



## AUDIOLOGICAL AND SPEECH MATERIALIZATION IN CORPUS CALLOSUM AGENESIS: TWIN CASE STUDY

### Clinical Research

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### ABSTRACT

**Introduction:** Agenesis of corpus callosum is a rare birth defect that occurs when the band of white matter connecting two cerebral hemispheres in brain fails to develop normally, typically between 12 to 20 weeks of pregnancy. Depending on the site of lesion and degree of damage to the corpus callosum, different symptoms get evident. Delayed motor and speech development are commonest among children with agenesis of corpus callosum. Other symptoms include vision, hearing, cognition and sensory deficit.

**Case Report:** The present case study illustrates 11 months old baby boy and 2 years old baby girl with a spoken language disorder in a known case of agenesis of corpus callosum. Detailed speech, language and audiological assessments were done to define child's level of linguistic functioning and hearing loss.

**Aim of Study:** To document the nature and characteristics of speech and language development in those individuals with agenesis of corpus callosum.

**Conclusion:** Although precise condition is unknown, agenesis of corpus callosum is a rare genetic condition that occurs among children and typically affects the speech and language development. Speech Language Pathologist plays a significant role in restoring, adjusting and compensating the development of speech and language in child with agenesis of corpus callosum.

### KEYWORDS

Corpus callosum, Receptive Expressive Emergent Language Scale (REELS), 3-Dimensional Language Acquisition Test (3-DLAT), Reynells Attention Scale, Delayed Speech and Language Development (DSL),

### INTRODUCTION

Agenesis of corpus callosum is a rare cerebral malformation in which there is a complete or partial absence of corpus callosum. Its incidence rate accounts for 0.7% to 5.3% per 100,000 cases<sup>1</sup>. The exact cause of agenesis of corpus callosum (AgCC) is usually idiopathic, but various other genetic and non-genetic factors like Chromosomal mutations, genetic inheritance, and prenatal infections are also the prime causes. Those individuals with Agenesis of Corpus callosum confronts difficulty in understanding literal and non-literal aspects of language and social cues, delayed motor milestones, sensory issues (eg; visual impairments, defective sensory perception and discriminations), bradykinesia, clumsiness and poor motor coordination, Central auditory processing disorders, hypotonia, distorted head or facial features, spasms, emotional swings and seizures. Other disorders of the corpus callosum include dysgenesis, (i.e.; corpus callosum is malformed or incomplete) and hypoplasia, (i.e.; corpus callosum is thinner)<sup>2</sup>. The specific symptoms depend on the site of lesion and extent of damage to corpus callosum. Multidisciplinary team approach plays a crucial role in bringing about diagnosis and intervention for the child with agenesis of corpus callosum.

### CASE REPORT 1

A 11 months old baby boy, belonging to a nuclear family reported with the chief complaints of bilateral decrease in hearing and delayed motor and speech milestones which were insidious and progressive in nature. A significant perinatal history revealed delayed birth cry which sustained for an hour and admission in Neonatal Intensive Care Unit (NICU) for 24 hours. However, a full term normal delivery (FTND) was reported with the birth weight of 2.5kg. No other notable ailment and congenital deformities in either of paternal or maternal family was delineated. Overall test findings and observations on Developmental Screening Test (DST) revealed chronological age of 10 months 27 days and developmental age of 3 months with development quotient of 36.08 suggestive of moderate deficit range in developmental functioning. Magnetic Resonance Brain Imaging (MRI) performed with non-contrast T<sub>1</sub> and T<sub>2</sub> W signal intensities in multiple plane flair revealed absence of splenium and partial agenesis of posterior aspect of body of corpus callosum.

### Speech and Language Evaluation

Childs mode of communication was predominately non verbal. Prerequisites for speech and language development such as object permanence, means-end relationship and attention to the sounds were

absent. Detailed OPM examination revealed inadequate lip seal which led to repeated drooling. Tongue movements were laboured and deglutition was slow as compared to the normative. All vegetative skills were inadequate and the child was solely on semi-solid and liquid diet.

Expressive vocabulary consisted of differentiated crying and vocal gestures primarily for hunger and other necessities. During the session, vocalizations of vowels like /a/, /i/ and different vocal signs of pleasure like social smile were observed. Frequent repetitive rotatory movements of hands along with continuous vocalizations were notable during speech examination.

Adequate eye contact was maintained with improper gaze pattern. Child prefers unoccupied and solitary play. Significant difficulty was evident on fine and gross motor skills. On formal testing like Receptive Expressive Emergent Language Scales (REELS) both Receptive and Expressive language Age (RLA and ELA) corresponded to 1-2 months. On 3 Dimensional Language Acquisition Test (3-DLAT), the child's age of comprehension, cognition and expressions were scattered within Group I. Childs attention concurs to level 1 on Reynell's Attention Scale which indicates child can pay attention but any new events are likely to distract.

### Audiological Findings

Otoscopic examination revealed no pathology of outer ear. Behavioural Observation Audiometry (BOA) showed moderate hearing loss as the child responded to different stimulus on a distance of less than 3 feet. On OAE screening, both DPOAE and TEOAE were referred.

Brainstem Evoked Response Audiometry (BERA) done using click stimuli presented at the rate of 21.1/sec with alternate polarity to estimate hearing sensitivity, wave V could be traced till 60 dBnHL in both the ears with good morphology and replicability suggestive of bilateral moderate to moderately severe hearing loss.

### CASE REPORT 2

A 2 year old female child reported to the hospital with the chief complaint of developmental delay and selective hearing loss. A detailed case history revealed post Neonatal Jaundice (NNJ) with exchange transfusion and delayed birth cry. APGAR score was markedly reduced with an outline of 4. Psychological evaluation

showed moderate intellectual disability. All motor and speech milestone were delayed. Moreover, maternal RH-incompatibility with history of spontaneous abortion was divulged. Ophthalmology examination was done to rule out homonymous hemianopsia and was further corrected with the help of spectacles assistance. MRI findings revealed agenesis of corpus callosum with poly micro gyria.

### Speech and Language Evaluation

OPM examination revealed all structures normal in appearance and feeble in function. Mild groping of mandible and tongue was observed. Reduced range and mobility of tongue was seen and severe drooling was present. Child does not follow unidirectional verbogestural command consistently. Prerequisites of speech and language development were spared properly. Reynell's attention scale finding corresponded to an attention skill of level 1 which indicates that the child can pay fleeting attention but any new event will distract. Child comprehends few common objects, understands very simple WH-questions and responds to rhythmical activities. Child expresses her need by vocalizing utterances and mimicking vocal stimulation. On formal test like REELS, RLA of 11-12 months and ELA of 9-10 months were indicated. A scattered result within Group 1 was obtained on 3 Dimensional Language Acquisition Test. Child prefers unoccupied and solitary play.

### Audiological evaluation

Comprehensive audiological test battery approach was conducted to determine hearing loss and other associated auditory disorder of the child. Different attentive and reflexive behaviours were observed during informal assessment which was done using different frequencies of stimuli at higher intensities (like Gadva, clap, wood rattle, damru and palm rattle). In addition, OAE screening revealed bilateral both DPOAE and TEOAE referred suggestive of hearing loss  $\geq 30$ -40 dBHL.

BERA was administered to determine hearing sensitivity with a transient stimulus using rarefaction polarity presented at the rate of 19.1/s which revealed bilateral Mild hearing loss.

### DISCUSSIONS

Corpus Callosum is a thick band of nerve fibres of white matter structure which resides underneath the cerebrum within the interhemispheric fissure. It plays a crucial role in connecting right and left cerebral hemispheres of the brain, thus facilitating transference of motor, sensory, cognitive and linguistic information<sup>3</sup>. Corpus callosum is derived from lamina terminalis in the portion of the neural tube, cephalic to the rostral neuropore which develops typically between 12 to 20 weeks of gestation period and experiences continuous structural changes even into adulthood. It can be divided into rostrum, genu, body and splenium from anterior to posterior, connecting different lobes of each cerebral hemisphere. Any intrinsic and extrinsic factors that interferes with the normal development of corpus callosum during the foetal life results in Agenesis of corpus callosum which can be either partial or complete. It occurs predominantly in males than in females with gender ratio of 2:1. Aforementioned case is an 11 months old baby boy and 2 years old baby girl with partial agenesis of corpus callosum.

Though the exact etiology of partial agenesis of corpus callosum is unknown, a chromosomal genetic mutation might be the probable cause for both the child. In spite of no abnormalities found during karyotyping, current evidences suggest that a combination of genetic mechanisms including single-gene Mendelian, single-gene sporadic mutations and complex genetics may be involved in aetiology of agenesis<sup>4</sup>. Moreover, the absence of splenium due to agenesis of corpus callosum and corresponding areas might be the cause of deficit in speech and language development and hearing impairment for both the cases. In addition failure in integration of visual information between the two hemispheres was the possible cause of homonymous hemianopsia for 2<sup>nd</sup> child. Necessary audiological rehabilitation was recommended immediately following diagnosis of hearing loss for both the patient. Primary priority was given for fitting of binaural hearing aids.

Intensive speech and language therapy was done for both the child. was done followed by other speech and language stimulations using different speech and language strategies like modelling, expansion, extension, parallel and self talk, correction, systemic desensitization, etc...Due to hearing impairment and moderate intellectual disability

for both the child, only an average speech and language output could be expected. Necessary parental counselling and intensive speech and language stimulations at home were the key recommendations for better prognosis.

### CONCLUSIONS

Although precise condition is unknown, agenesis of corpus callosum is a rare genetic condition that occurs among children and typically affects the speech and language development. Other different symptoms are manifested depending on the site of lesion and degree of damage to corpus callosum. Speech Language Pathologist plays a significant role in restoring, adjusting and compensating the development of speech and language in child with agenesis of corpus callosum.

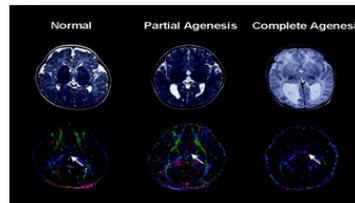


Figure 1.

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