CASE REPORT

Language and Speech Disorder in Agenesis of the Corpus Callosum

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Abstract

This study intends to provide information about a rare condition caused by a fiberal abnormality, as well as ongoing assessment and treatment procedures. One 42-month-old participant with corpus callosum agenesis was assessed using the TEDIL and TIGE assessment tests and had speech and language treatment for 18 months following that. The second examination was carried out while therapy was still in progress. In the beginning of the intervention process, the participant was found to produce only 5-6 single words while also exhibiting a variety of behavioral issues. The participant was able to use more words, word combinations, and pragmatic gestures after 18 months of sessions, as well as having better eye contact, imitation abilities, and less behavioral issues. The findings were similar to those of other studies focusing on corpus callosum agenesis, which indicates that the participant had developmental delays and speech and language disorders. It is believed that a multidisciplinary team would be more effective in conducting the intervention and assessment procedure.

Keywords: Agenesis; corpus callosum; developmental disorders; language disorder; speech and language therapy.

Agenesis of the corpus callosum (ACC) is a condition in which the commissural fibers are unable to cross the midline. Instead of a thick band, a blunt band forms, which proceeds posteriorly medial to the lateral ventricles. ACC is caused by a variety of factors. Epigenetic interactions, prenatal infections, maternal rubella, trisomy 8, 13, and/or 18, Andermann syndrome, Aicardi syndrome, and fetal alcohol syndrome, among other factors, may contribute to the diagnosis of people with ACC[1-3]. ACC can be diagnosed on its own or in combination with other conditions such as hippocampal abnormalities, cleft lip and palate, mental retardation, and hearing loss.

When looking through the literature, it becomes clear that most studies on people with ACC are conducted by professionals in the fields of neurology and genetics, and that these studies are done as case reports. There has been research on the development of language and speech in people who have agenesis, but these studies usually have results that can be used to profile the disease. Mentioned results show that individuals with AAC may have developmental delay, low muscle tone, poor motor coordination, feeding and swallowing problems, high tolerance to pain, sleep-related problems (sleepwalking, bedwetting, etc.), difficulty in maintaining attention, hyperactivity, restlessness, difficulty in executive functions such as problem solving, abstract thinking, making inferences, establishing cause-effect relationships, understanding/interpreting social cues, and understanding expectations. Individuals

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diagnosed with AAC have prolonged reaction and processing time in cognitive tasks, particularly when processing complex information[4,5]. Researchers claim that while normal intelligence levels can be seen in some cases, language and speech development is usually delayed[3]. According to studies undertaken to examine the language performance of people with ACC, these people have delayed language and speech development, difficulties in receptive and expressive language in nonverbal communication, and issues related to understanding emotions and thoughts of others[3]. Sensory problems and difficulties in sensorimotor, cognitive, emotional, and language skills are common in people with ACC[5,6]. Many cognitive skills such as learning, memory, recalling and problem solving were found to be impaired in Erickson et al.[6], conducted a study with 26 participants with ACC. In another study by Mangione et al.[7], significant differences in gross motor, fine motor, receptive language, and overall development were reported between the experimental and control groups. In a retrospective study conducted by Le Doussal et al.[8] over a 24-year period, it was discovered that neurodevelopmental outputs were typical or mildly impaired in 88% of the cases, that neurodevelopmental progress was slow in all participants, and that individuals with ACC were at high risk (64%) of speech disorders[8]. Individuals with ACC may be highly sensitive to nuanced language and subtext in social and emotional relationships, or they may misunderstandings during these sort of interactions[9]. The corpus callosum is crucial for lateralization of speech function to the left hemisphere and in ACC cases, there is no left hemispheric specialization for speech function. As a result, cortical language processing occur in two hemispheres and in auditory and visual language tasks, both hemispheres activate in incomplete AAC and predominantly right hemisphere activates in complete ACC[10,11]. It is also suggested in these research that there is a positive correlation between language specific lateralization and verbal IQ, and hence lack of language specific left hemisphere lateralization is linked to poor verbal IQ[10,11].

The aim of this study is to provide information on evaluation and intervention processes in a case of corpus callosum agenesis, which is not commonly seen.

Case Report

Participant

The study included a 3.8-year-old participant who applied to the Üsküdar University Language and Speech Therapy Application and Research Center (ÜSESKOM) with diffi-

culties related to speech output and intelligibility due to corpus callosum agenesis. When corpus callosum agenesis was suspected during a detailed fetal ultrasound examination performed during a routine control in the sixth month of pregnancy, the participant was referred to another hospital, where an evaluation was conducted by a committee consisting of gynecology, pediatric neurology, and neurosurgery clinics, and the participant was followed up with fetal magnetic resonance imaging (MRI) every three weeks (Figs. 1-3). In the eighth month of pregnancy, the participant was diagnosed '*Agenesis of the Corpus Callosum*'. At the age of one month, the participant was referred to rehabilitation by a pediatric neurologist, and at the age of two, the participant began receiving special education support and had private lessons, swimming and horseback riding training and enrolled in kindergarten, as well as three hours of physical rehabilitation per week.

The mother noted that the participant had some behavioral issues, as well as crying to express her self-care needs, and had involuntary movements in her body in circumstances of panic and fright during the initial interview. The mother also mentioned that the participant was so stubborn and also the speech intelligibility is very low even though the participant could produce all sounds. What the participant could do, according to the mother, is as follows:

- Making eye contact with parents.
- Maintaining joint attention for a certain period of time.
- Looking at her parents and laughing when she is joyful.
- Playing with toys at home.
- Practicing what is learnt during sessions after modelling cues home.
- Enjoying weekend activities.
- Trying to adjust new people and environment at kindergarten.
- Not crying at kindergarten.
- Being extremely fond of father.
- Enjoying spending time on mobile phone.
- Being able to say '*mother*' and '*father*'.

Data Collection Method

In the study, detailed assessment of the participant's speech and language skills were conducted twice, at the beginning and after the 18th session of the therapy process. The language development was evaluated with '*Test of Early Language Development*' (TEDIL- Topbaş & Güven, 2014), the Turkish version of TELD-3 (Test of Early Language Development) which aims to assess receptive and expressive lan-
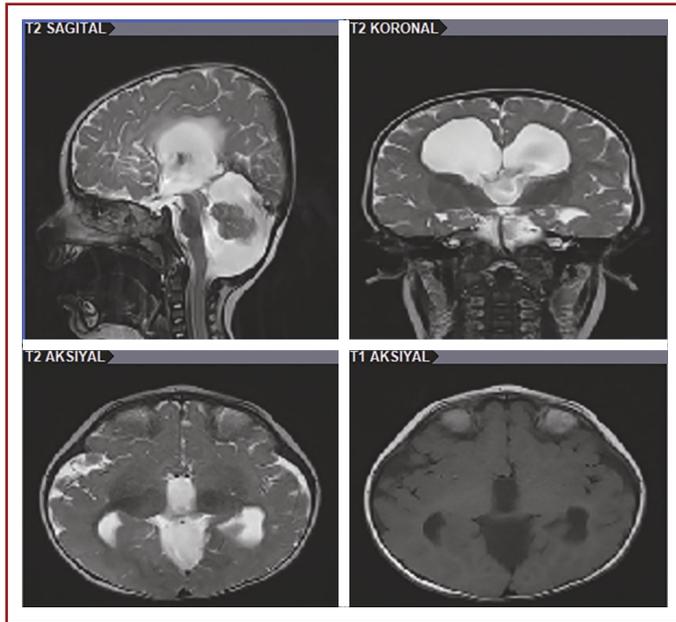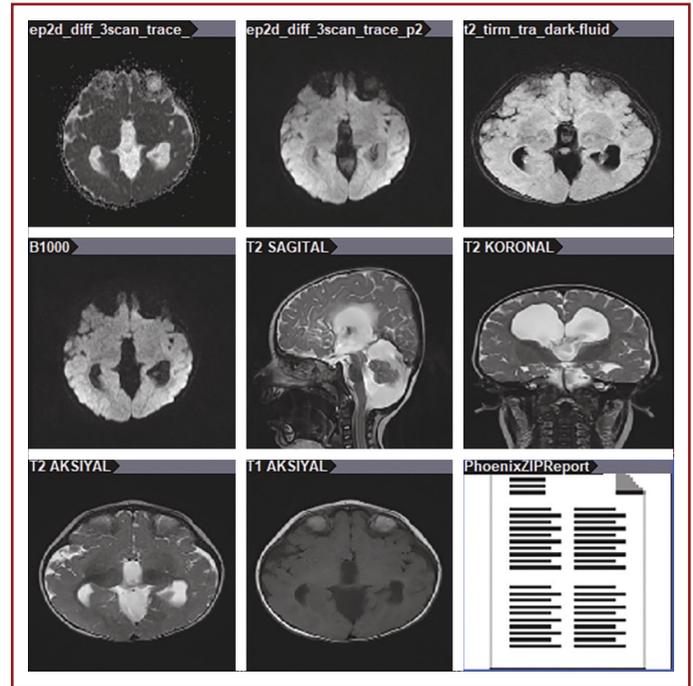
Figure 1. Magnetic resonance imaging (MRI) showing corpus callosum agenesis (CCA).
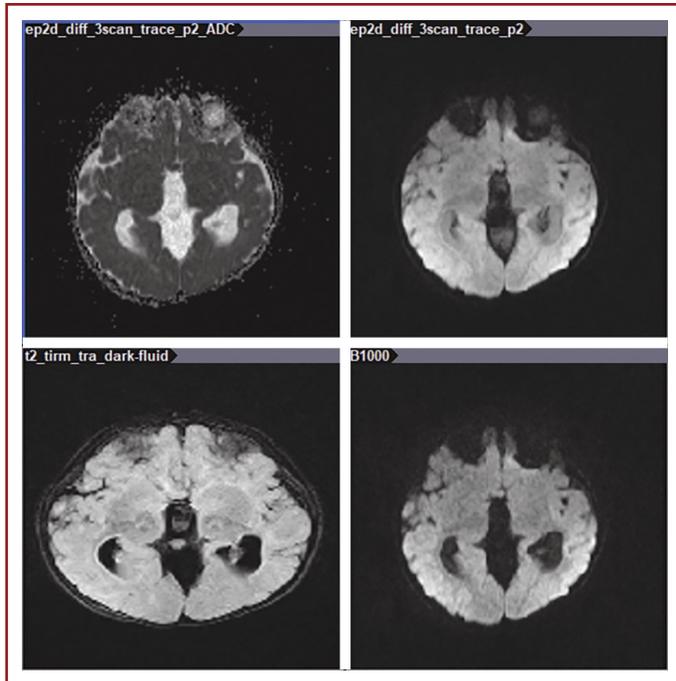
Figure 2. Magnetic resonance imaging (MRI) showing corpus callosum agenesis (CCA).

guage skills of children aged 2;0-7;11. TEDIL includes two forms, Form A and Form B, and 76 items in total. Besides TEDIL, Turkish version of the '*MacArthur-Bates Communicative Development Inventory*' (Fenson et al., 1993; MB-CDI) (Türkçe İletişim Gelişimi Envanteri, Acarlar et al., 2009; TIGE) was used to gather information. The TIGE consisted of two forms: TIGE I-TIGE II which evaluate the language skills of

Figure 3. Magnetic resonance imaging (MRI) showing corpus callosum agenesis (CCA).

'16-36 months' children based on parental opinion. TIGE II (16-36 months) was used in present study. In addition to standardized testing procedures, the '*Pediatric Family Interview Form*', which were prepared to be used within USESKOM, were completed. During informal assessment procedures, the parents are interviewed with detailed open-ended questions and the participant was observed in terms of motor and language development during free play.

Initial Assessment

During the initial evaluation phase, the participant did not reply to any instructions in the TEDIL and scored zero points in the receptive-expressive language domains. The parents were unable to complete TIGE II due to a lack of data on which to fill out the language performance form. The language samples acquired were not qualified or sufficient for transcription, hence transcription and analysis of the sample were not possible. It was observed that the participant did not have any linguistic output and lacked preverbal pragmatic skills. Additionally, evidence of apraxia was discovered during an oral motor evaluation.

Second Assessment

A total of 18 weekly speech and language therapy sessions (45 minutes each) were completed. The participant's eye-contact skills improved after the first four sessions compared

to the previous sessions. The participant was more interested in activities involving oral motor skills, but the verbal output was still lacking. The parent was given the TIGE II again at the second assessment, and TEDIL was administered to the participant once again. Based on the data obtained after conducting the TIGE test, it was discovered that the participant was significantly behind peers. The participant was only able to say 8 of 711 words across 21 categories in TIGE II. In vocabulary section of TIGE, only two of the five items, '*often*' and '*sometimes*' were reported to have been used. Due to a lack of relevant and sufficient language output, the grammar section of TIGE II was unable to be completed. The longest sentences heard from the case were "*Işığı kapa (Turn off the light)*" and "*Ersel gitti (Ersel is gone)*" and both of which were two-word combinations, as the family reported in the last section of the TIGE II exam. When the data was analyzed, it was discovered that the expressive language skills of the participant were well behind of peers.

The participant's TEDIL test results are listed in the table below (Table 1).

The difference between the TEDIL receptive and expressive Language Subtest scores is higher than 15, indicating that the participant seems to have a language disorder. Receptive language was also found to be significantly higher than expressive language.

Interviews with parents were conducted in order to assess the process and determine the impact of the intervention on language development. The participant's parents reported that the participant demonstrated positive improvements in initiating and maintaining communication. While the participant could only produce 5-6 words at the beginning of therapy, after 18 sessions, the participant was able to produce approximately 20-25 words. Furthermore, it was noted that the participant progressively started to show object through gestures, as well as performing verbal and motor imitations. It was reported that the participant produced two word combinations in some situations and behavioral problems such as crying and hitting were reduced.

During the initial assessment, it was noted that the participant lacked a sufficient language output to assess language performance. However, the results of the post-therapy assessment revealed that, while the participant could not yet perform at a level comparable to peers in terms of language, she was improving and positive improvements in the development of behavior and language performance developed in participant allowing evaluation.

Discussion

The language abilities of a participant diagnosed with ACC were assessed before and after speech and language therapy in this study. Similar to other studies in the literature, it was found that she had developmental delay compared to peers and did not have a language development suitable for her chronological age[3,5,12]. Multiple structural and functional problems such as developmental delay, low muscle tone, poor motor coordination, sucking/ eating problems, sleep-related problems (sleepwalking, bedwetting, etc.), difficulty maintaining interest, hyperactivity, and restlessness, which are frequently reported in individuals with ACC, were also present in the participant of this study[12-14]. In our study, we found that the participant with ACC was in the high-risk group for a diagnosis of speech and language disorders, similar to findings of Le-Doussal et al[8]. Receptive language skills are better than expressive language abilities in individuals with ACC, however their performance differs from their peers[7]. Relevant assessments have also indicated that our participant had considerable delays in word and sentence use. Language performance in the ACC can be defined within the framework of issues noticed in delayed language and speech, receptive and expressive language in nonverbal communication, as noted by Badon et al.[3] and it was also accurate in our study. In addition to the language delays, the individual has been observed during therapy to have a variety of sensory and motor deficits, which is consistent with the literature[4,5]. Unlike typically developing peers, our participant was found to have impairments in fine and motor abilities, as well as difficulty walking and engaging in tasks that needed fine motor skills as reported in the literature[7]. Even after short-term (18-session) therapy, encouraging improvements in the participant were observed related

Table 1. Findings related to TEDIL receptive and expressive language subtests

Receptive language raw score	Expressive language raw score	Receptive language std. score	Expressive language std. score	Receptive language equivalent age (Month)	Expressive language equivalent age (Month)
18	8	105	59	39	12

*: The participant is 3.5 years old at the time of this assessment; TEDIL: Test of Early Language Development (Topbaş&Güven, 201).

to difficulties in the first assessment such as the absence of verbal output, difficulty exhibiting pragmatic skills such as having eye contact-joint attention, and frequent behavioral problems. In circumstances where verbal output is limited, reinforcing preverbal pragmatic gestures such as pointing rather than unpleasant behavior such as '*crying*' has improved communication efficiency. Although there are few studies on the effectiveness of speech and language therapy in ACC cases, based on the findings of this study, it is believed that with an effective intervention process, individuals who have been diagnosed by an interdisciplinary team can develop skills to compensate for their deficits.

Conclusion

Different assessments to determine general development, language, and speech characteristics in ACC are thought to be studied with studies involving more participants for more detailed and consistent results. Clinicians and researchers will benefit from new studies assessing the efficacy of speech and language treatment in which alternative goals are pursued in areas such as voice, swallowing, or prosody, and/or evidence-based language intervention approaches are applied systematically in ACC cases. Cases with ACC should be followed up in a multidisciplinary framework by experts in disciplines such as pediatrics, pediatric neurology, genetics, endocrinology, psychology, physiotherapy, and speech-language therapy, because they frequently present with a clinical picture that combines multiple disorders. This suggestion is supported by our findings during the assessment and therapy processes of the participant we presented.

Other diagnoses or other conditions that may impact language function, such as cerebellar atrophy, which can be seen on MRI scans (Figs. 1-3), were also present in our participant. However, due to the participant's severe language development impairments, evaluating the effects of these factors on language independently wasn't practicable. However, it may be proposed that these factors be considered when designing a study, if any at all possible.

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