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Case Report

Cochlear Implantation in a Child with Patau Syndrome

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This study aimed to present the first cochlear implant surgery performed on a patient with Patau syndrome. In the auditory brainstem Response test performed on the 37th month, I-III-V waves at 100 dB were not obtained in the right ear, while I-III-V waves at 90 dB were obtained in the left ear. In the free-field audiometry test done in the first year, the threshold value of cochlear implantation was found to be 45 dB. While the Meaningful Auditory Integration Scale test result was 35/40, the Meaningful Use of Speech Scale test result was 13/40. The cochlear implantation was observed and found that hearing results are good and had a positive effect on the quality of life.

KEYWORDS: Auditory brainstem response, cochlear implantation, Patau syndrome, sensorineural hearing loss, trisomy 13 syndrome

INTRODUCTION

The trisomy 13 was first defined in 1960 by Patau. Congenital heart defects (%80), holoprosencephaly, cleft lip and palate, microphthalmia, neural tube defects, nasal malformation, microcephaly, ear malformation, hearing loss, gastrointestinal pathologies (omphalocele, hernia), urogenital problems (polycystic kidneys, micropenis, or hypertrophy of the clitoris), and polydactyly may be seen in trisomy 13 patients.[1-4] Trisomy 13 is one of the most common trisomies and its incidence is 1/5000. Diagnosis is made at birth or during the prenatal period. The average life is 7-10 days and 90% of the patients live less than 1 year.[2,5] Baty et al[6] showed higher survival ratio in girls than boys. The reason for Patau's syndrome is that there are 3 chromosomes in the 13th chromosome. The most common reason for this is non-disjunction of the 13th chromosome during the meiosis.[2,3] Prognosis is better in patients with mosaic Patau syndrome and unbalanced translocation.[2]

This study aimed to present the first cochlear implant (CI) surgery performed on a patient with Patau syndrome.

CASE REPORT

We present a case of CI in a male with Patau syndrome. Informed consent was taken from the patient's parent. During pregnancy, his mother was 42 years old and his father was 43 years old. The patient whose birth weight was determined to be 1800 g was born with normal deliveryat 38th week. There is a history of intensive care for 85 days after birth and phototherapy for 15 days. In feet, polydactyly and polycystic kidneys were present. He underwent omphalocele, undescended testis, inguinal hernia, cataract, tracheotomy, and cleft palate-lip surgeries. The diagnosis of trisomy 13 was confirmed with the genetic test. The cranial magnetic resonance imaging (MRI) was normal. After swallowing therapy, swallowing function was relieved and tracheotomy was closed. In addition, esophageal dilatation was performed at 2 years of age due to esophageal stenosis.

The patient is 5 years old at the moment. The audiometry performed at the 34th month and 37th month did not reveal any sound search behavior. In the auditory brainstem response (ABR) test performed on the 37th month, I-III-V waves at 100 dB were not obtained on the right ear, while I-III-V waves at 90 dB were obtained on the left ear. In Ankara Developmental Screening Inventory, general development was 8 months, language cognitive development was 7 months, fine motor development was 6 months, rough motor development was 8 months, and social skill-self-care was 9 months. Temporal bone MRI was normal and bilateral mastoid ventilation disorder was detected in high-resolution computerized tomography (Figure 1). We performed right CI when he was 3 years old. The Nucleus® CI 422 implant was fully inserted through the round window without any intraoperative complications. Impedance, stapes reflex, and neural response telemetry were obtained within normal limits. In the free-field audiometry test in the first postoperative year, the threshold value of CI was 45 dB (Figure 2). In order to evaluate the postoperative 18th-month auditory

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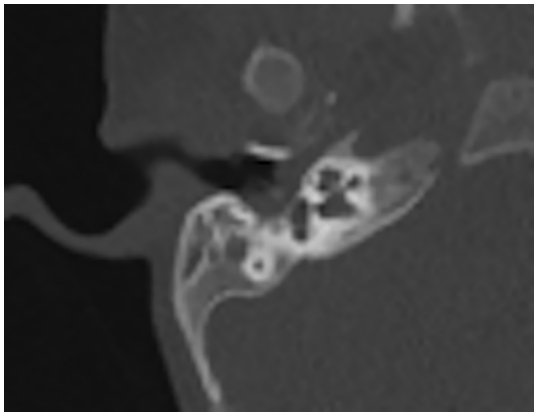

Figure 1. The cochlea is normal on HRCT. However, there is mastoid ventilation disorder. HRCT, high-resolution computerized tomography.

performance of CI, the Meaningful Auditory Integration Scale (MAIS) and Meaningful Use of Speech Scale (MUSS) tests from the EARS protocol tests were used.[7] While the MAIS test result was 35/40, the MUSS test result was 13/40.

DISCUSSION

Trisomy 13 is one of the most frequent trisomy disorders. The prevalence of trisomy 13 is predicted as 1/5000 or 1/6400.[2,4,5] The advanced maternal age is one of the risk factors and the prognosis of these patients is poor.[2,5] Baty et al[6] notified that the maternal and paternal age in patients with trisomy 13 was higher than the normal population (respectively; 31.3 years and 33.7 years; normal population: the mean maternal age 26.0 years and the mean paternal age: 28.2 years). Both maternal and paternal age of our patient was higher than the normal population.

Nelson et al[8] reported that the median survival time is 12.5 days for these patients. Additionally, they reported that 10-year survival is 12.9%.[8] In the literature, there are a few cases with longer life reported.[2] Ilıopoulos et al[9] reported a patient who lived 146 months. They claimed that the patient lived longer because of the absence of holoprosencephaly and cardiac anomaly. Fogu et al[10] presented a 12-year-old patient with Patau syndrome. The long-life expectancy is usually very low by virtue of cardiovascular and cerebral malformations.[2] In contrast, Nelson et al[8] reported that cardiac and neurological diagnoses are not associated with shorter survival. Intensive treatment in Patau syndrome is controversial due to poor prognosis.[2,8]

In a retrospective study, otolaryngologic operations in patients with trisomy 18 and 13 were reported to rank fourth in the top 10 rankings. Additionally, this study showed that the most frequent surgeries were tympanostomy tube placement (25%), cleft lip repair (17%), tracheostomy, (16.5%), tonsillectomy and/ or adenoidectomy (16%), and cleft palate repair (13%).[4] Therefore, otolaryngologists are an important part of the follow-up and treatment of these patients. Although these patients have a high mortality rate, it has been reported that the increase in life expectancy and quality of life because of these patients' good care and surgical interventions and the wishes of families to maintain treatments have changed the approach to treatment.[4]

The CI surgery has not been previously reported in these patients. Fukushima et al[11] have reported a 23-week-old girl with bilateral sensorineural hearing loss. In the ABR test, they showed that no wave was obtained in the left ear and that there were prolonged latencies of waves I, III, and V in the right ear. They found that cochlear nerve fibers and Corti organ were not developed in the upper turns. Additionally, they demonstrated that there are deformities related

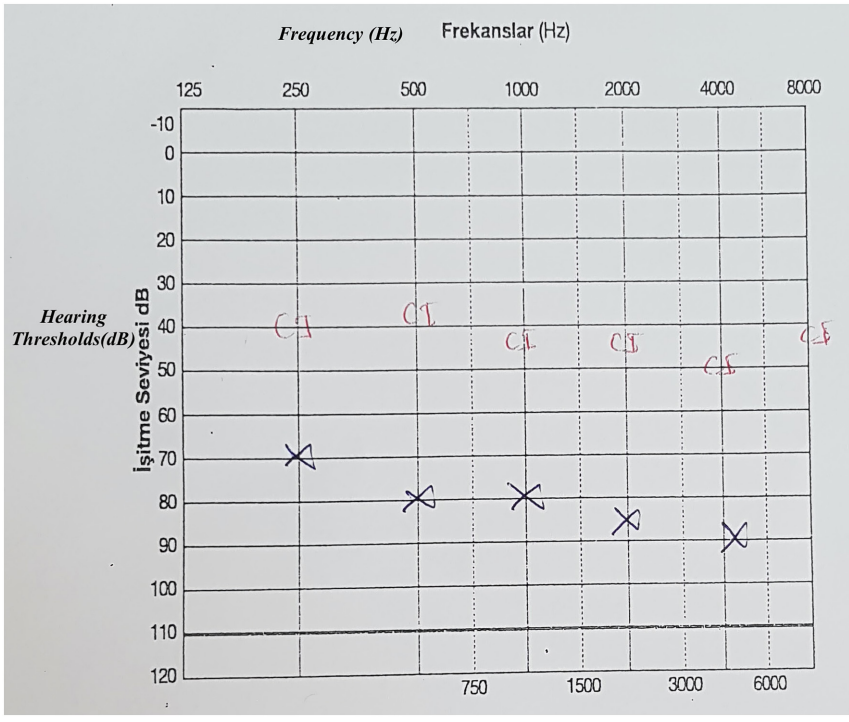

Figure 2. The mean threshold of right CI was measured as 45 dB in the audiometry of the postoperative first year. CI, cochlear implantation.

to semicircular canals, cochlear aqueduct, endolymphatic duct, and modiolus. In our patient's ABR test, I-III-V waves are not obtained at 100 dB in the right ear, while I-III-V waves are obtained at 90 dB in the left ear. Additionally, impedance, stapes reflex, and neural response telemetry were obtained within normal limits intraoperatively. Postoperative CI hearing thresholds were found to be satisfactory. In the postoperative auditory evaluation results, it is seen that the patient partially benefited from CI. Considering that these patients and their families have to cope with many problems, we think that even the slightest improvement in auditory perception will make the lives of these patients easier. The patient and his family are happy with the hearing level and CI application, and hence, the postoperative rehabilitation program continues.

CONCLUSION

Although long life is not generally expected in these patients, each patient should be evaluated separately. Although aggressive treatments are generally not recommended due to poor prognosis, some patients have a better prognosis and this topic should be taken into consideration when planning treatment. This case is the first CI surgery performed on a patient with Patau syndrome. Additionally, the CI is observed and found that the hearing results are good and had a positive effect on the quality of life.

Informed Consent: Written informed consent was obtained from all participants who participated in this study.

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REFERENCES

1. Bruns DA, Campbell E. Nine children over the age of one year with full trisomy 13: a case series describing medical conditions. *Am J Med Genet A*. 2014;164A(12):2987-2995. [CrossRef]
2. Williams GM, Brady R. Patau syndrome. *Stat Pearls* [internet]; 2019. Treasure Island, FL: Stat Pearls Publishing. Available at: http://www.ncbi.nlm.nih.gov/books/NBK538347/. PubMed PMID: 30855930.
3. Kamal M, Varghese D, Bhagde J, Singariya G, Simon AM, Singh A. Anesthesia in a child operated for cleft lip associated with Patau's syndrome. *Braz J Anesthesiol*. 2018;68(2):197-199. [CrossRef]
4. Karimnejad K, Costa DJ. Otolaryngologic surgery in children with trisomy 18 and 13. *Int J Pediatr Otorhinolaryngol*. 2015;79(11):1831-1833. [CrossRef]
5. Duarte AC, Menezes AI, Devens ES, Roth JM, Garcias GL, Martino-Roth MG. Patau syndrome with a long survival. A case report. *Genet Mol Res*. 2004;3(2):288-292.
6. Baty BJ, Blackburn BL, Carey JC. Natural history of trisomy 18 and trisomy 13: I. Growth, physical assessment, medical histories, survival, and recurrence risk. *Am J Med Genet*. 1994;49(2):175-188. [CrossRef]
7. Esser-Leyding B, Anderson I. Anderson I. EARS(R) (Evaluation of Auditory Responses to Speech): an internationally validated assessment tool for children provided with cochlear implants. *ORL J Otorhinolaryngol Relat Spec*. 2012;74(1):42-51. [CrossRef]
8. Nelson KE, Rosella LC, Mahant S, Guttmann A. Survival and surgical interventions for children With trisomy 13 and 18. *JAMA*. 2016;316(4):420-428. [CrossRef]
9. Iliopoulos D, Sekerli E, Vassiliou G, et al. Patau syndrome with a long survival (146 months): a clinical report and review of literature. *Am J Med Genet A*. 2006;140(1):92-93. [CrossRef]
10. Fogu G, Maserati E, Cambosu F, et al. Patau syndrome with long survival in a case of unusual mosaic trisomy 13. *Eur J Med Genet*. 2008;51(4):303-314. [CrossRef]
11. Fukushima H, Schachern PA, Cureoglu S, Paparella MM. Temporal bone study of trisomy 13 syndrome. *Laryngoscope*. 2008;118(3):506-507. [CrossRef]