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To study the prevalence of associated syndrome in children with anomalies of Corpus Callosum

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Abstract

Background: The corpus callosum (CC) is the largest of three forebrain commissures in humans. Agenesis of the corpus callosum is caused by abnormality of fetal brain growth between the 3-12weeks of pregnancy.

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: Possible autosomal dominant syndrome and Possible autosomal recessive syndrome seen in 26.67% children.

Conclusion: Common syndromes associated with anomalies of CC identified in the present study were Aicardi syndrome, Frontonasal dysplasia and Dandy Walker related syndromes

Keywords: CC, Syndrome, Children

Introduction

The corpus callosum (CC) is the largest of three forebrain commissures in humans. Agenesis of the corpus callosum is caused by abnormality of fetal brain growth between the 3-12weeks of pregnancy. The possible causes may include chromosomal abnormalities, inherited genetic factors, prenatal infections or injuries, exposure toxins, structural blockage by cysts and also metabolic abnormalities. The estimated prevalence of CC anomalies is 0.3% to 0.7% in patients undergoing neuroimaging in the general population but 2% to 3% in a developmentally abnormal cohort. In many cases, there are associated with genetic disorders which are being increasingly characterIsticssome syndromes which frequently manifest with anomalies of CC are acrocallosal syndrome; Aicardi syndrome, septo-optic dysplasia etc.[1-3]

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

Table 1: Diagnostic categories in patients with corpus callosum anomalies (n=30)

Category	Frequency	Percentage
Possible chromosomal syndrome	4	13.3
Multiple malformation syndrome	6	20
Possible autosomal dominant syndrome	8	26.7
Possible autosomal recessive syndrome	8	26.7
Other / Unknown disorder	4	13.3
Total	30	100

Discussion

In the recent study done by Hashim et al, they retrospectively reviewed 125 patients with corpus callosum abnormalities and found that the presence of cardiac disease was frequently (21%) associated with anomalies of CC. Children under our study were screened for the presence of additional malformations and it was observed that 16.7% had digital malformations and 3.3% had cardiac malformations .Renal malformations were found in 6.7 % whereas skeletal malformations were seen in 6.7% of cases. Therefore screening for additional malformations should be done in all the cases of anomalies of CC especially when additional malformations are present or if there is significant growth retardation.

They also found that in 37% (46/125) of the patients the cause of anomalies of CC was identifiable. In the present study it was found that in 86.7 % cases the clinical cause was identifiable: of these 13.3% had possible chromosomal disorder, 20% had multiple malformation syndrome, 26.7% had a possible autosomal dominant disorder and 26.7% had possible autosomal recessive disorder. In autosomal dominant syndromes, the recurrence risk is 50% and autosomal recessive disorder has recurrence risk of 25%. If we know the inheritance pattern of the identified

syndrome, appropriate antenatal counseling can be offered to the parents.

Conclusion

Common syndromes associated with anomalies of CC identified in the present study were Aicardi syndrome, Frontonasal dysplasia and Dandy Walker related syndromes

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