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PICTORIAL ESSAY OF CONGENITAL ANOMALIES OF BRAIN AND SPINAL CORD

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ABSTRACT
Nightmares along with happy dreams is the scenario of parents of congenital anomalies child. Antenatal diagnosis of congenital anomalies need high index of clinical suspicion. If suspected on ultrasound, further investigation be should be carried out to find out any chromosomal defects. Genetic counselling plays crucial role in chromosomal anomalies. Here we present a spectrum of congenital anomalies of brain and spinal cord at various stages of development.

Keywords: Congenital Anomalies, Malformations

INTRODUCTION:

Congenital anomalies of brain are commonly encountered in day to day practice., diagnosing it correctly is of paramount importance.

Congenital malformations of brain can be broadly categorized into disorders affecting:

NEURULATION:

Dorsal induction:

Primary neurulation:

- Myelomeningocele
- Chairi type malformation

Secondary neurulation:

- Diastometamyelia
- Meningocele
- Lipoma
- Dermal sinus with / without cyst
- Tethered cord /Tightfilumterminale
- Caudal regression syndrome

Ventral induction:

Dandy Walker Malformation
Joubert's syndrome

NEURONAL PROLIFERATION, DIFFERENTIATION, HISTIOGENESIS:

Neurocutaneous syndromes –

- NF 1 NF 2.
- Tuberous sclerosis.

Congenital vascular malformations –

- Developmental venous anomaly

Cavernoma
Embryonal neoplasms
Aqueduct stenosis.
Arachnoid cyst
Microcephaly
Megalencephaly

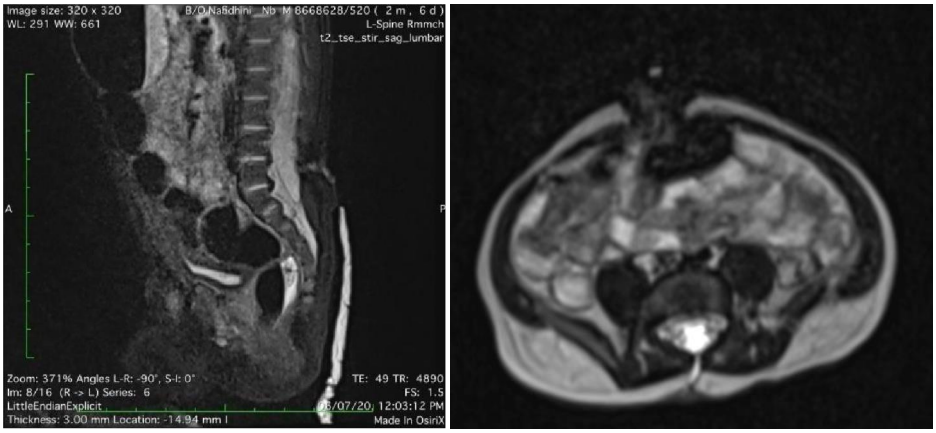
NEURONAL MIGRATION:

Cortical dysplasia
Band heterotopia
Schizencephaly
Patchy/polymicrogyria
Corpus callosal agenesis with/without lipoma

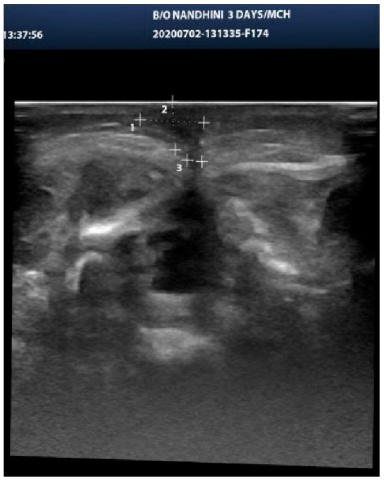
METABOLIC, DEMYELINATION, ENZYME DISORDERS
ACQUIRED DEGENERATION:

Hydranencephaly
Periventricular leucomalacia
Hemiatrophy
Multicysticencephalomalacia

DISORDERS OF DORSAL INDUCTION-
DORSAL DERMAL SINUS



a b



c

Fig-1: Dorsal dermal sinus

a. Thin T2 hyperintense fluid filled sinus tract 1.6 mm thickness extending from skin surface to posterior thecal sac for a length of 8 -10 mm between spinous process of L5 - S1 vertebrae.
b. Associated small cystic swelling 5 x 3 mm skin surface. Non fusion of L3 L4 L5 spinous process seen. S/O Congenital Dorsal Derrmal sinus with superficial cyst at skin surface and associated spina bifida occulta.
c. USG: anechoic elongated structure extending superiorly from the skin surface into the spinal canal.[1,2,3,4]

DIASTOMETAMYELIA

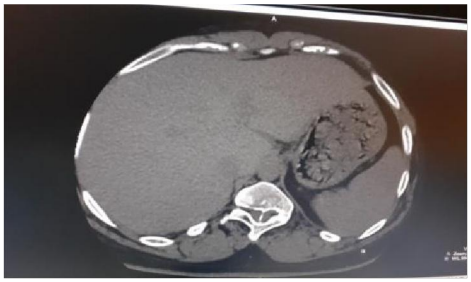


Fig-3: Type 1 Diastometamyelia

Split cord malformation- (type of spinal dysraphism- spina bifida occulta)
Longitudinal split in the spinal cord[5.6 7]
Type 1:Classic,Duplicated Dural sac with common midline spur
Type 2: Single dural sac with no spur

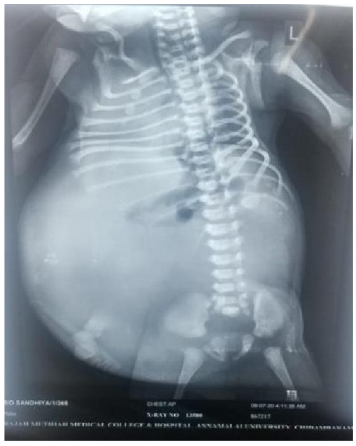


Fig-4: Caudal regression syndrome[8]

Partial agenesis of sacrum and coccyx

MENINGOCELE

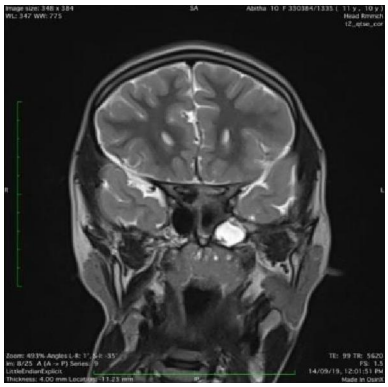


Fig-5: Meningocele

Well defined T2 hyperintense cystic lesion seen in left petrous apex which has communication with left Meckel's cave.[9]

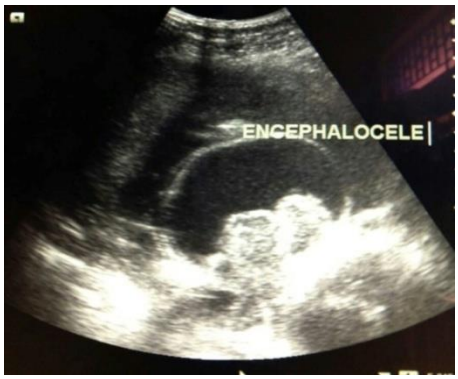


Fig-6: Encephalocele

Cystic mass with herniated brain tissue at cranial end

DISORDERS OF VENTRAL INDUCTION:

DANDY WALKER COMPLEX (includes continuum of Dandy Walker malformation, Dandy Walker variant and mega cisterna magna)[10,11 12]

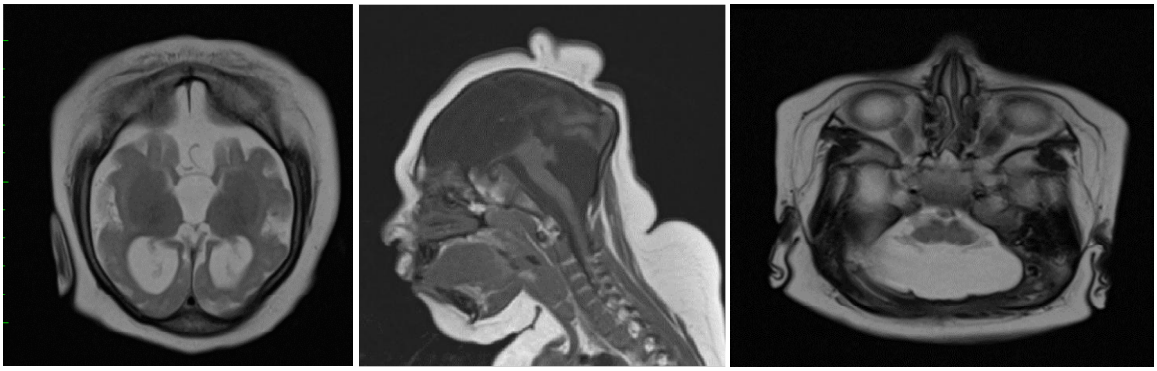


Fig-7: Dandy walker malformation causing obstructive and associated corpus callosal agenesis

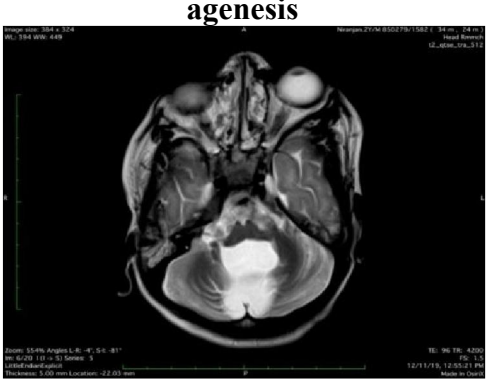


Fig-8: Dandy Walker Continuum.

Superior cerebellar vermian hypoplasia with inferior cerebellar agenesis.
Posterior fossa cyst seen, communicates with fourth ventricle, volume of posterior fossa normal, no torcular inversion seen. S/O **Dandy Walker Continuum.**

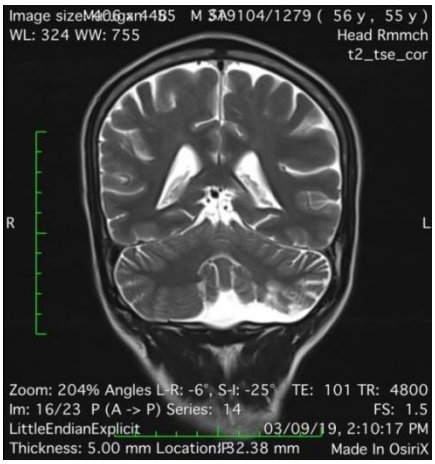


Fig-8:Dandy Walker Variant

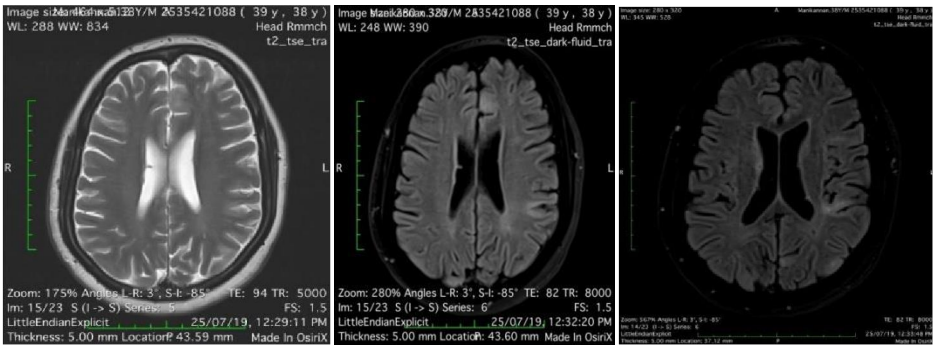
Inferior vermian Hypoplasia with large CSF intensity area in posterior fossa, communicating with 4th ventricle, giving keyhole appearance **S/ O Dandy Walker Variant**

DD POSTERIOR FOSSA CYST:

- ➢ Dandy Walker continuum,
- ➢ Posterior fossa arachnoid cyst,
- ➢ Cystic neoplasms, dermoid, epidermoid and enterogenous cyst.

NEURONAL PROLIFERATION, DIFFERENTIATION, HISTIOGENESIS:
NEUROCUTANEOUS SYNDROMES

TUBEROUS SCLEROSIS[13]



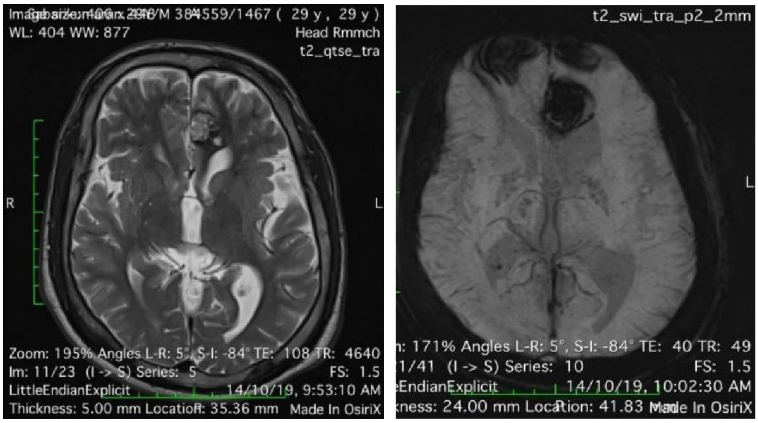
a **b** **c**

Fig-9: Tuberous sclerosis

a. Subependymalhamartomas-Nodular T1 hyper /T2 hypointense B/L subependymal regions,blooming+.

b,c. Cortical Tubers (Calcified)- Cortical, subcortical T2/ FLAIR hyperintensities seen in left medial frontal and parietooccipital lobe. Radial bands - Radial white matter T2 /FLAIR hyperintensities

VASCULAR MALFORMATIONS OF THE BRAIN:
CAVERNOMA

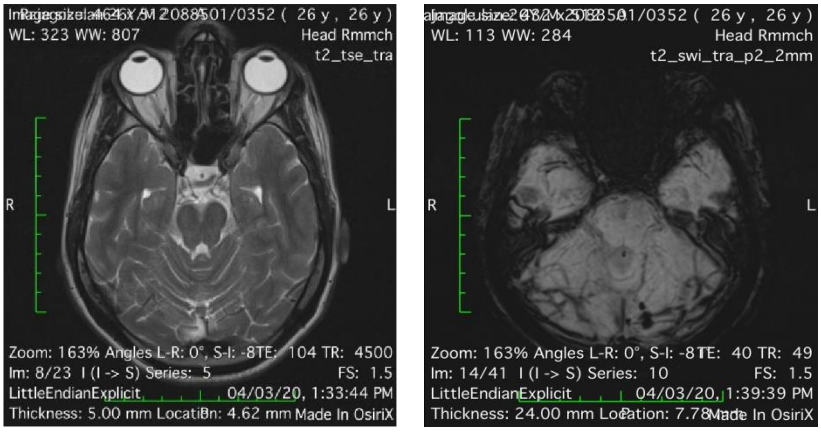


a **b**

Fig-10: Zabramski type II cerebral cavernoma

a. T1 T2 heterointense lesion with internal T1 T2 hyperintensities left inferior frontal lobe (parasagitally),T2 peripheral hypointense rim
b. Blooming +.
To Look for Developmental venous anomaly in post contrast - for surgical planning

Developmental Venous Anomaly (DVA)[14]

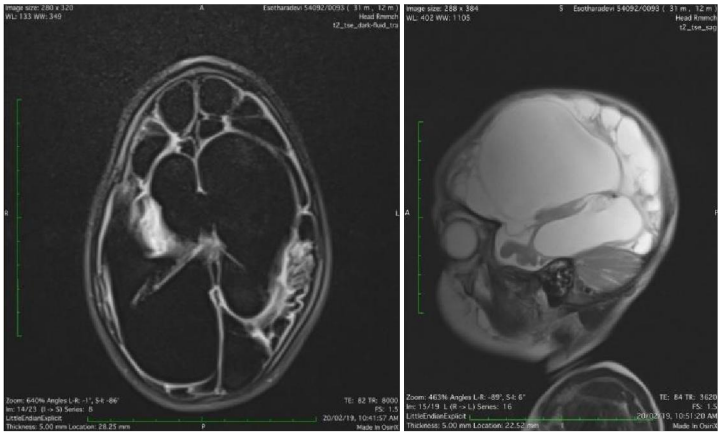


a **b**

Fig-11: Developmental Venous Anomaly

a. Abnormal tortuous superficial cortical vein in left superior cerebellar region,internally drains into left transverse sinus

AQUEDUCT STENOSIS [15]

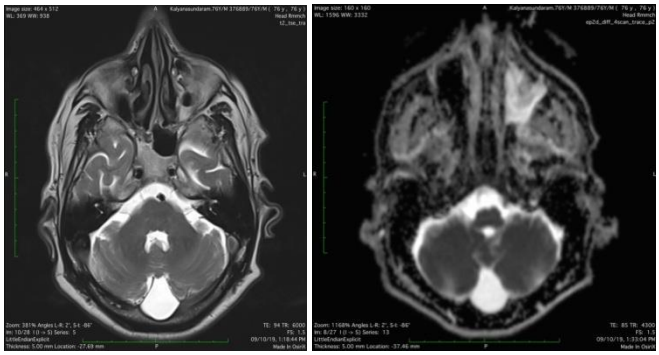


a **b**

Fig-13:Aqueduct stenosis

a) Diffuse enlargement of bilateral lateral ventricles,3rdventricle with mildly dilated 4th ventricle,T2 hyper FLAIR hypointense,Loss of Flow void in T2- narrowing at aqueduct. b) T2 hyperintense cystic changes cerebral cortex with relatively normal appearing gurus around central sulcus -Onstructivetriventricular hydrocephalus due to Aqueduct stenosis with encephalomalacia.

ARACHNOID CYST[16]



a **b**

Fig-14: Retro cerebellar arachnoid cyst

a) T2 hyperintense cystic lesion in retro cerebellar region, b) no Diffusion restriction.

NEURONAL MIGRATION
MALFORMATIONS OF CORTICAL DEVELOPMENT

Fig-15: Band heterotopia / double cortex syndrome with pachygyria

Band of grey matter located deep and roughly paralleling cortex .Gyri appear thickened .

SCHIZENCEPHALY:[17]

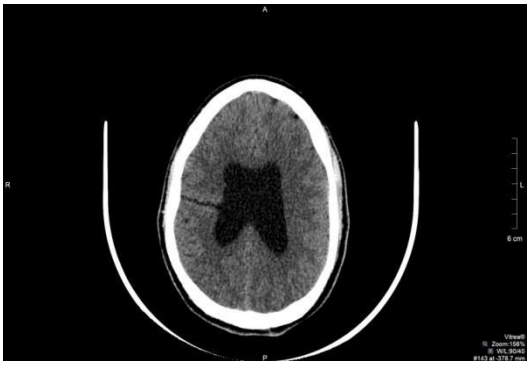
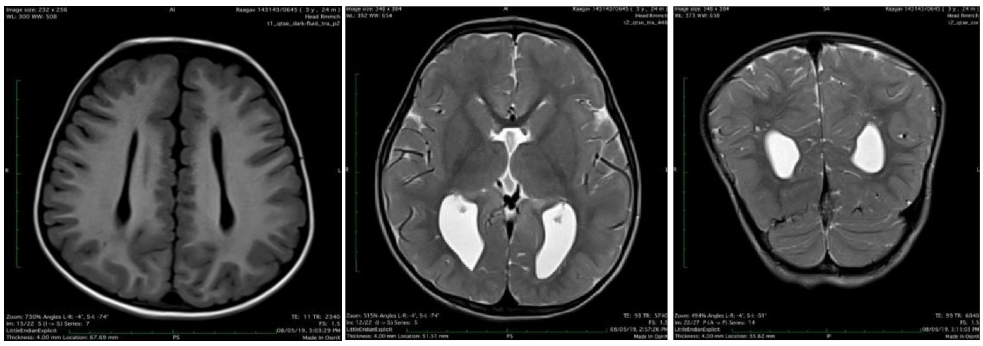


Fig-16: Type 1Schizencephaly

Closed-lip (type 1) schizencephaly is a thin, gray matter lined cleft in parenchyma. Usually the ventricular margin shows an out pouching at the site of closed-lip schizencephaly, acting as an important clue.

Open-lip schizencephaly is a larger, gray matter lined, with obvious defect in ventricular margin. Closest differential of open-lip schizencephaly is porencephaly. But porencephalic cysts are lined by gliotic white matter

CORPUS CALLOSUM ANOMALIES[18]



a **b** **c**

Fig-17: Corpus Callosal agenesis

a) Corpus callosum absent except genu, high riding third ventricle with spoke-like orientation of gyri around it.
b) Lateral ventricles are widely separated, parallel and non-converging.
c) Colpocephaly (dilated occipital horns) and frontal horns thinner and laterally pointing. Lateral ventricles are indented superomedially by the longitudinal white matter bundles (Probst bundles)

CORPUS CALLOSAL DYSGENESIS

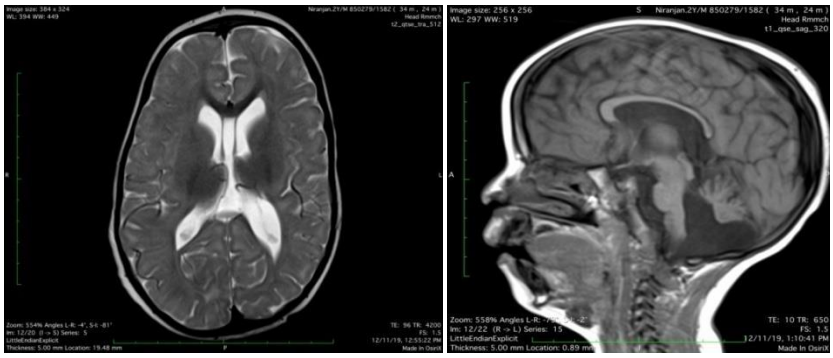
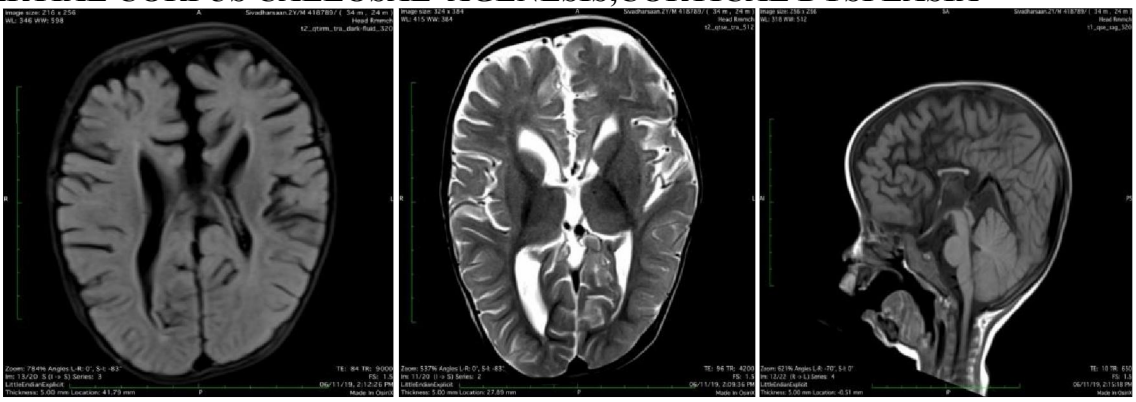


Fig-18: Corpus CallosalDysgenesis

Thinning of body and splenium of corpus Callosum.

PARTIAL CORPUS CALLOSAL AGENESIS,CORTICAL DYSPLASIA



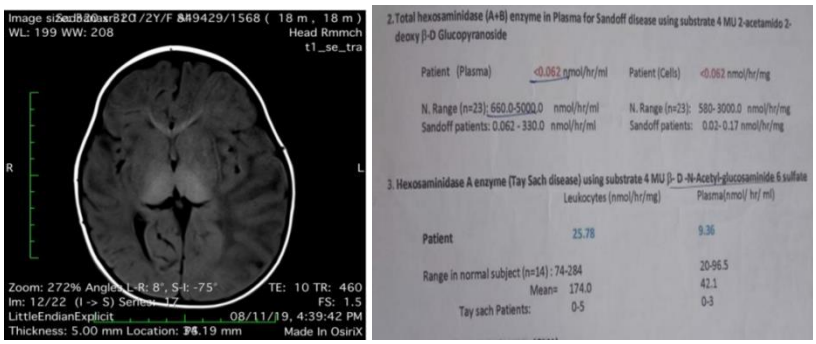
a b c

Fig-19: Partial corpus callosal agenesis (a, b), cortical dysplasia (c)

a) Genu and body – rudimentary, Rostrum, splenium absent
b) Colpocephaly.
c) Some cortices of bilateral frontal, anterior temporal lobe thinned out with paucity of subcortical white matter-S/ O Cortical Dysplasia

METABOLIC - ENZYME DISORDERS

SANDHOFF DISEASE[19]



a b

Fig-20: Sandhoff disease

a) Diffuse T1 hyper T2 FLAIR hypointensities in bilateral thalamus
b) Reduced Total Hexosaminidase levels
DD : TaySach's disease :ventral thalamus hypointense, Dorsal thalamus hyperintense

ACQUIRED DEGENERATION

HEMIATROPHY

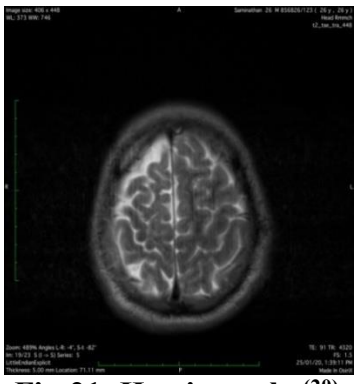


Fig-21: Hemiatrophy[20]

- Diffuse atrophy with volume loss - right cerebral hemisphere
- DD: Dyke Davidoff Mason syndrome

CYSTIC ENCEPHALOMALACIA

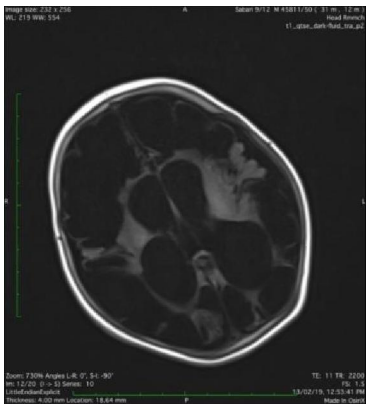


Fig-22: Cystic Encephalomalacia

Supratentorial white matter T1 FLAIR hypo/T2 hyperintensities with exvacuo dilatation of bilateral lateral and 3rd ventricle

CONCLUSION

Neuro imaging of various congenital malformations is essential to assess the prognosis and decide the management .It also helps in planning future pregnancies with prior genetic counselling in selected cases.

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