients with different epilepticsyndromes.Brain Dev2011;33: 294–300.

4-Nickel M, Simonati A, Jacoby D, et al. Disease charac-teristics and progression in patients with late-infantileneuronal ceroid lipofuscinosis type 2 (CLN2) disease:an observational cohort study.Lancet Child AdolescHealth2018;2: 582–90.

5- Williams RE, Adams HR, Blohm M, et al. Management strategies for CLN2 disease. Pediatr Neurol. 2017;69:102-112.