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To study of clinical characteristics of children diagnosed with anomalies of Corpus Callosum

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Abstract

Background: The corpus callosum is the largest interhemispheric connective structure in the brain. Anomalies of the corpus callosum are observed in a variety of conditions that disrupt early cerebral development, including chromosomal and metabolic disorders, as well as intrauterine exposure to teratogens and infection.

Methods: Hospital based study conducted on children attending the Pediatric genetics, Pediatric Neurology OPD and admitted to various wards of Department of Pediatrics, PGIMER

Results: All the cases in the study group were analysed for the presence of seizures and 33.3% children were found to have history of seizures. 6.7% children in study group had significant history of birth asphyxia.

Conclusion: Amongst identifiable causes anomalies of corpus callosum are also associated with genetically inherited syndromes. Identification of these syndromes / disorders by clinical history, examination and appropriate imaging and laboratory evaluation enables appropriate genetic counseling and antenatal counseling of the parents.

Keywords: CC, Syndrome, Children

Introduction

The corpus callosum is the largest interhemispheric connective structure in the brain . Anomalies of the corpus callosum are observed in a variety of conditions that disrupt early cerebral development, including chromosomal and metabolic disorders, as well as intrauterine exposure to teratogens and infection . Recently, a gene defect, haploinsufficiency of AKT3, was also reported, which can be a cause of microcephaly and agenesis of the corpus callosum. Callosal anomalies are often accompanied by other central nervous system (CNS) and/or somatic anomalies. Szabo et al. studied the birth prevalence and clinical spectrum of corpus callosal anomaly patients in Hungary. Glass et al. researched the prevalence and demographic risk factors of callosal anomaly in patients over 20 years in California. They reported that patients with corpus callosal anomalies had other associated CNS and/or somatic anomalies, and more severe callosal anomaly was associated with more functional impairment. [1-4]

Materials and methods

Study Setting: Hospital based

Children attending the Pediatric genetics, Pediatric Neurology OPD and admitted to various wards of Department of Pediatrics, PGIMER.

Study design: Observational, Cross-sectional

Participant: Patients (Children),Confirmed cases with anomalies of Corpus Callosum, detected by neuroimaging were enrolled for the study .

Duration: 18 months (July 2016 to December 2017).

Inclusion criteria

Children with anomalies of Corpus Callosum diagnosed on neuroimaging.

Exclusion Criteria

Participants / Parents refused to participate or to give consent

Statistical Analysis

The study was an observational / cross sectional study and did not involve detailed statistical analysis .In patients who were tested, variables such as age of the patient , IQ level were recorded and genotype / phenotype correlation was done .

Results

Around 1000 patients from NDC clinic and 400 patients from Genetics clinic were followed up over a study period of 18 months (July 2016 to December 2017) and 40- 45 patients were found to have Corpus callosum anomalies out of which 30 patients were enrolled in the study.

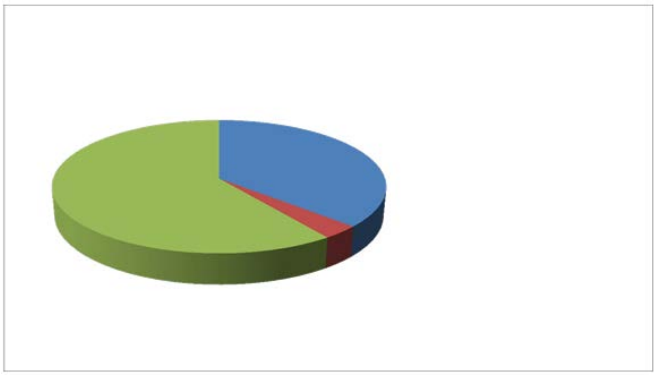
Out of 30 children enrolled in the study 17 were males and 13 were females with varied geographical distribution with maximum (13) number of children coming from Punjab followed by 7 from Haryana, 6 from HP and 2 each from UP and Jammu & Kashmir.

Results

All the cases in the study group were analysed for the presence of seizures and 33.3% children were found to have history of seizures. 6.7% children in study group had significant history of birth asphyxia.

The study revealed that 86.7% children with corpus callosum anomalies had developmental delay in either of the 4 domains (Gross motor, Fine motor, Language, social adaptive).

Out of 30 children only 3.3 % children had neurocutaneous markers in the form of cafe au lait spots. 73.3 % of the children in the study group had facial dysmorphism



Graph 1

Discussion

he proportion of patients with isolated agenesis of the corpus callosum that showed normal development was 19 out of 26, in a previous study by Mangione et al. . However, in the present study, only one out of 13 cases of isolated agenesis or hypoplasia of the corpus callosum developed normally. This difference may be caused by the use of different definitions of normal development. Mangione et al. defined delayed development as 79 or less on the Development Quotient calculated from Child Developmental Inventory (DQ-CDI), and some patients with convulsion, hypotonia, and strabismus were included in the normal developmental group. However, the definition of normal development in this study was stricter, in that the absence of developmental delay, intellectual

disability, abnormal neurologic findings, and epilepsy was required. A previous study by Szabo et al. [1] that applied criteria similar to that of this study reported that 13 out of 18 patients with isolated agenesis or hypoplasia showed developmental delay, intellectual disability, or epilepsy. Another possibility explanation for the low rate of normal development in this study is that we only recruited subjects who visited the rehabilitation clinic in our hospital; not patients with incidental corpus callosal anomaly. Therefore, the results of this study should not be considered to be representative of the prevalence in the general population of Korea.

In patients with CP in this study, other accompanying CNS and/or somatic anomalies showed worse effects on gross motor function, as measured by GMFCS, compared with isolated callosal anomaly. A previous study by Tang et al. [5] also reported that patients with accompanying abnormal sulcal morphology, cerebellar abnormalities, vermian abnormalities, and brain stem abnormalities showed poor neurodevelopmental outcomes. They analyzed the causal relationship of structural anomaly in fetal MR images with functional impairments, and reported that patients with abnormal sulcal morphology and infratentorial abnormalities had poor outcomes. Our study did not try to elucidate the relationship between specific brain structure, and the function of patients with CP. However, patients with other associated CNS abnormalities had worse levels of gross motor function.

Conclusion

Amongst identifiable causes anomalies of corpus callosum are also associated with genetically inherited syndromes. Identification of these syndromes / disorders by clinical history, examination and appropriate imaging and laboratory evaluation enables appropriate genetic counseling and antenatal counseling of the parents.

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