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RESEARCH ARTICLE

PRENATAL DIAGNOSIS OF NEURAL TUBE DEFECTS USING ANTENATAL ULTRASOUND

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Abstract

Objective: To determine the prevalence and causes of neural tube defects (NTDs) using antenatal ultrasound and clinical history.
Introduction: NTDs are common congenital malformations affecting the central nervous system, with maternal folic acid deficiency being a key risk factor. NTDs are classified as open or closed, with open spina bifida being the most common compatible with life.
Methods: The study included 2730 antenatal patients. Excluded were those with prior NTD diagnoses.
Results: Twenty-two cases of NTDs were identified (prevalence: 0.80%). The most common types were anencephaly (63.6%), open spina bifida (13.6%), encephalocele (9.09%), and myelomeningocele (9.09%). Key risk factors included non-use of folic acid (50%) and a history of congenital anomalies (18%). Discussion: NTDs arise from failed neural tube closure, affecting brain and spinal cord development. Early detection via ultrasound, fetal MRI, and maternal screening can improve diagnosis. In cases compatible with life, further renal and neurological evaluations are recommended.
Conclusion: Antenatal ultrasound is essential for diagnosing NTDs, with MRI used for confirmation. Prevalence in this study was 0.80%, and anencephaly was the most common defect. Folic acid supplementation is critical for reducing NTD incidence.

Introduction:-

- Ntds are the 2nd most common congenital malformations in humans affecting the development of central nervous system
- Multifactorial causes have been described of which most common association is maternal deprivation of folic acid.
- It is divided broadly into 2 types : open and closed. Most of them are life threatening and of them the most compatible with life is closed spina bifida

Materials and Methodology:-

- Patients and treatment:
- Inclusion criteria: all the antenatal mothers who visited radiology department for routine antenatal scans
- Exclusion criteria: previously diagnosed patients.

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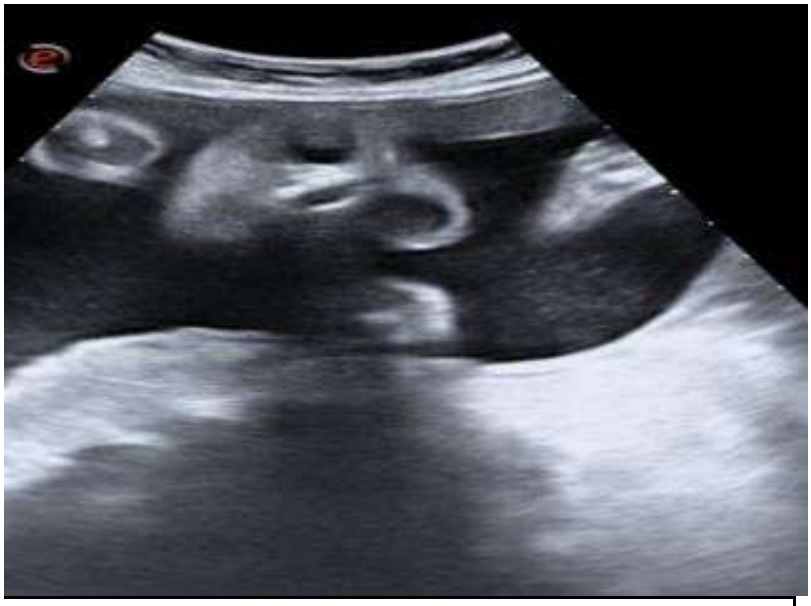
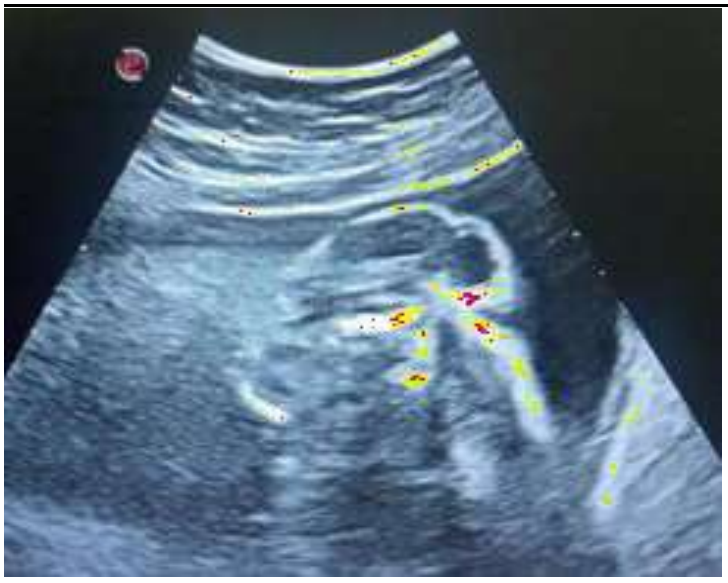
Results:-

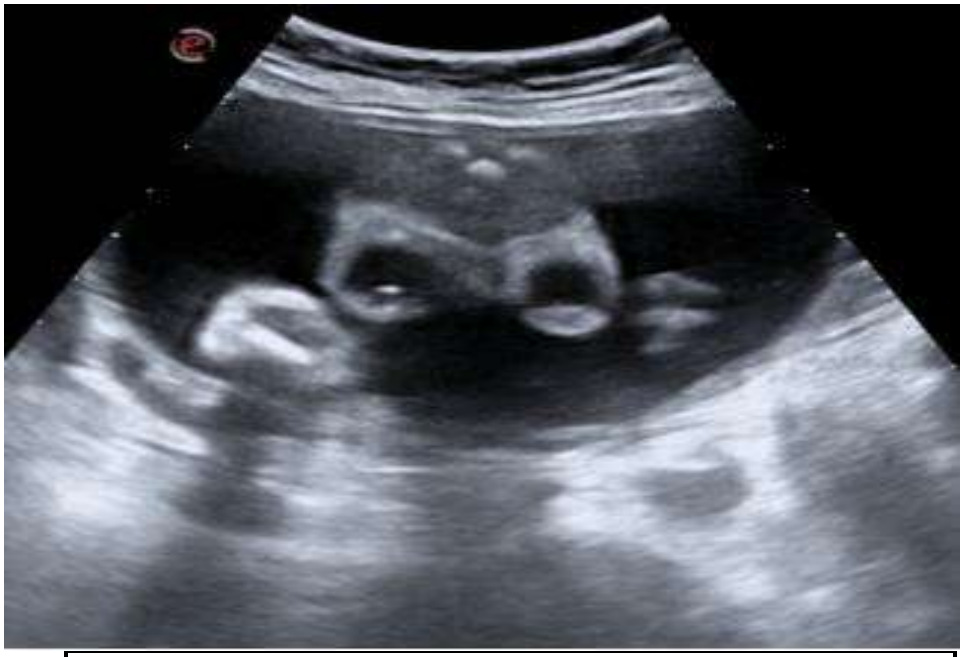
- The study population constituites of 2730 number of patients among whom 22 are diagnosed with neural tube defects in the above mentioned period of time
- Most of them had a history of non usage of folic acid during the 1st trimester of which some of them were not aware of pregnancy until 2nd trimester , few had previous history of neural tube defects few of them had a history of consanguineous marriage, others had no specified history



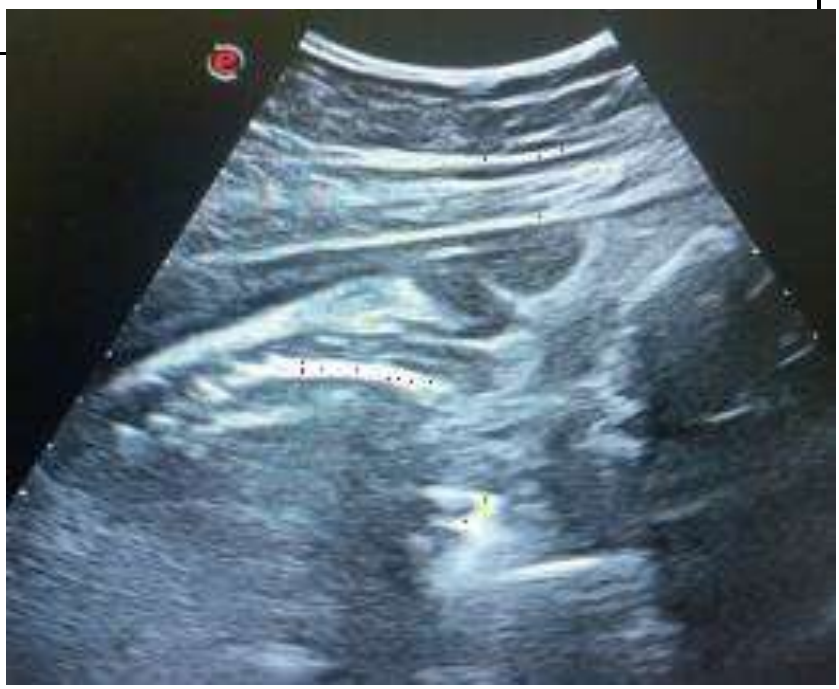
FROG EYE SIGN - ANENCEPHALY

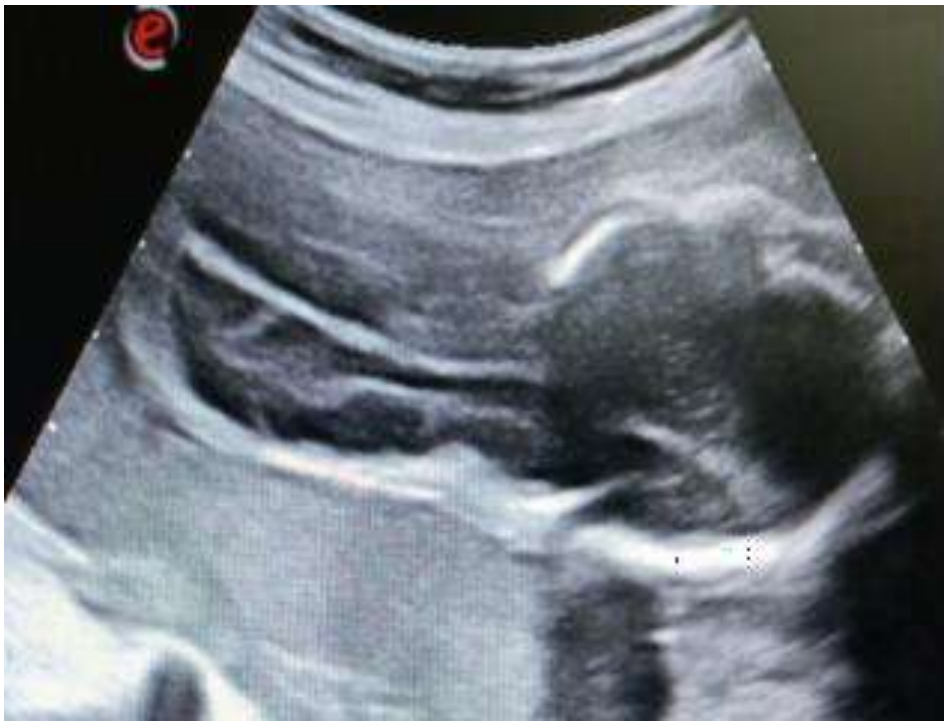


Defect Noted In Upper Cervical Regoin With Hernaition Of Meninges And Neural Elements - Cervical Myelomeningocele.

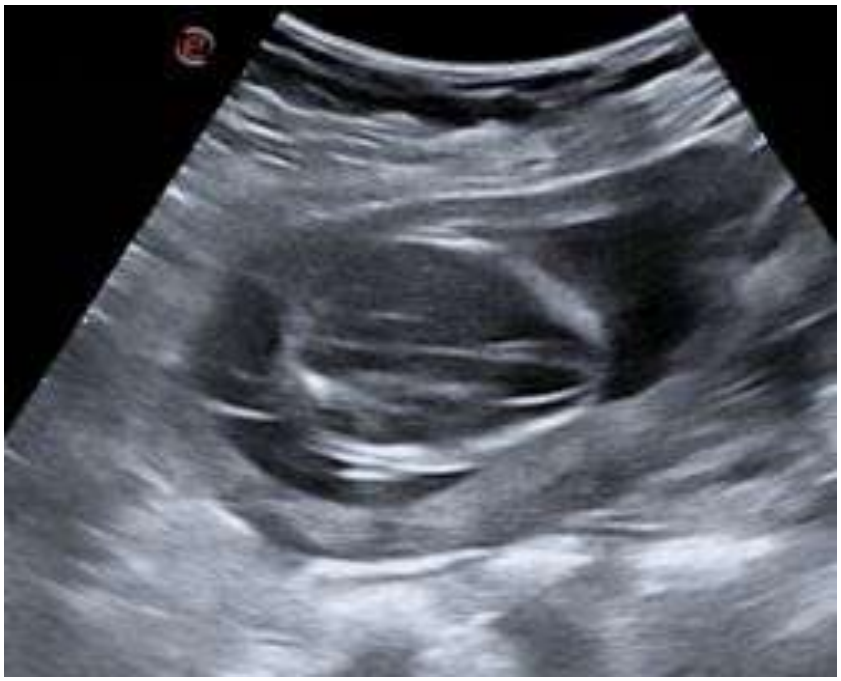


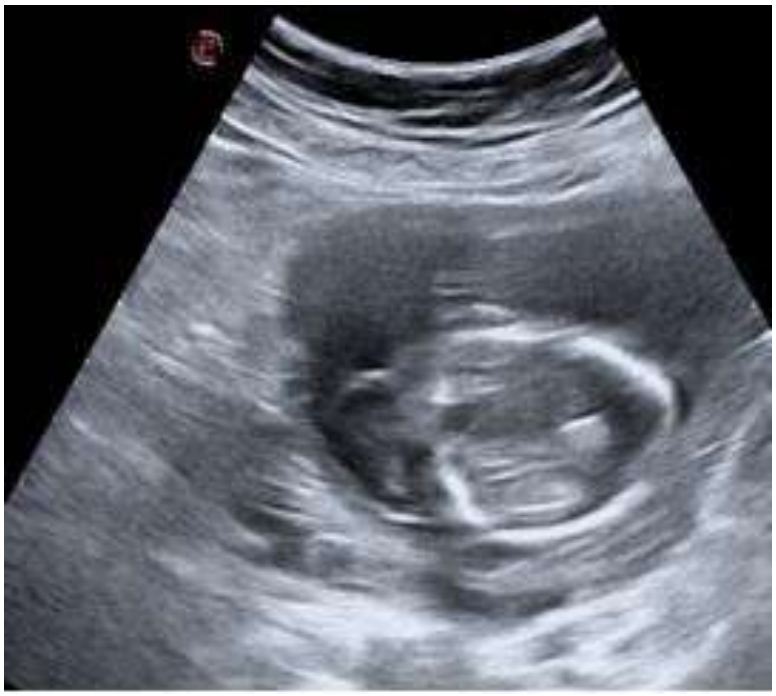
ABSENT CALVARIUM WITH LARGE AMOUNTS OF BRAIN TISSUE PROTRUDING OUTSIDE - **EXENCEPHALY**



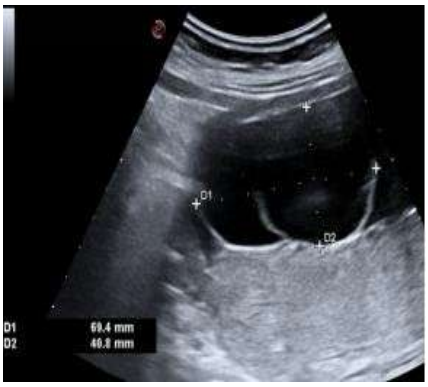


A defect of size 2 cms is noted in the posterior crainial vault with herniation of brain parenchyma – Occipital Encephaloce Le.





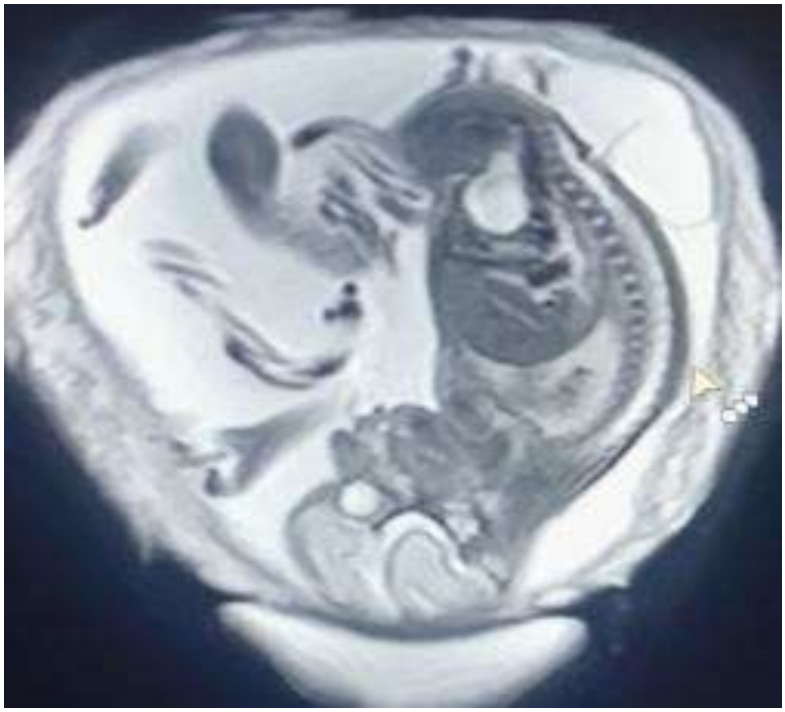
Small Defect Of Size 8mm Is Noted In The Posterior Cranial Vault With Herniation Of Meninges Along With Brain Tissue - Occipital Encephalocel E.



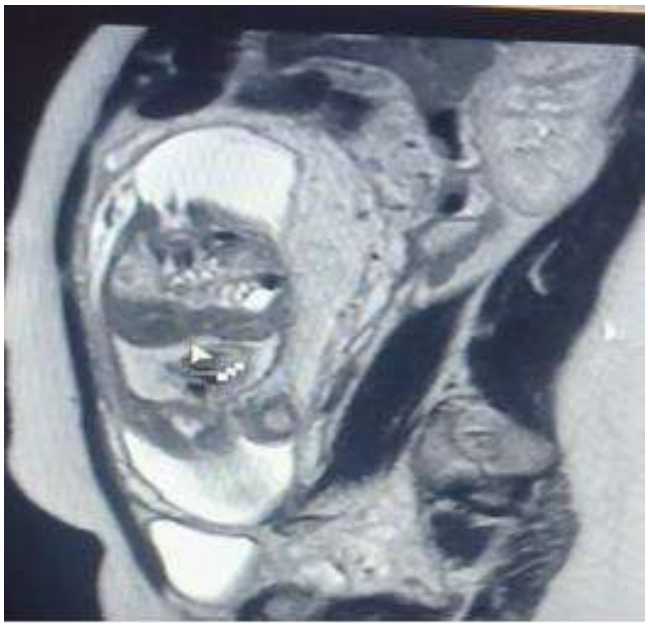
GROSS IMAGE OF THE SAME FETUS SHOWING LUMBAR MYELOMENINGOCELE

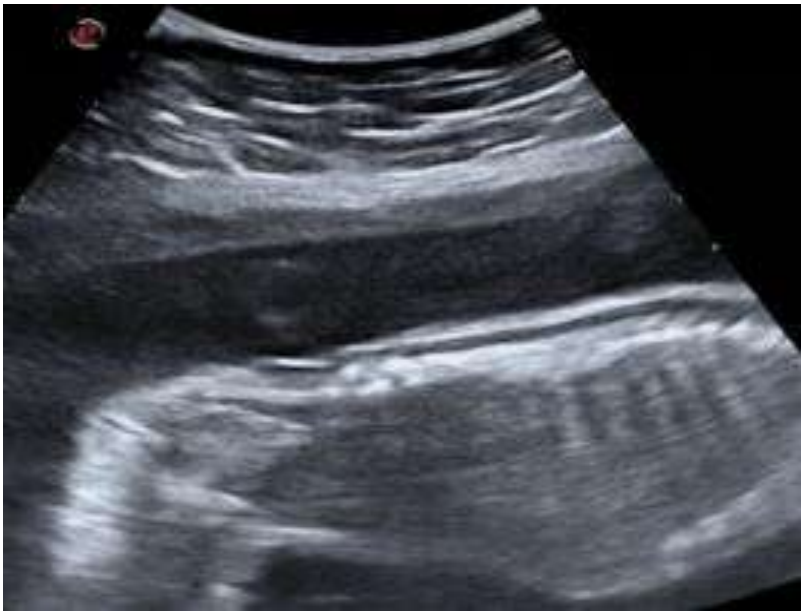
ULTRASOUND IMAGES SHOWING DEFECT OF SIZE 9MM IS NOTED IN THE LOWER LUMBAR WITH HERNIATION OF MENINGES AND NEURAL ELEMENTS

HERNIATED CONTENTS MEASURE 6.9cm* 4.08 cm

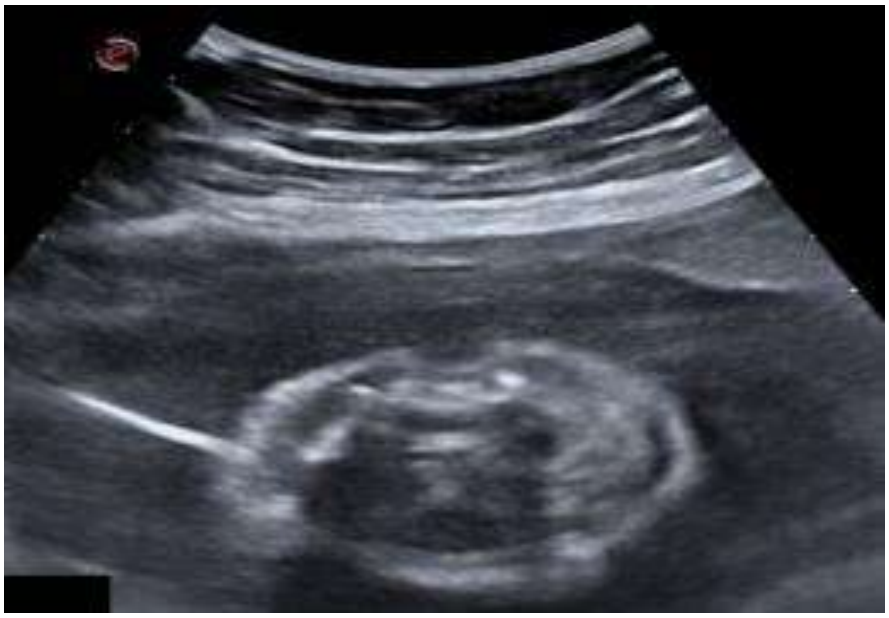


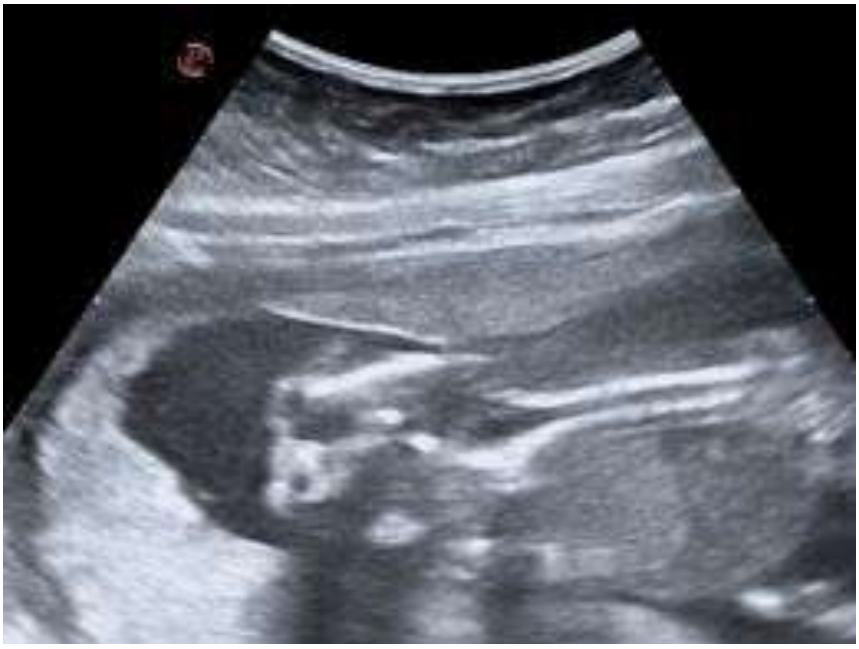
T2wmri Images Showing Defect Of Size 9mm In The Lower Lumbar Region With Herniation Of Meninges And Neural Elements – Lumbar Myelomeningocele.





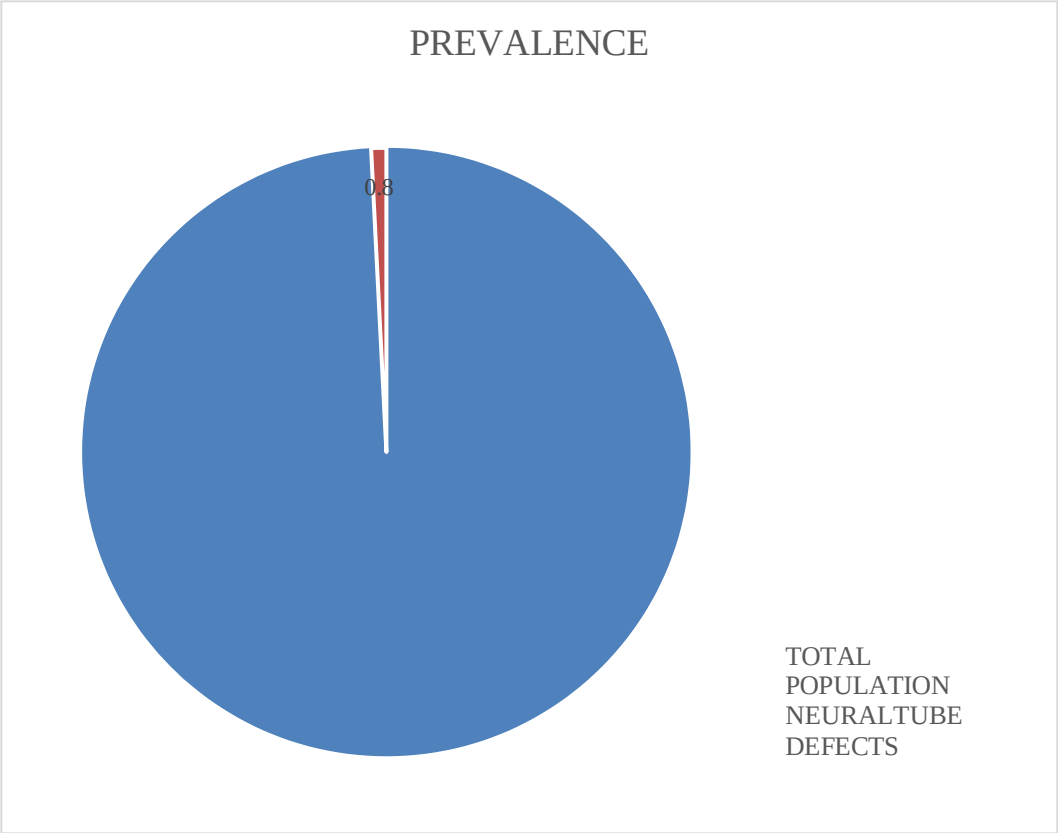
Both Fetuses Show Skin Defect With Incomplete Formation Of Posterior Neural Arch – Open Spina Bifida.

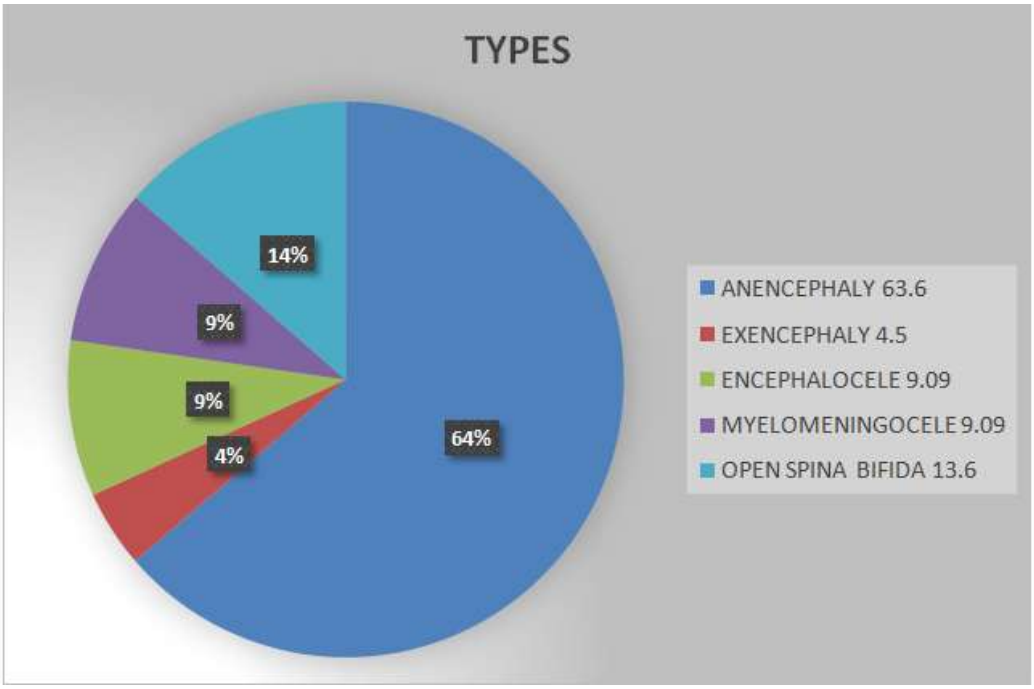




Results:-

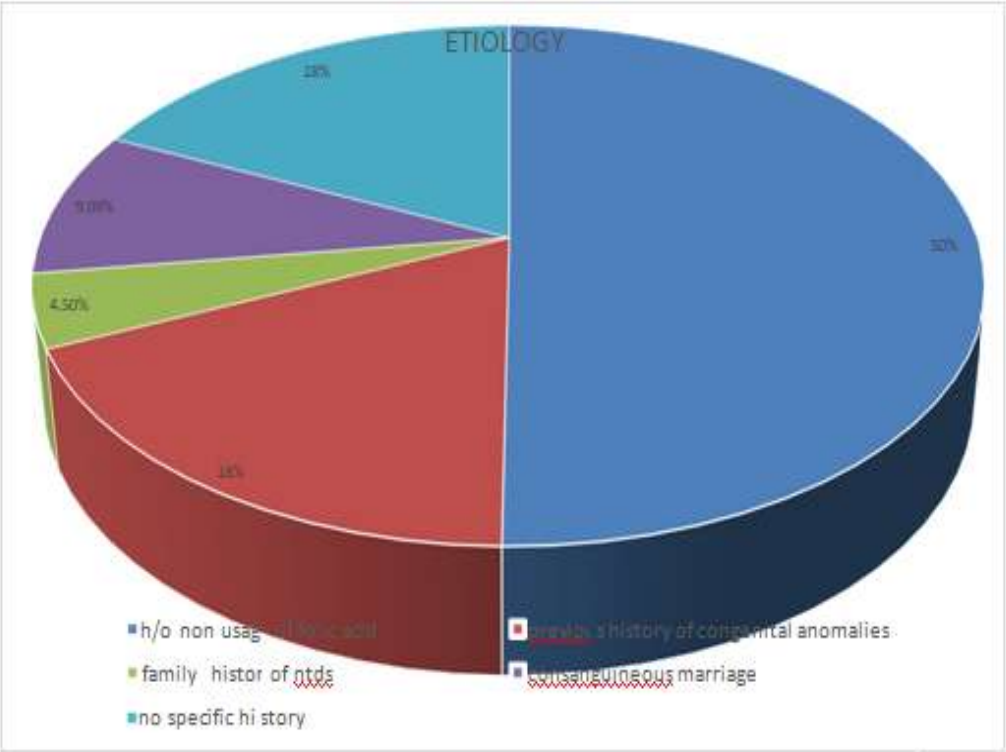
Noof cases	NoofNTDs	percentage
2730	22	0.80





Noofcases	22	%
h/ononusageoffolicacid	11	50%
previoushistoryof congenitalanomalies	4	18%
familyhistoryof ntds	1	4.5%

consanguineousmarriage	2	9.09%
nospecifichistory	4	18%



Type of NTD	No of patients	Percentage
ANENCEPHALY	14	63.6
EXENCEPHALY	1	4.5
ENCEPHALOCELE	2	9.09
MYELOMENINGOCELE	2	9.09
OPEN SPINA BIFIDA	3	13.6

Discussion:-

- Neural tube defects occur in the developing brain and spinal cord. Also the overlying bones are involved
- Broadly classified into:
- Spina bifida open and closed when caudal neuropore fails to close
- Anencephaly when cranial neuropore fails to close
- At day 21 - neurulation starts - neural tube forms and then closes(the neural folds develop and then fuse)
- Fusion of neural tube begins in the mid-line caudal neuropore closes at last
- When all segments not fused it is called craniorachischisis - extremely rare
- Spina bifida - defect in posterior mid-line neural arch
- Anencephaly - when cranial neuropore fails to close
- Spina bifida occulta - vertebral arches of a single vertebra fail to fuse. No skin defect to outside.
- Meningocele - meninges with cerebrospinal fluid are the herniated contents
- Myelomeningocele - Dura, arachnoid, and neural tissue protrude from vertebral canal through Spina bifida defect in posterior mid-line neural arches

- Craniorachischisis - neural folds corresponding to future brain and spinal cord fail to fuse and fail to differentiate, invaginate, and separate from surface ectoderm. Most severe form.

Investigations

- Antenatal ultrasound:
- Following are few important markers to diagnose an open ntd
- The 3 lucencies are suggestive of 4th ventricle, brainstem and cisterna magna
- Brainstem should lie below the maxillo occipital line
- Crash sign - reduced distance between aqueduct and occiput
- Alfa feto protein
- Beta hcg
- UE3
- Fetal MRI
- If born and are compatible with life, then renal tests are suggested such as Ultrasound of KUB Urodynamic studies as neurogenic bladder is common
- Ct brain for hydrocephalus
- Definitive treatment includes surgical repair

Conclusion:-

- Antenatal ultrasound is the 1st investigation which is used to evaluate various fetal cns anomalies, especially neural tube defects
- Findings which are not conclusive on ultrasound can be sent for further evaluation by fetal mri
- Prevalence of ntds according to my study for the given population is approximately 0.80%
- Of them Anencephaly has been reported to be the most common approximately 63.6 % next being open spina bifida 13.6%
- Next most common being the Encephalocele and Myelomeningocele which were 9.09%
- Rest of them such as Exencephaly represent remaining 4.5%

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