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Original Article

Cochlear Implantation Outcomes in Children with Agenesis of the Corpus Callosum: A Retrospective Study and A Review of the Literature

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OBJECTIVES: The aim of the present study was to analyze the outcomes of cochlear implantation (CI) in patients with agenesis of the corpus callosum (CCA). A literature review and a retrospective analysis of our cochlear implant database were performed.

MATERIALS and METHODS: To the best of our knowledge, in the English literature, there was only one case reported with CCA who had undergone CI surgery. This case had Donnai–Barrow syndrome. In the Cukurova University School of Medicine Department of Otorhinolaryngology database, 5 of the 1317 patients who underwent CI surgery who had CCA were selected. The patients' demographic characteristics, operative findings, surgical outcomes, and additional disabilities were investigated. The patients' preoperative and postoperative Listening Progress Profile (LiP) and Meaningful Auditory Integration Scale (MAIS) tests were done to analyze the auditory performances.

RESULTS: The participants of the study were 5 (0.38%) individuals (2 male and 3 female patients; ages 5.5, 7.5, 8, 9, and 12 years). Two of the patients had total agenesis, and the other three had partial agenesis of the CCA. In the histories of the patients, one patient had parental consanguinity, and one had febrile convulsion. No patient had an additional disability. None had experienced device failure. No patients were non-users or limited users of cochlear implants. Postoperative LiP and MAIS test scores were improved for all patients nearly as the patients without any deformity. They showed normal auditory performance in the analysis in their postoperative 48 months of follow-up.

CONCLUSION: Patients who had CCA are good candidates for CI surgery.

KEYWORDS: Cochlear implant, corpus callosum, agenesis, auditory performance

INTRODUCTION

The corpus callosum (CCA) is an interhemispheric fiber bunch located in the middle part of the brain. In humans, it contains approximately 200 million axons [1]. Although its presence has been known for centuries, some of its functions could still not be well understood. It is essential for learning, discrimination, sensory experience, memory, and synchronicity of sleep [1, 2]. It consists of four parts: rostrum, genu, splenium, and body. Agenesis of the CCA could be total (absence of all anatomical parts of the CCA) or partial (absence of at least one, but not all, parts of the CCA). Its diagnosis is based on ultrasound and magnetic resonance imaging (MRI) [2-6].

In the literature, the true incidence of CCA is varied because of the asymptomatic cases. However, the reported incidence in children with developmental disabilities is 2%-3% [7-10]. It is frequently associated with different somatic, physical, and genetic diseases, such as attention deficit disorder, autism spectrum disorder, and sensorineural hearing loss (SNHL). CCA has been reported in >50 congenital human syndromes, such as Donnai–Barrow syndrome, Chudley–McCullough syndrome, Aicardi syndrome, and Kabuki syndrome [11-15].

This study was presented at the 10th Asia Pacific Symposium on Cochlear Implants and Related Sciences Congress, April 30-May 3, 2015, Beijing, China.

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To the best of our knowledge, there was only one case reported in the English literature who had CCA and underwent cochlear implantation (CI) surgery. This patient had Donnai–Barrow syndrome. In the present study, we examined the literature and patients in our tertiary referral cochlear implant center database who had CCA radiologically and received cochlear implants. The auditory performances before and after CI and comorbid abnormalities of the patients were analyzed.

MATERIALS AND METHODS

PubMed was searched for studies written in English with subjects "CCA AND SNHL," in which we found 44 studies. When we searched the subjects "CCA AND SNHL AND CI," we found only one case report study. After this literature review, we investigated our database.

Of the 1317 patients who underwent CI surgery at the Cukurova University School of Medicine Department of Otorhinolaryngology, Adana, Turkey between January 2000 and March 2017, 5 (0.38%) cochlear-implanted patients who had CCA were selected. Their medical records were documented. The study was approved by the local ethics committee. Written informed consents from all participants were obtained.

At our clinic, all cochlear implant candidates who had profound SNHL were examined for compliance. The patients consulted to both the psychiatry and neurology departments. Moreover, pure tone audiometry, auditory brainstem response tests, and radiological investigations including computerized tomography of the temporal bone and MRI of the inner ear were performed routinely to all candidates. For the purpose of examining the preoperative and postoperative auditory performances of the CI candidates, we performed the Listening Progress Profile (LiP) and Meaningful Auditory Integration Scale (MAIS) tests that were selected from the EARS protocols [16, 17]. In our study, we selected five patients who neuroradiologically had CCA and received cochlear implants. We investigated their comorbid abnormalities, etiology, and preoperative and postoperative 48 months of auditory performances. These patients were implanted with the same cochlear implant device (MED-EL Company, Austria) at a round window.

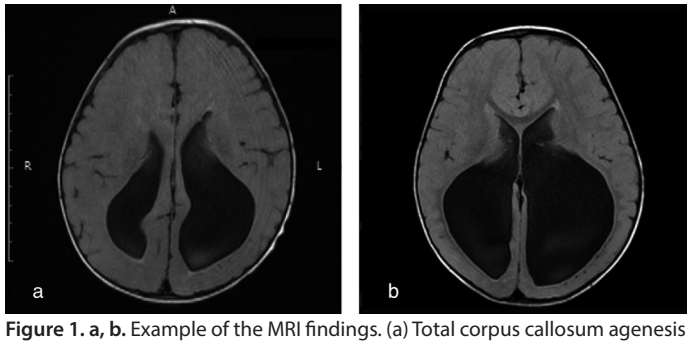

Figure 1. a, b. Example of the MRI findings. (a) Total corpus callosum agenesis and colpocephaly and (b) partial corpus callosum agenesis (absence of the splenium and posterior of the body) and colpocephaly in the T2 FLAIR MRI.

Statistical Analysis

Statistical analyses were performed using the The Statistical Package for the Social Sciences (SPSS) statistical package version 18.0 (IBM Corp.; Armonk, NY, USA).

RESULTS

A total of five patients were included in the study. The study comprised two male and three female patients. The ages of the patients were 5.5, 7.5, 8, 9, and 12 years. Two of the patients had total agenesis, and the other three had partial CCA. In addition to CCA, two patients had cerebellar vermis hypoplasia, and two patients had colpocephaly in the radiological examinations. Examples of the MRI findings of total CCA and partial CCA are shown in Figure 1. With respect to their histories, one patient had parental consanguinity, and one had febrile convulsion. One patient had mild mental retardation. All the patients' perioperative tests were successful. The demographic characteristics of the patients are summarized in Table 1.

No patients were non-users or limited users of cochlear implants. No patient had device failure problem. The LiP and MAIS test scores were improved in the postoperative period, and they showed to be nearly normal as patients without any deformity in the auditory performances. The preoperative and 48-month postoperative results of the tests are shown in Table 2.

Table 1. The demographic characteristics of the patients

	Age (year), sex	Age during implantation (month)	Approach	MRI findings	Preoperative tests	Risk factors for SNHL
Case I	5.5, female	18	Round window	Partial CCA (absence of the splenium and posterior of the body)	Successful	No
Case II	7.5, male	15	Round window	Total CCA, cerebellar vermis hypogenesis	Successful	Mild mental retardation
Case III	8, female	19	Round window	Partial CCA (absence of the splenium and posterior of the body), colpocephaly	Successful	Parents were consanguineous and brother has also total bilateral sensorineural hearing loss
Case IV	9, male	27	Round window	Partial CCA (absence of the splenium and posterior of the body), cerebellar vermis hypogenesis	Successful	Febrile convulsion at 10 months
Case V	12, female	72	Round window	Total CCA, colpocephaly	Successful	No

SNHL: sensorineural hearing loss; CCA: corpus callosum agenesis

Table 2. Postoperative LiP and MAIS test scores

	Preoperative LiP	Postoperative LiP 12 months	Postoperative LiP 48 months	Preoperative MAIS	Postoperative MAIS 12 months	Postoperative MAIS 48 months
Case I	0/42	30/42	42/42	2/40	32/40	40/40
Case II	3/42	34/42	42/42	2/40	33/40	40/40
Case III	3/42	36/42	42/42	0/40	33/40	40/40
Case IV	4/42	42/42	42/42	3/40	38/40	40/40
Case V	2/42	34/42	42/42	2/40	36/40	40/40

LiP: Listening Progress Profile; MAIS: Meaningful Auditory Integration Scale tests

DISCUSSION

The corpus callosum malformations were associated with several different syndromes, including Kabuki (Niikawa–Kuroki) syndrome, Chudley–McCullagh syndrome, and Donnai–Barrow syndrome [11, 12, 18, 19]. The genetic heritage of the CCA may be autosomal recessive, X-linked recessive, or autosomal dominant form [20].

The *PAX2* gene is necessary for normal cochlear development and is necessary for CC development [21, 22]. Some of these malformations have been influenced by different types of the Pax gene homeobox regulators.

In recent years, promising radiological studies in the evaluation of the auditory cortex and auditory pathways in humans and also patients with CCA are reported. The new techniques used in these studies for structural connectivity framework and functional auditory areas in the cortex were functional MRI, diffusion tractography, probabilistic tractography, high-angular-resolution diffusion-weighted imaging, and high resolution 7 Tesla-diffusion tensor imaging, among others [23-25]. Severino et al. [25] demonstrated that even the absence of a small callosal segment may impact global brain connectivity and modularity organization of the brain.

Familial cases of CCA without other anomalies are rare. Wilson et al. [26] reported two brothers with complete CCA, developmental retardation, and cranial problems. Shapira and Cohen [27] defined two sisters from consanguineous parents with CCA. Parrish et al. [28] published 11 cases with CCA. They reported a high incidence of anomalies and no togetherness with deafness.

Donnai–Barrow syndrome or facio-oculo-acoustico-renal syndrome is a rare disease especially with dual sensory loss, for instance, bilateral SNHL and visual problems [12]. It is characterized by craniofacial abnormalities (ocular hypertelorism and enlarged fontanelle), ocular findings (high myopia, retinal problems, progressive loss of vision, and iris coloboma), SNHL, CCA, intellectual disability, exomphalos, and hernia of the diaphragm.

The corpus callosum is one of the most frequently seen malformations in the brain, and it is rarely associated with SNHL. We found only one case with Donnai–Barrow syndrome as reported by Bruce et al. [29] who had CCA and underwent CI surgery. The case was born at full term. The parents were consanguineous. The patient's MRI demonstrated an absent CCA. Hearing loss was first detected at aged 1 month. He started to use bilateral hearing aid at aged 5 months. Although there were extensive developmental retardation and speech problems, the patient's non-verbal communication talents were well improved. He had undergone CI surgery to the right ear at aged 39 months. His perioperative test was successful, and no complication occurred. His verbal communication improved by 2 years.

Cochlear implantation is a common surgical technique for patients who had total SNHL. Wrong indications of this procedure can cause a large number of limited-user or non-user patients in the long term and waste of public money. This would also discourage further funding for this procedure in many countries [30].

In our tertiary referral cochlear implant center database, we had five patients who radiologically had CCA and received cochlear implants. The auditory performances before and after CI and the comorbid abnormalities of the patients were analyzed. During their follow-up, the auditory performance analysis scores improved nearly the same as the patients in the same ages without any abnormality.

CONCLUSION

The corpus callosum (CCA) is rarely associated with SNHL. Patients who had CCA without a syndrome or an additional disability are good candidates for CI surgery.

Ethics Committee Approval: Ethics committee approval was received for this study from the Cukurova University School of Medicine Ethics Committee.

Informed Consent: Written informed consent was received from the patients who participated in this study.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept – S.O., U.T.; Design – S.O.; Supervision – S.O., U.T., F.C., M.K.; Resource – S.O., O.S.; Materials – S.O., O.S., O.T., F.C., M.K., M.D., P.S., N.T., F.A.; Data Collection and/or Processing – S.O., U.T., O.S., O.T., M.K., M.D., P.S., N.T., F.A.; Analysis and/or Interpretation – S.O., O.S., O.T., M.D.; Literature Search – S.O., U.T., O.S., O.T., M.D., P.S., N.T., F.A.; Writing – S.O.,O.S.; Critical Reviews – S.O., U.T., F.C., M.K.

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