

Spectrum of Prenatally detected Callosal Abnormalities

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INTRODUCTION

- Corpus callosum (CC) is the main junction between the two cerebral hemispheres. It consists of roughly 180 million neuron
- The CC extends frontal lobe to quadrigeminal plate
- It has three parts Rostrum, Genu, Body & Splenium.
- It concerns with Learning cognition attention span , judgement and memory
- Prevalence of callosal abnormalities is
 - 0.3% and 0.7% in the general population
 - 2% and 3% in the developmentally disabled population

Aims & Objectives

- To present sonographic pattern of prenatal callosal abnormalities in Jharkhand region (tribal population).
- To assess other CNS and extra CNS abnormalities associated with callosal abnormalities in a fetus.
- To analyse the genetic evaluation conducted in these fetuses.

Material and Method

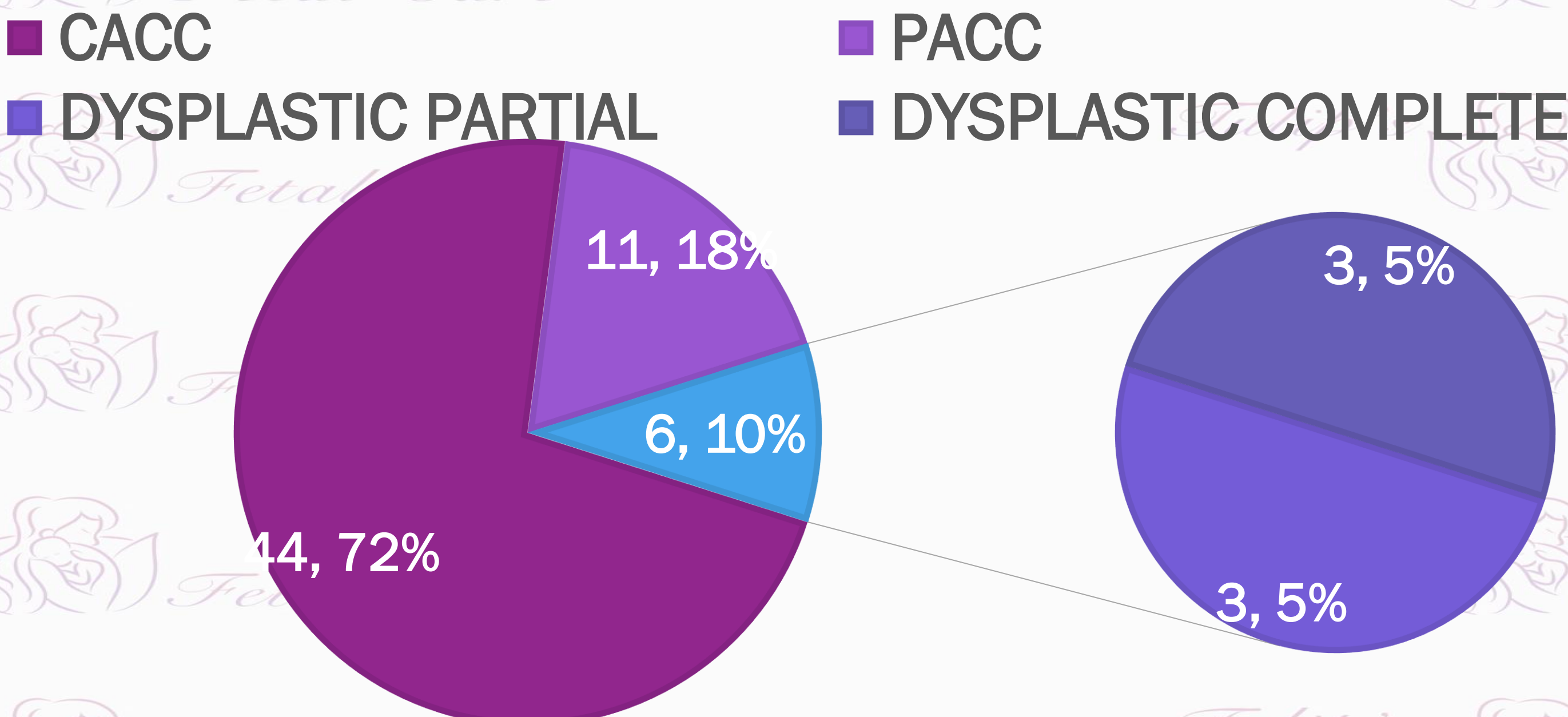
- It was a retrospective study conducted over a period of 8 years and five Maternal / Fetal care units.
- Inclusion – All cases with confirmed diagnosis of Agenesis / partial agenesis / dysgenesis of corpus callosum with 2D and 3D images available for reanalysis.
- Exclusion – Suspected / non confirmed diagnosis, images not available.
- All the cases were examined by experts in fetal medicines with high-end ultrasound systems equipped with transabdominal and transvaginal volumetric transducers
- Data collected – Demographic trends, prenatal screening status, callosal and CSP description, other CNS and extra CNS abnormalities and genetic testing if available.

Results

- 61 cases with Average maternal age 27.5 years 18-45 years
- 59 natural , 2 IVF, 59 singleton, 1 DCDA, 1 MCDA
- Most were primi / 2nd gravidas (22 each)
- Previous affected child / termination of pregnancy was seen in 9 cases.
- Screening for aneuploidies - 9 cases and it was positive for trisomy 21 in 2 cases trisomy 18 in 3 cases and positive for 13,18 and 21 in one case
- There were 44 cases of CACC, 11 cases of PACC and 6 cases of Dysplastic CC
- The CSP was absent in 44 cases , wide in 5 cases, dysmorphic I 10 cases and in 2 cases it appeared normal,

Results

CALLOSAL DESCRIPTION



Extra CNS Malformations

Cardiac 18	Face 13	Skeletal 10	Renal 6	Others 7
Minor (ARSA, EICF, SUA) – 8 Cardiomegaly – 4 Small VSD – 3 DORV (with PS) – 2 HLHS 1 RA/RV dilated - 2	Absent nasal Bone 2 Mid face hypoplasia (Binder's) 1 CLP 4 Micro/retrognathia 3 Ocular defects – 3 Arrhinia - 1	CTEV – 4 Clenched fists with override of fingers – 1 Ectrodactyly – 1 Short long bones – 3 Kyphoscoliosis - 2	Pyelectasis Hypospadias Renal cystic dysplasia Bilateral enlarged echogenic kidneys Pelvic horseshoe kidneys Right renal agenesis, Small left kidney	Early onset FGR- 3 Fetal hydrops (cytic hygroma) – 2 CHAOS – 1 Choledochal cyst – 1 Pleural effusion – 1 Non visualisation of Stomach bubble - 1

Genetic testing 12 cases

Aneuploidies 2

CMA /WES

NORMAL CMA / KT -3

DNA STORAGE 1

VOUS 2

Pathogenic variant 3

Result – survival and postnatal outcome

Continuation of pregnancy was seen in 6cases (6ACC / 2Dysplastic CC/ 1 PACC). IUD at 30 weeks and early neonatal death was seen in 2 cases (CACC), Post birth seizure disorder was seen in one case (dysplastic CC).

Other CNS Malformations

Ventriculomegaly	Supratentorial 6	Posterior fossa 17/61
Lateral ventricle only 26/61 Polyventriculomegaly 6/61	Disorders of cortical migration 3/61 Peri ventricular leukomalacia 1/61 CP cyst 1/61 Pericallosal Lipoma 1/61 Thickened fornixes 1/61	DWM 8/17 Mega cisterna Magna 3/17 Cerebellar hypoplasia 6/17

Discussion & Conclusion

- Fetal Callosal abnormalities can be detected by expert fetal ultrasound at mid- trimester scan.
- One should be watchful for indirect signs of ACC which can be easily demonstrated in axial views.
- Other system survey should also be done to rule out syndromic associations.
- More emphasis on prenatal / postnatal genetic work up
- Larger studies with long term follow up of survivors is needed in