world birth defects day march 3

World Birth Defect day

3rd March 2023

CME Program

Organized by:

Department of Obstetrics and Gynecology

Nobel Medical College Teaching Hospital (P) LTD.

affiliated to Kathmandu University

Overview of World Birth Defect Day

Prof Dr Gehanath Baral

Department of Obstetrics and Gynecology

2022 World Birth Defects Day Partners : 2022

14	South East Asia	Nepal	Nobel Medical College and Teaching Hospital, Biratnagar	Gehanafh Baral	baraldr@gmail.com

2022 World Birth Defects Day Partners : 2021

28	South East Asia	Nepal	B and C Medical College Teaching Hospital	Gehanafh Baral	baraldr@gmail.com

Neonate/Infant/Child Mortality trend

Limited public health intervention

Expanded public health intervention

Key facts on Birth defects

- 6% (8 million) born globally each year
- Leading causes of death in infants and young children
- Types: Structural and Non-structural
- 50% of structural cannot be linked to a specific cause
- Known causes or risk factors
  - Non-genetic: can be mitigated or removed before conception or early pregnancy to prevent some birth defects
    - insufficient folate status, poorly managed diabetes, obesity, lack of protection against infectious diseases, some teratogenic medications, smoking, alcohol, and other risk factors.

WBDD

- **The first WBDD: 2015**
  - Aim to provide one global voice and a platform to all organizations and institutions engaged in birth defects related surveillance, research, prevention and care activities.
- **First four years (2015-2018)**
  - Raising awareness about the importance of birth defects.
- **Five-years vision: 2019-2023**
  - 130 organizations formally joined
  - To accelerate **coordinated efforts** to enable organizations and institutions

THE WBDD VISION (2019-2023)

1. **Reduce the occurrence** of birth defects for which there are proven prevention strategies,
2. **Improve quality of life** of all individuals and families affected by birth defects,
3. **Advance knowledge on epidemiology** of birth defects by initiating new monitoring and research programs and strengthening the existing ones to better understand how birth defects impact children, families and communities,
4. **Secure financial and public support** for **prevention efforts and for research** to find causes of birth defects and identify best practices for treatment and care of children with birth defects,
5. **Develop training courses** and/or disseminate the existing ones.

Five-years goal: 2019-2023

1. Raising awareness
2. Mobilizing resources
3. Commitment to improve surveillance, research, prevention and care

Reduce the Risk of Birth Defects

1. Folic acid 400 µg/day
2. Consult before stopping or starting any medicine
3. Vaccines: flu shot and whooping cough vaccine (Tdap) during each pregnancy
4. Pre-pregnancy body mass index [BMI]: ≤30
5. Avoid during pregnancy: Smoking, Alcohol, tobacco, and other drugs

Teratogens

Infections

- Toxoplasmosis (an infection that spreads through cat feces)
- Other infections like group B streptococcus, listeria, candida and sexually transmitted infections (STIs)
- Rubella
- Cytomegalovirus (CMV)
- Herpes simplex virus
- Syphilis

Medications

- Antiepileptic drugs (AEDs)
- Antimicrobials
- Anticoagulants (blood thinners)
- Antithyroid medications
- Vitamin A (a common ingredient in skincare products)
- Hormonal medication

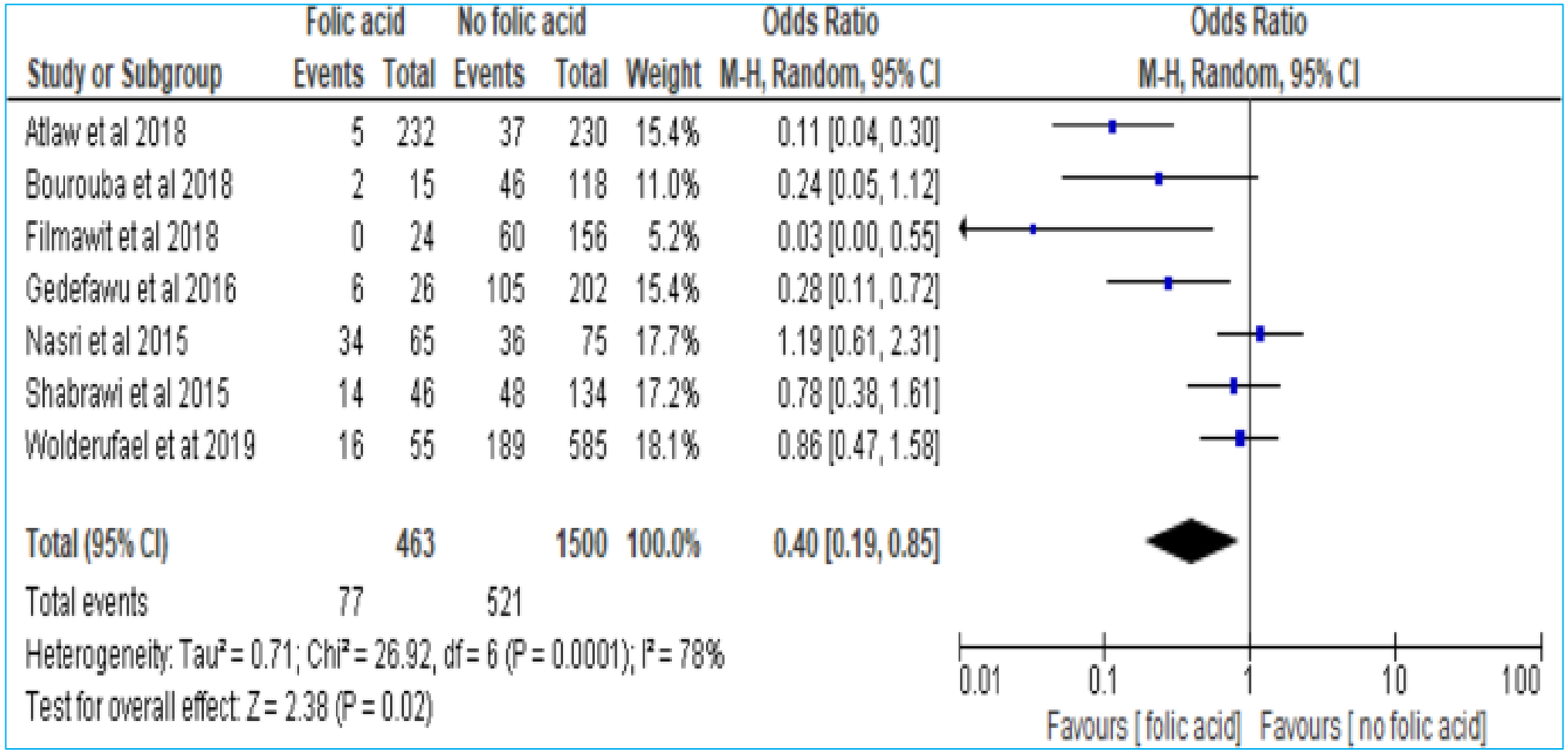
Environmental toxins, chemicals or other physical agents

- Radiation exposure (from X-rays) or chemotherapy.
- Hot tubs, saunas or other heat sources that raise your body temperature.
- Mercury (found in certain types of fish).
- Lead (commonly found in paint and pipes in older homes).
- Toxic chemicals or heavy metals found in the workplace or manufacturing facilities.

Common birth defects teratogens cause

- Brain or spinal cord issues like anencephaly.
- Physical or structural malformation like small bones or missing body parts.
- Cleft lip and palate.
- Cognitive impairment or neurological issues.
- Cardiovascular issues or heart conditions.

Meta analysis of folic acid and NTD



Department of Obstetrics and Gynecology

Dr Pratibha Kaphle

Most common birth defects

Birth defects	
Congenital heart defects	1 in every 110 births
Hypospadias	1 in every 200 births
Ventricular septal defect	1 in every 240 births
Clubfoot	1 in every 593 births
Down syndrome	1 in every 700 births
Pulmonary valve atresia and stenosis	1 in every 1,052 births
Cleft lip with cleft palate	1 in every 1,563 births
Cleft palate	1 in every 1,687 births
Atrioventricular septal defect	1 in every 1,859 births
Limb defect	1 in every 1,943 births

CDC
CENTERS FOR DISEASE CONTROL AND PREVENTION

Burden and consequence in Nepal

- The prevalence of birth defects was found to be 5.8 per 1000 live births
- The commonly occurring birth defects were
  - cleft lip and palate - 6.13%
  - anencephaly - 3.95%
  - clubfeet - 3.95%
  - eye abnormalities - 3.95%
  - meningomyelocele - 3.36%
  - cleft lip - 2.77%

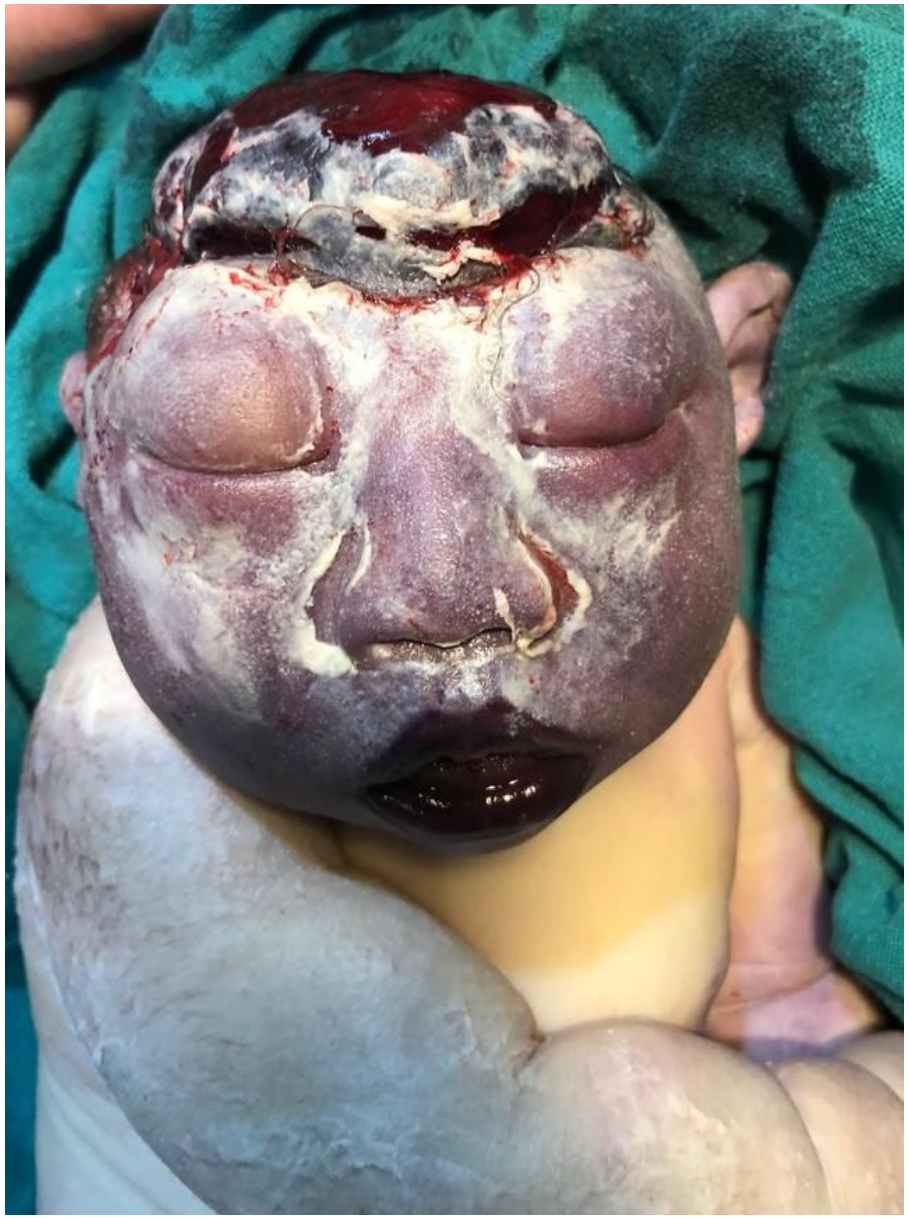
Selected Major Congenital Anomalies

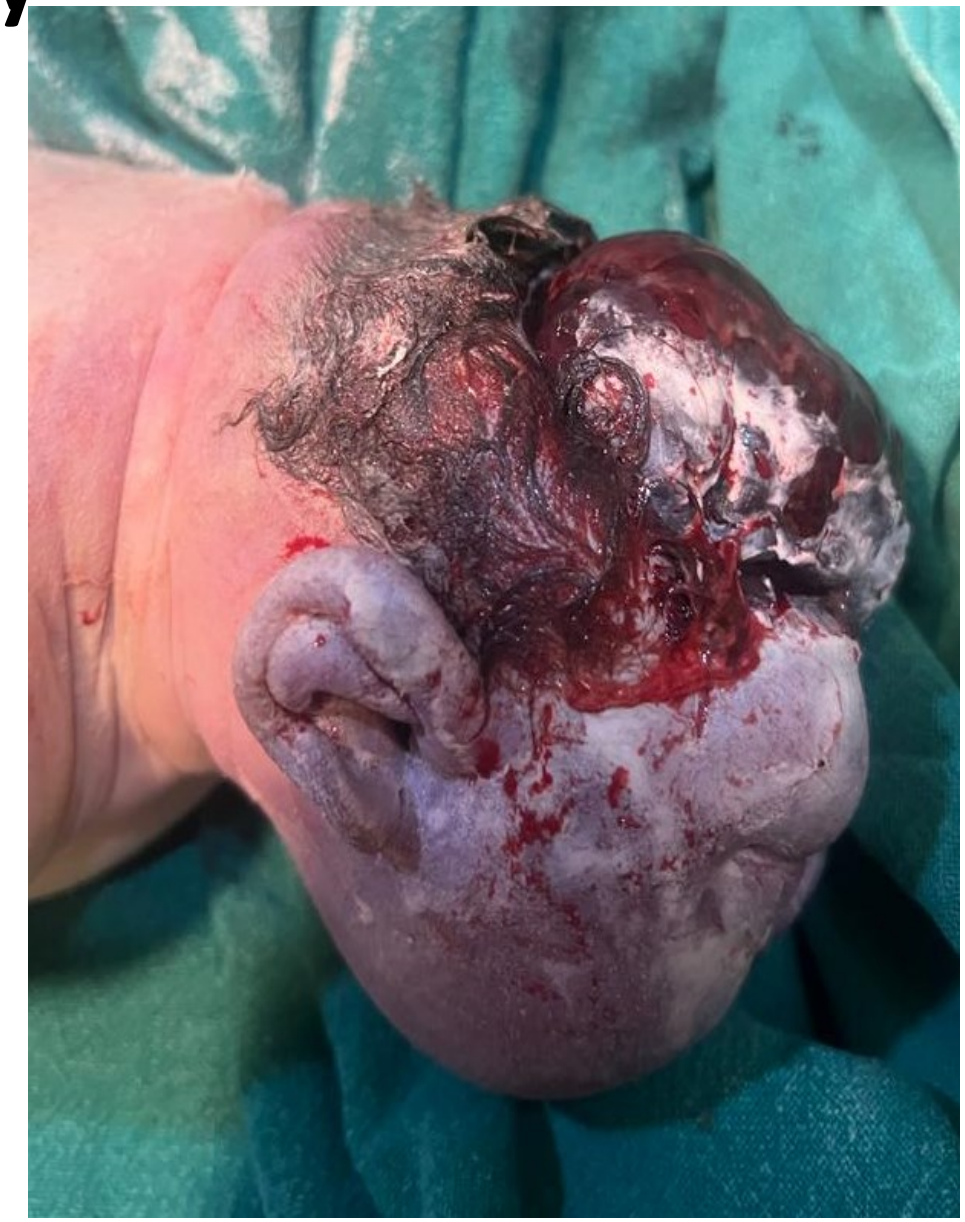
External	Internal
1. Neural Tube Defects Anencephaly, Craniorachischisis, Iniencephaly, Encephalocele, Spina bifida 2. Microcephaly 3. Microtia/Anotia 4. Orofacial clefts 5. Cleft lip only 6. Cleft palate only Cleft lip and palate 7. Exomphalos (omphalocele) 8. Gastroschisis 9. Hypospadias 10. Reduction defects of upper and lower limbs 11. Talipes equinovarus/club foot	1. Congenital heart defects Hypoplastic left heart syndrome Common truncus Interrupted aortic arch Transposition of great arteries Tetralogy of Fallot Pulmonary valve atresia Tricuspid valve atresia 2. Esophageal atresia/tracheoesophageal fistula 3. Large intestinal atresia/stenosis 4. Anorectal atresia/stenosis 5. Renal agenesis/hypoplasia
Chromosomal: Down Syndrome(Trisomy 21)	

Selected external minor congenital anomalies

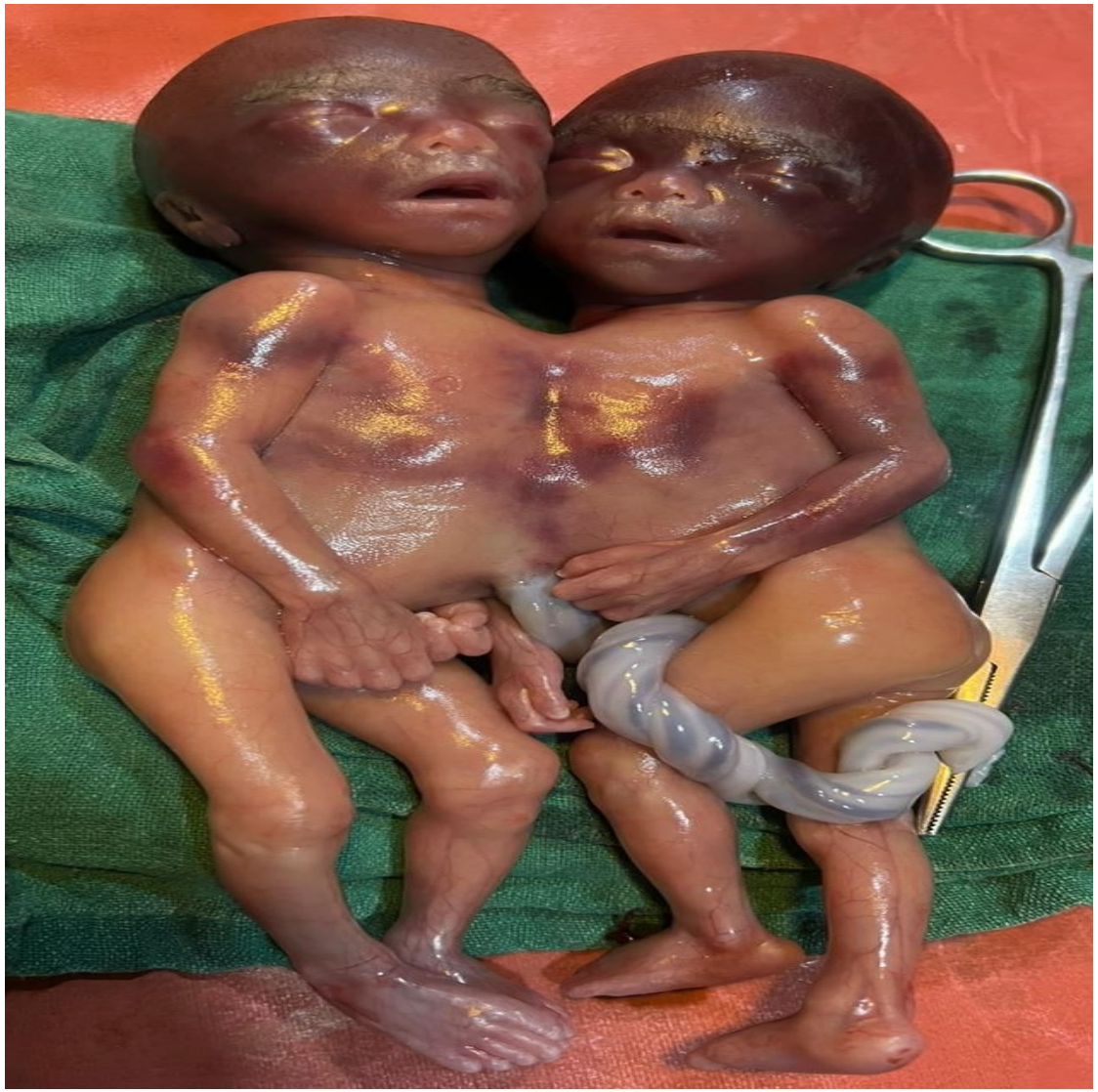
Absent nails	Lop ear
Accessory tragus	Micrognathia
Anterior anus (ectopic anus)	Natal teeth
Auricular tag or pit	Plagiocephaly
Bifid uvula or cleft uvula	Polydactyly , involves hand and foot
Branchial tag or pit	Preauricular appendage, tag or lobule
Camptodactyly	Redundant neck folds
Cup ear	Rocker-bottom feet
Cutis aplasia (if large, this is a major anomaly)	Single crease, fifth finger
	Single transverse palmar crease
Ear lobe crease	Single umbilical artery
Ear lobe notch	Small penis (micropenis)
Ear pit or tag	Syndactyly involving second and third toes
Extra nipples (supernumerary nipples)	
Facial asymmetry	Tongue-tie (ankyloglossia)
Hydrocele	Umbilical hernia
Hypoplastic fingernails toenails	Undescended testicle
Iris coloboma	Webbed neck (pterygium colli)

Anencephaly

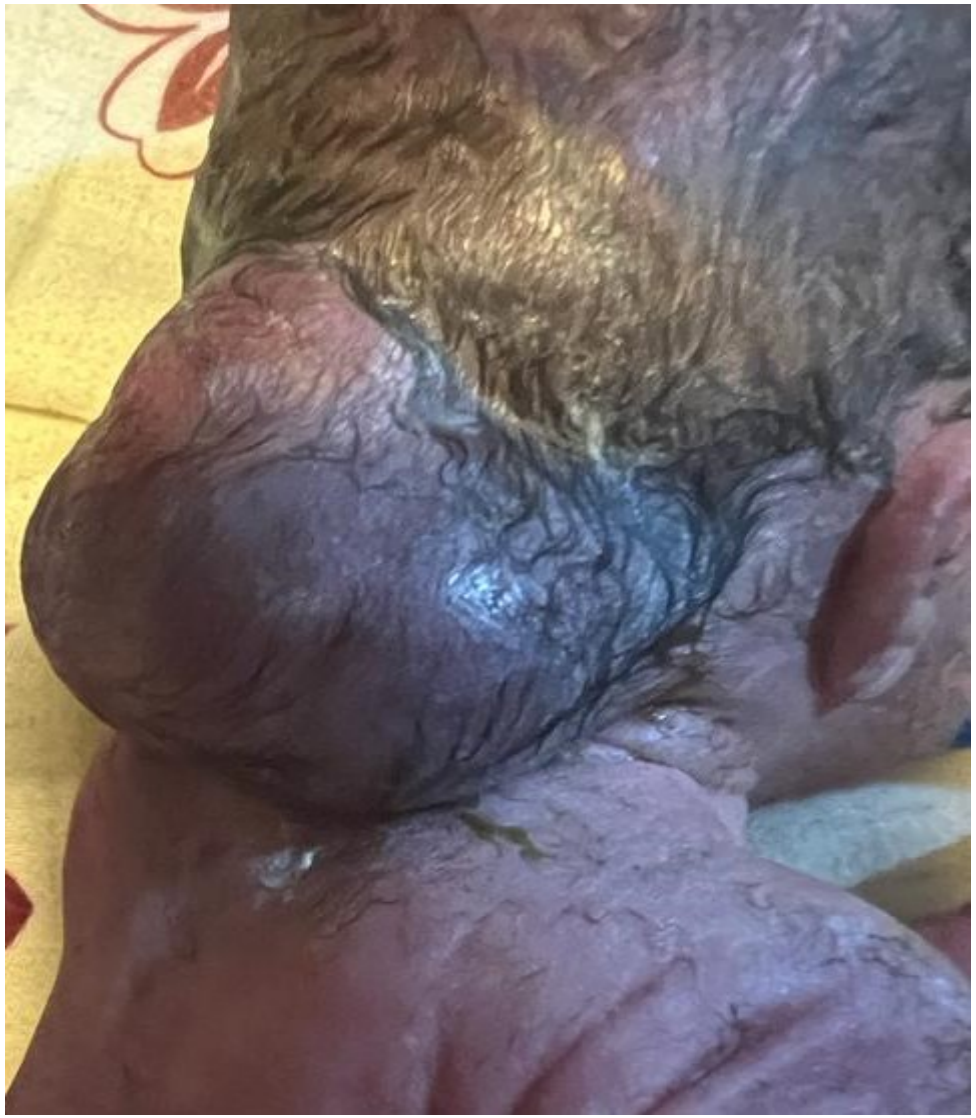




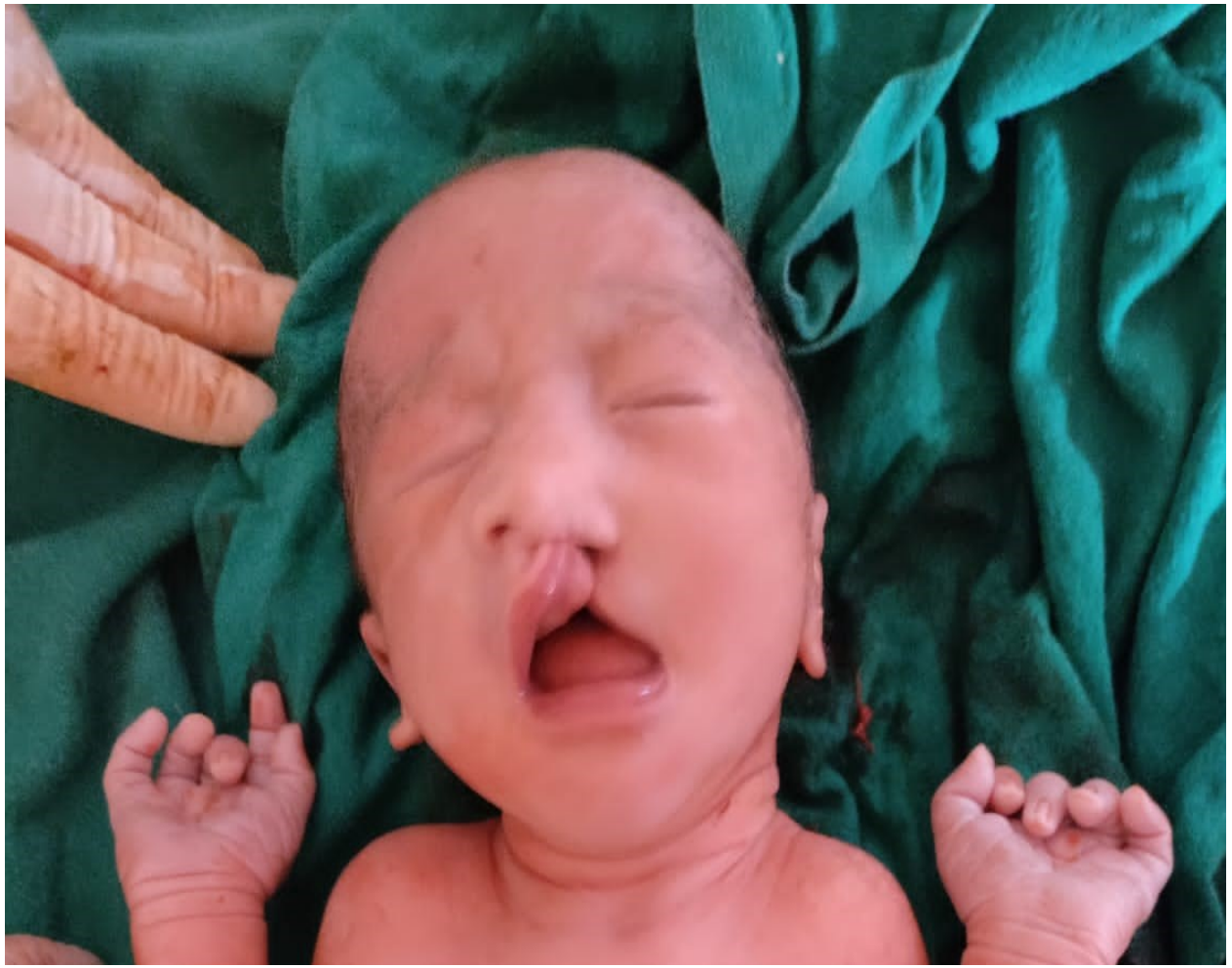
Conjoined twins



Encephalocele



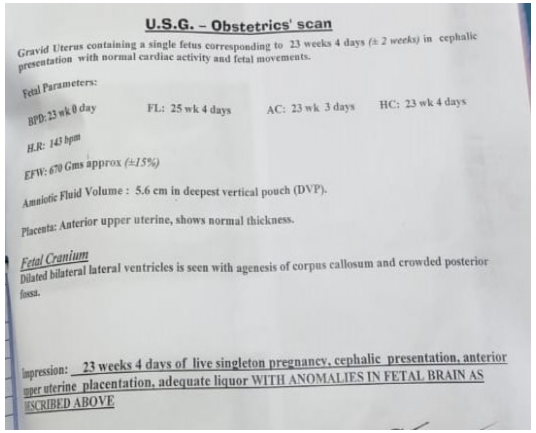
Cleft lip and palate



Clubfoot

Sacrococcygeal teratoma

Hydrocephalus



U.S.G. – Obstetrics' scan

Gravid Uterus containing a single fetus corresponding to 23 weeks 4 days (± 2 weeks) in cephalic presentation with normal cardiac activity and fetal movements.

Fetal Parameters:

BPD: 23 wk 0 day FL: 25 wk 4 days AC: 23 wk 3 days HC: 23 wk 4 days

H.R: 143 bpm

EFW: 670 Gms approx (±15%)

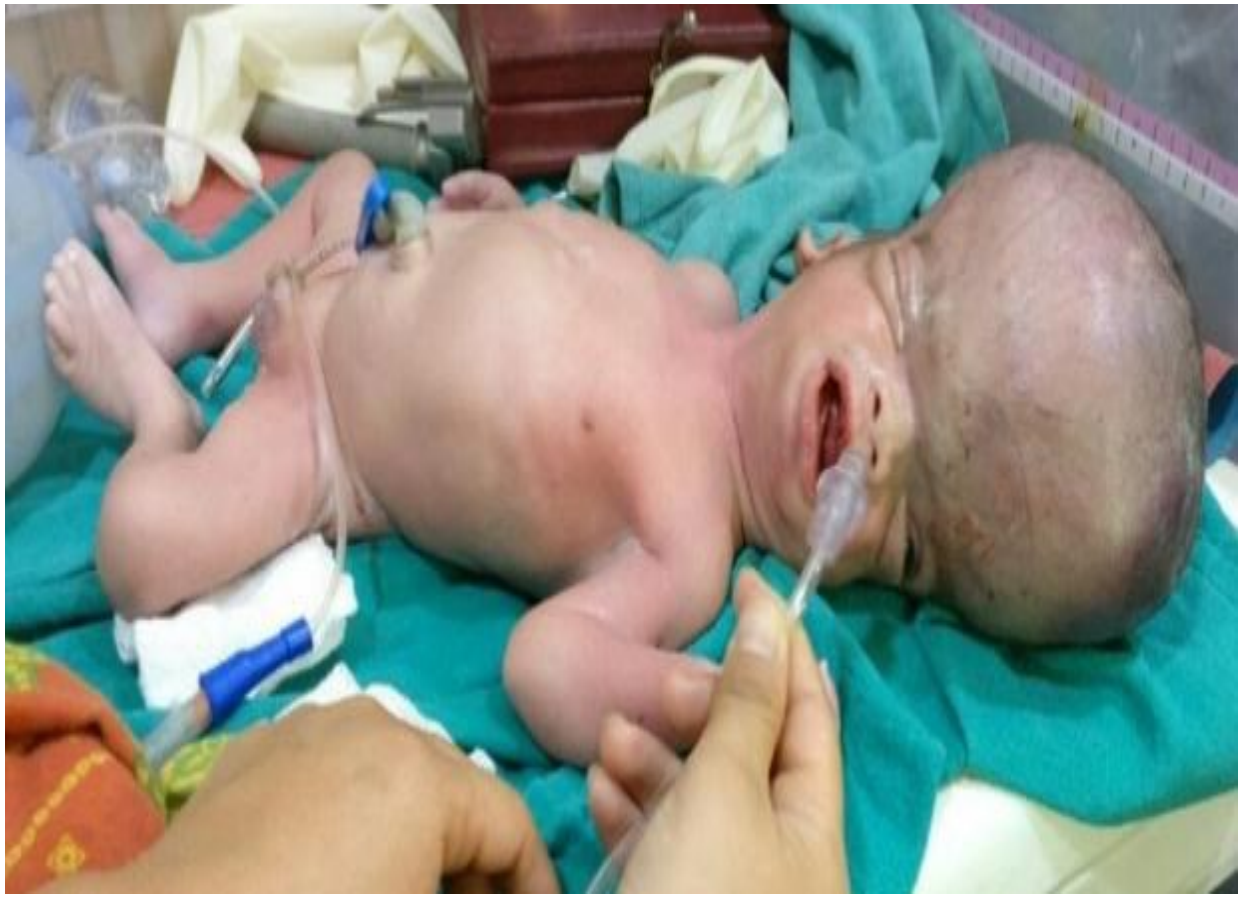
Amniotic Fluid Volume : 5.6 cm in deepest vertical pouch (DVP).

Placenta: Anterior upper uterine, shows normal thickness.

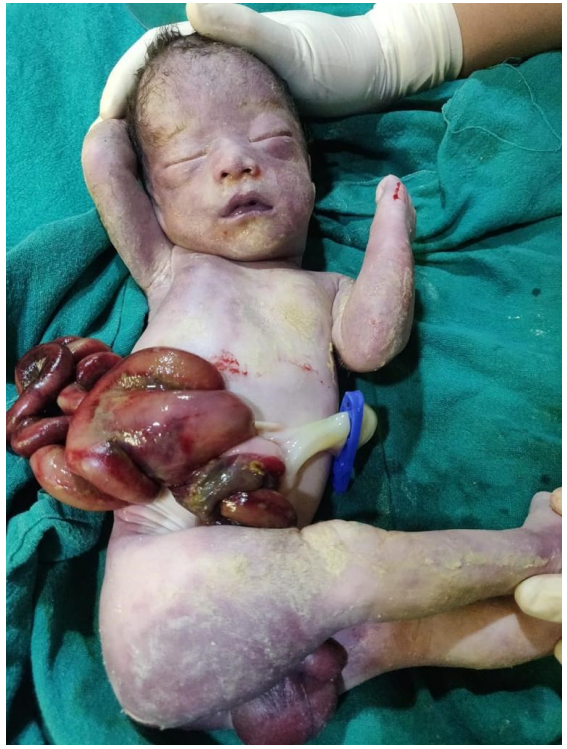
Fetal Cranium

Dilated bilateral lateral ventricles is seen with agenesis of corpus callosum and crowded posterior fossa.

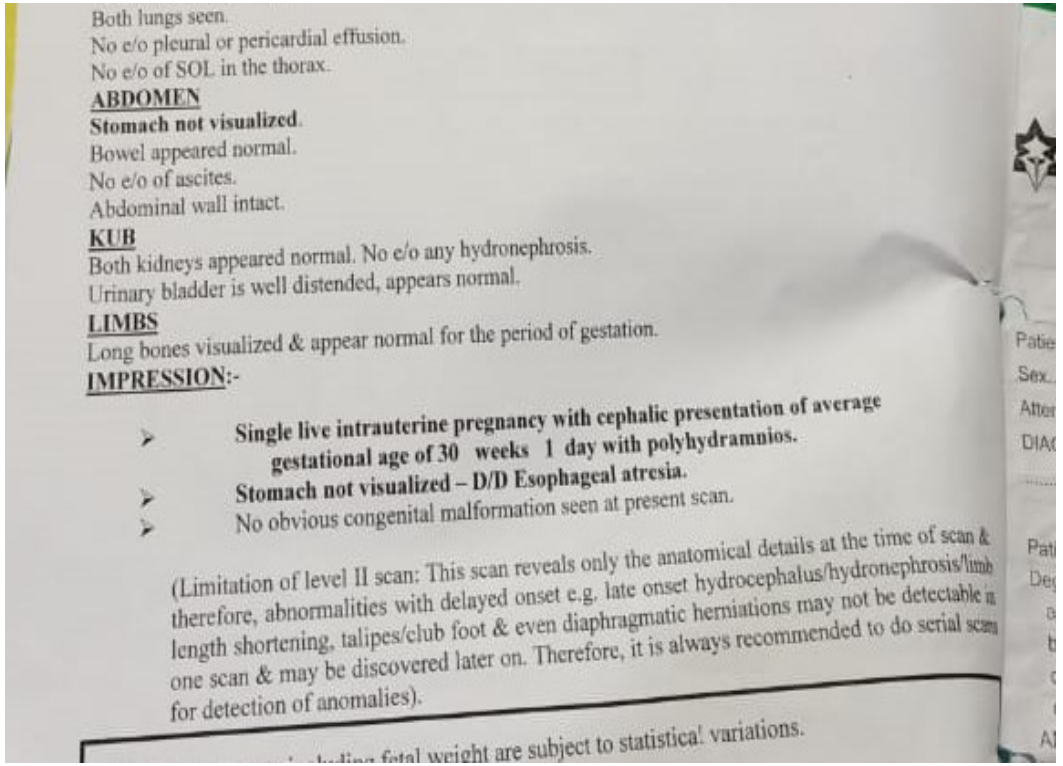
Impression: 23 weeks 4 days of live singleton pregnancy, cephalic presentation, anterior upper uterine placentation, adequate liquor WITH ANOMALIES IN FETAL BRAIN AS DESCRIBED ABOVE



Gastroschisis



Oesophageal atresia



Both lungs seen.
No e/o pleural or pericardial effusion.
No e/o of SOL in the thorax.

ABDOMEN

Stomach not visualized.
Bowel appeared normal.
No e/o of ascites.
Abdominal wall intact.

KUB

Both kidneys appeared normal. No e/o any hydronephrosis.
Urinary bladder is well distended, appears normal.

LIMBS

Long bones visualized & appear normal for the period of gestation.

IMPRESSION:-

- **Single live intrauterine pregnancy with cephalic presentation of average gestational age of 30 weeks 1 day with polyhydramnios.**
- **Stomach not visualized – D/D Esophageal atresia.**
- No obvious congenital malformation seen at present scan.

(Limitation of level II scan: This scan reveals only the anatomical details at the time of scan & therefore, abnormalities with delayed onset e.g. late onset hydrocephalus/hydronephrosis/limb length shortening, talipes/club foot & even diaphragmatic herniations may not be detectable in one scan & may be discovered later on. Therefore, it is always recommended to do serial scans for detection of anomalies).

...including fetal weight are subject to statistical variations.

Others

- Hydrops fetalis
- Omphalocele
- Meningomyelocele
- Congenital heart defects: Hypoplastic left heart
- Multicystic dysplastic kidney
- B/L Renal agenesis
- Limb defects: Aplasia, hypoplasia

Risk factors

- Birth defects are result of one or more genetic, infectious, nutritional or environmental factors
- Infections: Syphilis, Rubella
- Radiation exposure
- Alcohol, drug intake
- Nutritional : Iodine, Folate deficiency
- Obesity
- Maternal diabetes
- Age
- Genetic
  - Consanguinity

Screening

- Preconception screening
  - Carrier state
  - Family history
- During pregnancy
  - 1st trimester : NT/NB scan, dual/combined test, CVS
  - 2nd trimester: Anomaly scan, quadruple test, Amniocentesis, cfDNA analysis
- Neonatal screening

Screening for Down syndrome:

1st Trimester Screening Tests

Maternal Serum Markers

-Preg. asso. Placental Protein A (PAPP-A)

-Free ß hCG

65%

Fetal Marker- Nuchal fold> 4mm at 8-12 wks GA

57-73%

85%

2nd Trimester Screening Tests

Maternal Serum Markers

↓ AFP

↓ E3

↑ hCG

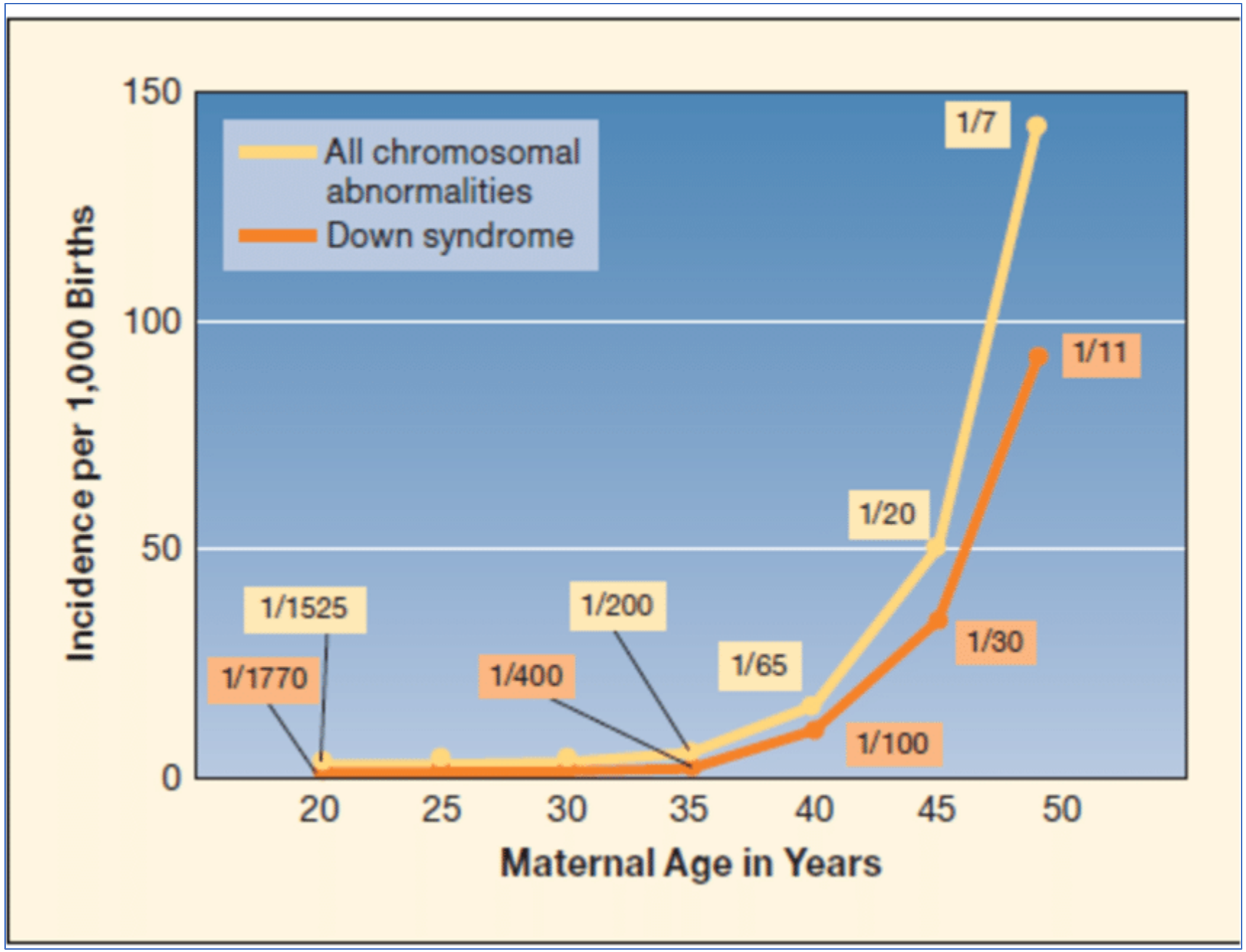
Triple Test 69%

↑ Inhibin

76%

Quadruple Test

94%


150
100
50
0
Incidence per 1,000 Births
All chromosomal abnormalities
Down syndrome
1/7
1/11
1/20
1/30
1/65
1/100
1/200
1/400
1/1525
1/1770
20
25
30
35
40
45
50
Maternal Age in Years

Conclusion

- Congenital anomalies can be single or multiple, major or minor defects.
- Early screening and diagnosis during pregnancy offers less psychological trauma to the patient for termination.
- Supplementation with prophylactic folic acid and identification and modification of possible risk factors may improve future pregnancy outcome.

Department of Radiology

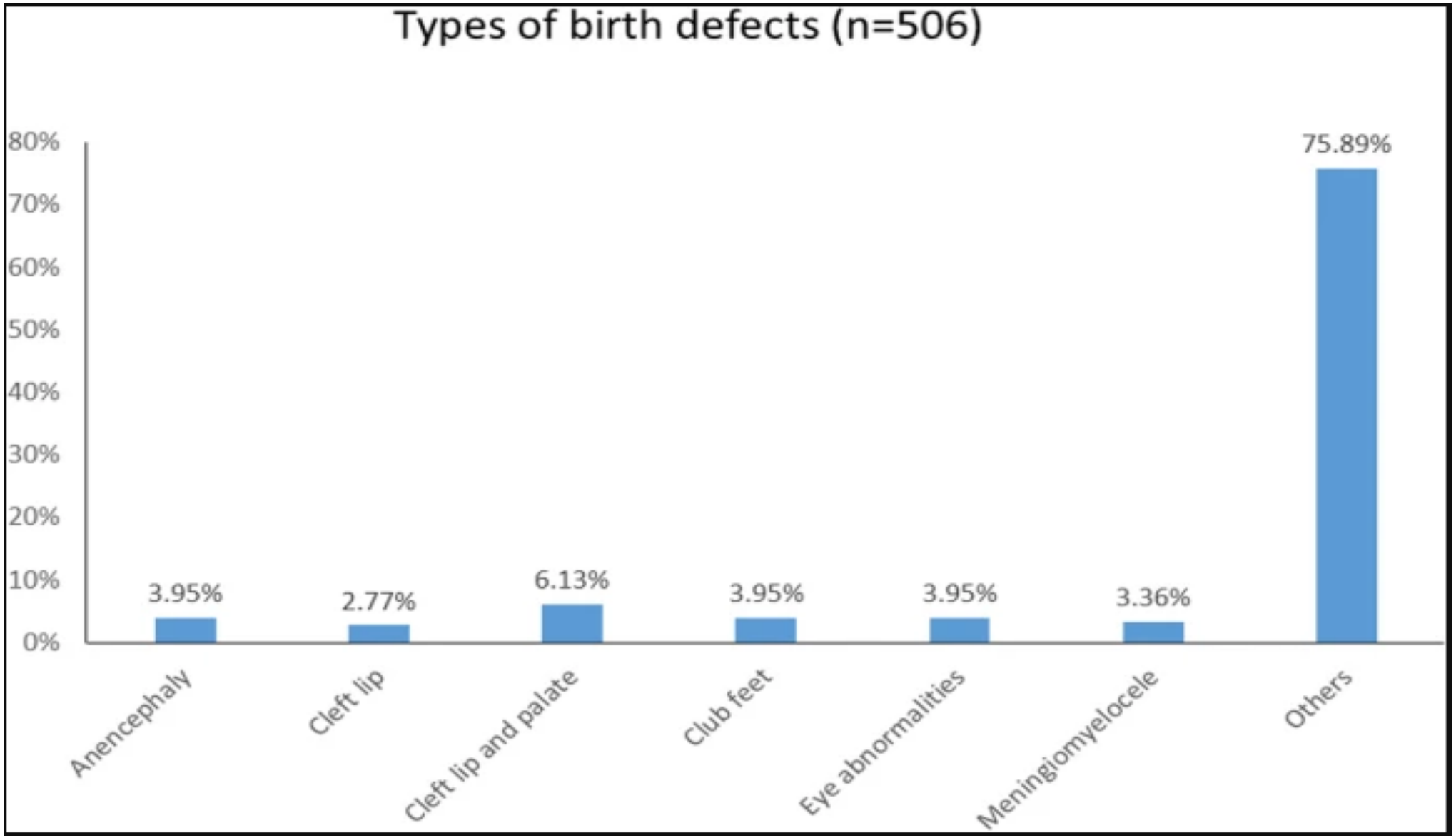
Dr Sandhya Gautam

USG detection of congenital anomalies

- Low risk population: sensitivity- 14-85%, specificity-93-99%
- US (n=200,000 pregnancy): sensitivity- 61.4 % at 18-22wks
- CNS anomalies: commonly detected with USG like anencephaly and hydrocephaly

Influencing factors

- Quality of the equipment
- Prevalence of the particular defect
- Number of studies done per pregnancy
- Type of defect
- Gestational age
- Maternal body habitus
- Amount of amniotic fluid
- Patient bladder preparation
- Examination protocol
- Obesity: Image quality deteriorates as BMI increases



Burden and consequence of birth defects in Nepal-prospective cohort study; SOURCE:BMC pediatrics

Name:
Age: years/ Date:
Technical conditions:
Placenta-
Presentation –
AFI:
Fetal Movement:
Fetal Parameters:

Measurements	cm	Gestational Age
BPD	cm	w d
HC	cm	w d
AC	cm	w d
FL	cm	w d
Average GA		w d
EFW	gms	
FHR	bpm, regular	
Cervical Length	cm	

Abnormal findings: No

Conclusion:

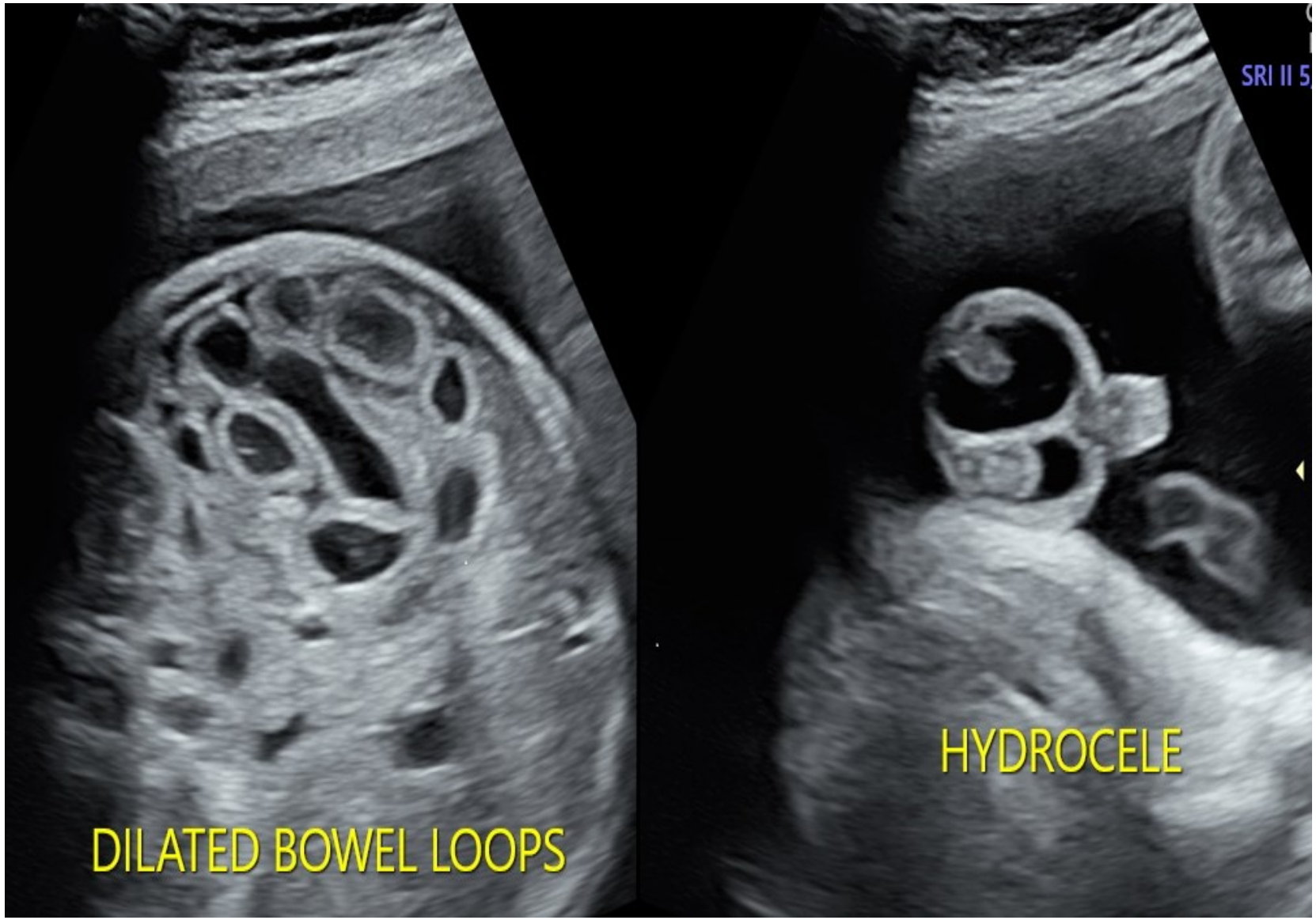
Dr.
Radiologist
NMC No. -

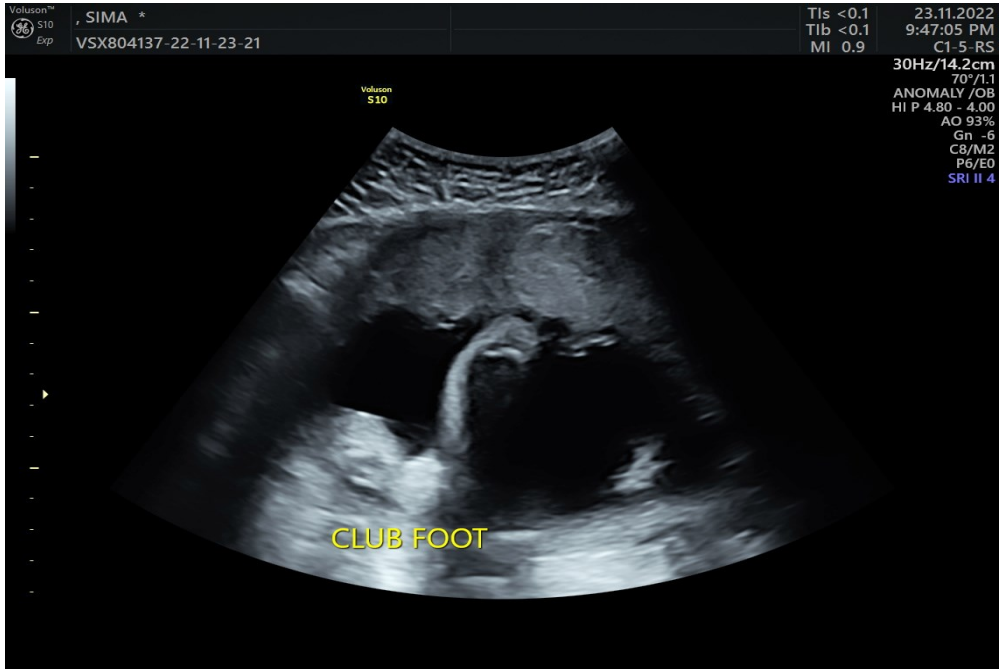
Prepared by:

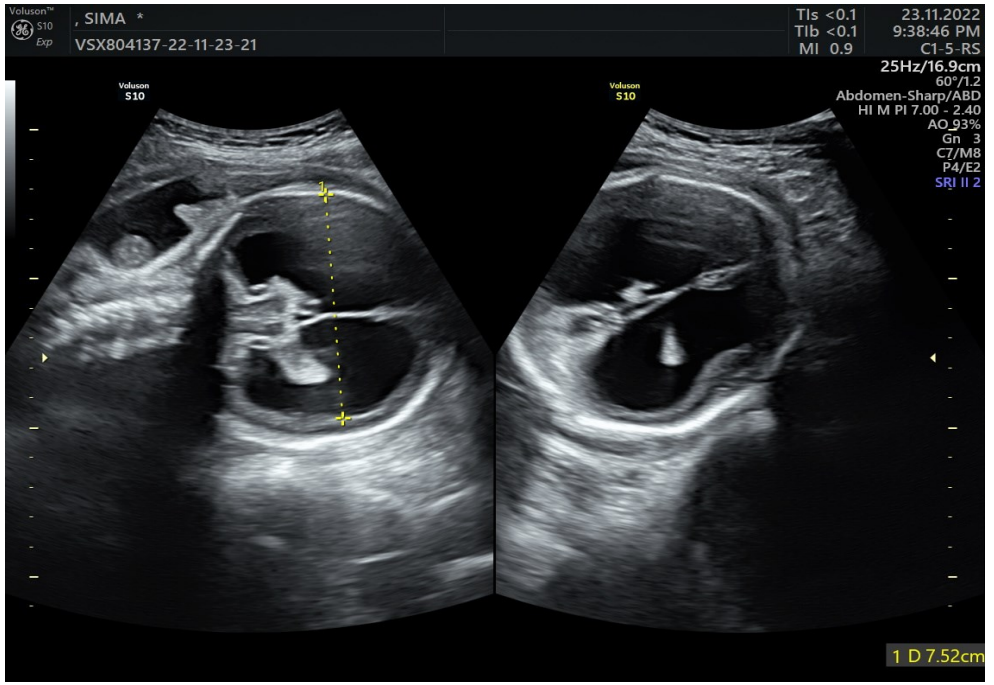
Sonographic appearance of fetal anatomy (N: Normal. Ab.: Abnormal, Nv.: Not visualized, Gray: Optional)	N	Ab	Nv
Head			
• Shape			
• Cavum septum pellucidum			
• Midline falx			
• Lateral ventricle			
• Cisterna Magna			
Face			
• Upper lip			
• Midline profile			
• Orbits			
• Nose			
• Nostrils			
Neck			
Thorax			
• Shape			
• No masses			
Heart			
• Heart Activity			
• Size			
• Cardiac Axis			
• 4- chamber view			
• LVOT			
• RVOT			
Abdomen			
• Stomach			
• Bowel			
• Kidney			
• UB			
• Cord insertion			
• Cord vessels			
Spine			
Limbs			
• Right arm including hand			
• Left arm including hand			
• Right leg including foot			
• Left leg including foot			

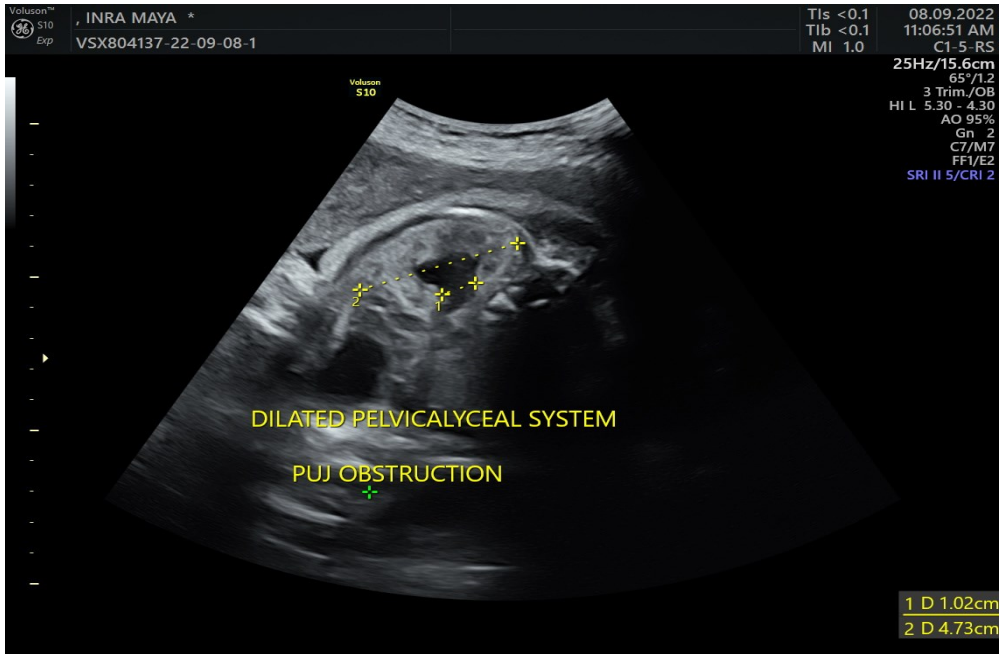
Hospital service

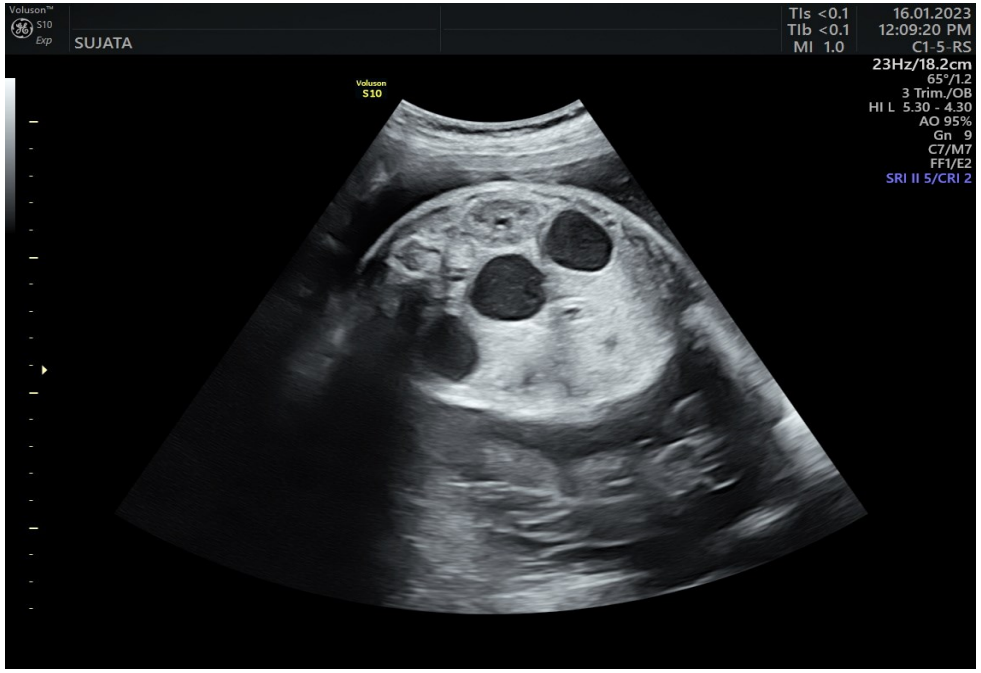
- Total number of anomaly scan done in NMCTH (Aug 2022-Jan2023): **456**
- Anomaly scan at hospital= 6-8/day
- Most common: Neurological and GIT
- Less common: Musculoskeletal and other systemic anomalies



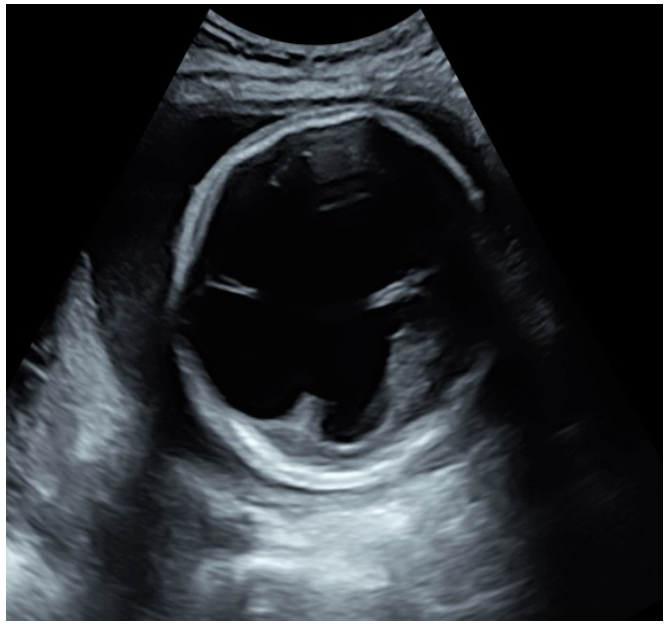

, SIMA *
VSX804137-22-11-23-21
TIs <0.1
TIb <0.1
MI 0.9
23.11.2022
9:47:05 PM
C1-5-RS
30Hz/14.2cm
70°/1.1
ANOMALY /OB
HI P 4.80 - 4.00
AO 93%
Gn -6
C8/M2
P6/E0
SRI II 4
CLUB FOOT

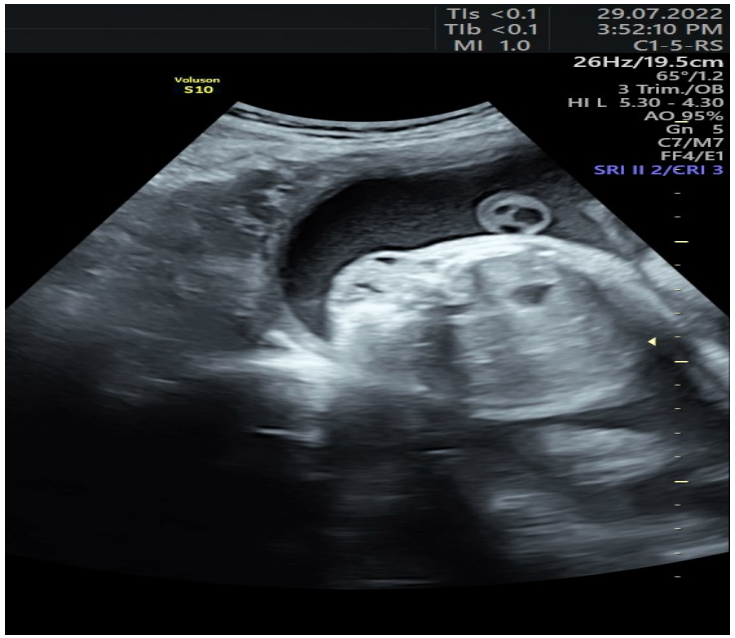

, SIMA *
VSX804137-22-11-23-21
TIs <0.1
TIb <0.1
MI 0.9
23.11.2022
9:38:46 PM
C1-5-RS
25Hz/16.9cm
60°/1.2
Abdomen-Sharp/ABD
HI M PI 7.00 - 2.40
AO 93%
Gn 3
C7/M8
P4/E2
SRI II 2
1 D 7.52cm

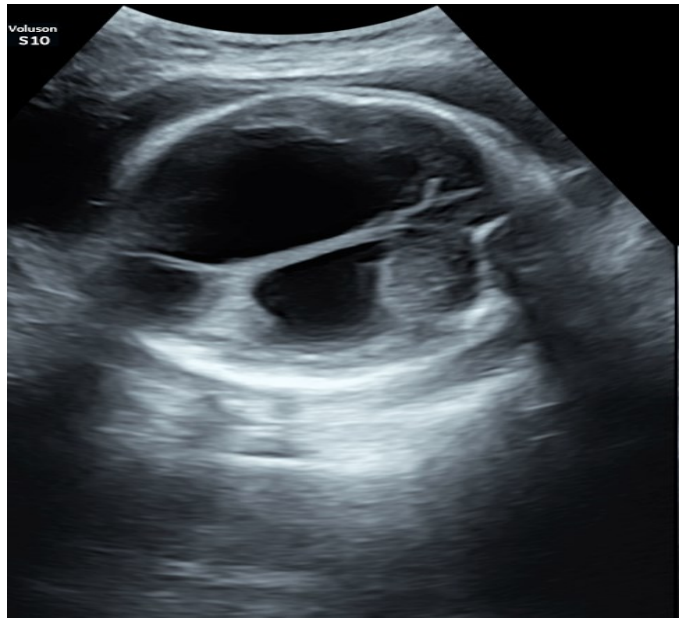

, INRA MAYA *
VSX804137-22-09-08-1
TIs <0.1
TIb <0.1
MI 1.0
08.09.2022
11:06:51 AM
C1-5-RS
25Hz/15.6cm
65°/1.2
3 Trim./OB
HI L 5.30 - 4.30
AO 95%
Gn 2
C7/M7
FF1/E2
SRI II 5/CRI 2
DILATED PELVICALYCEAL SYSTEM
PUJ OBSTRUCTION
1 D 1.02cm
2 D 4.73cm


SUJATA
TIs <0.1
TIb <0.1
MI 1.0
16.01.2023
12:09:20 PM
C1-5-RS
23Hz/18.2cm
65°/1.2
3 Trim./OB
HI L 5.30 - 4.30
AO 95%
Gn 9
C7/M7
FF1/E2
SRI II 5/CRI 2

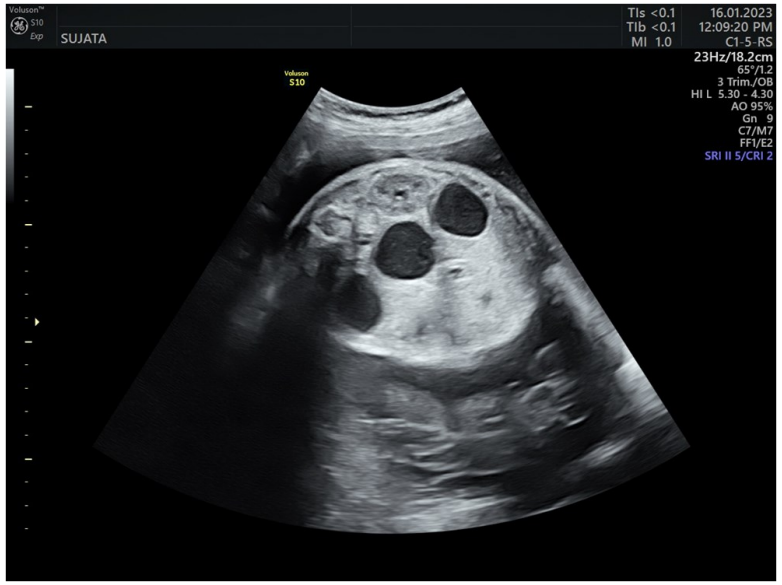
29 yrs female at 31weeks of pregnancy



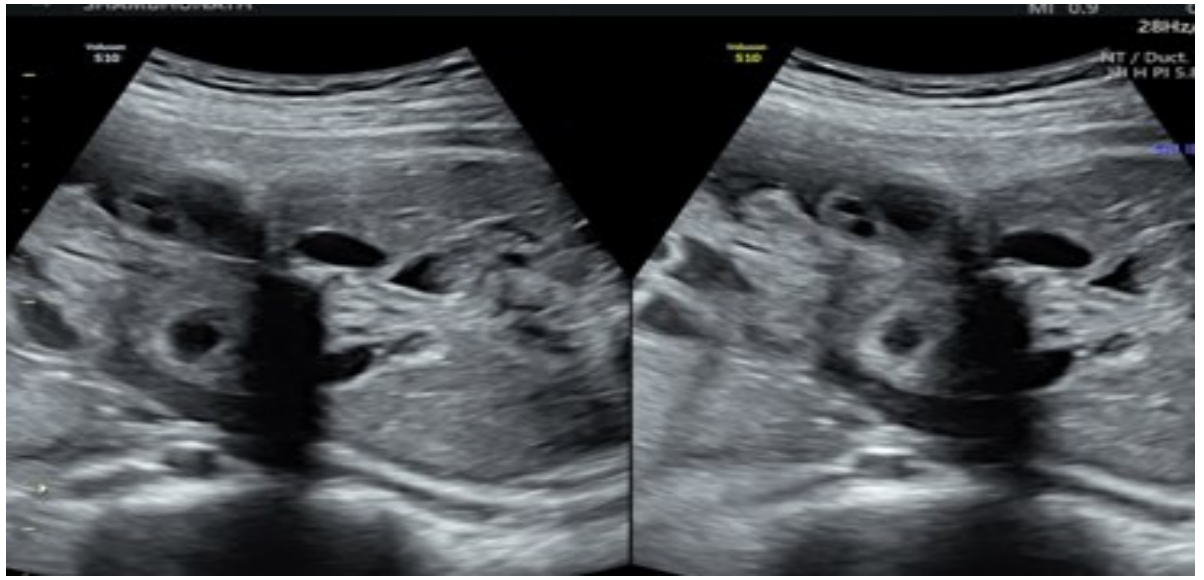




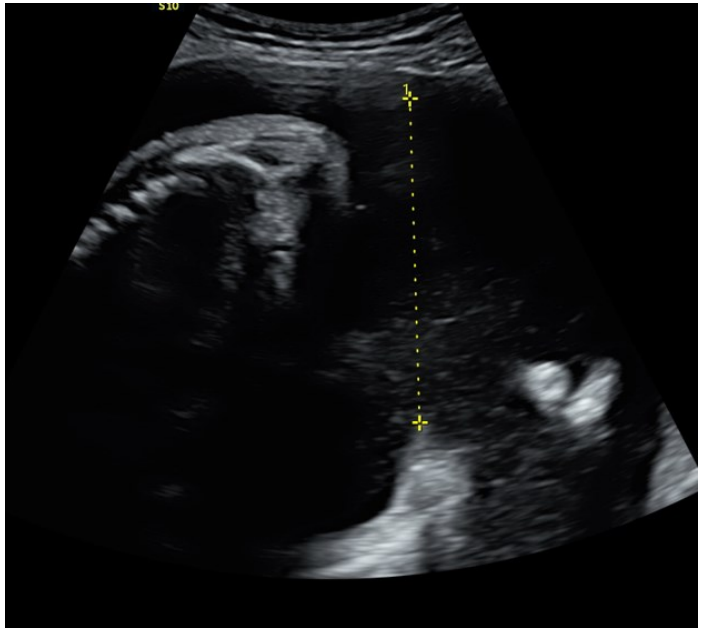
Triple bubble sign

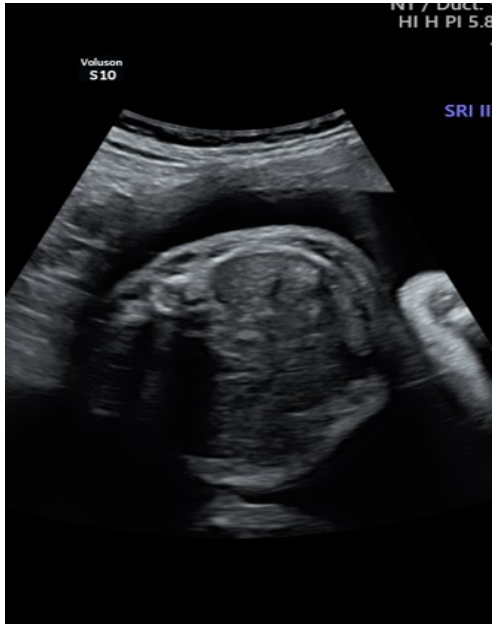


Gastroschisis



TEF





Department of Neonatology

Dr Subash Bhattarai

Epidemiology

- 1 in 33 infants/3.2 million disabilities per year/90% in L-MICC
- Death: 7% or 270 000 NND/year and 170 000 between 1 month - 5 years
- Most common severe birth defects: heart and neural tube defects

- Genetic
- Nongenetic Teratogenic Etiology
  - Maternal DM
  - Drugs and Chemicals
  - Maternal Infection
- Multifactorial Disorders
  - Cleft lip, Palate
  - Congenital Heart Disease
  - Neural tube Defect

Common Major Birth Defect We Encountered

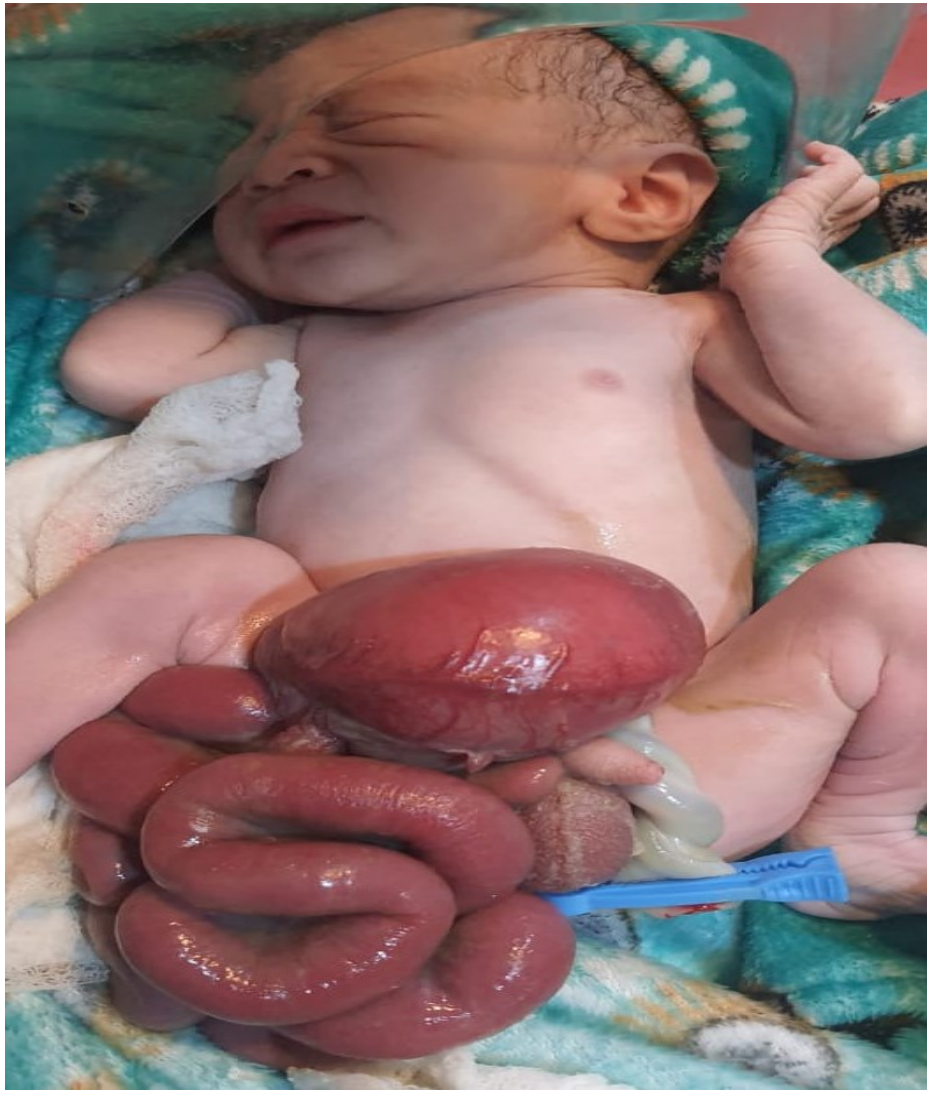
- Cleft lip with and without cleft palate/Cleft palate: 8
- Various CHDs: 7
  - HOCM: 2
  - TAPVC: 2
  - TOF: 2
  - Single Ventricle Physiology:1
- Down Syndrome: 6

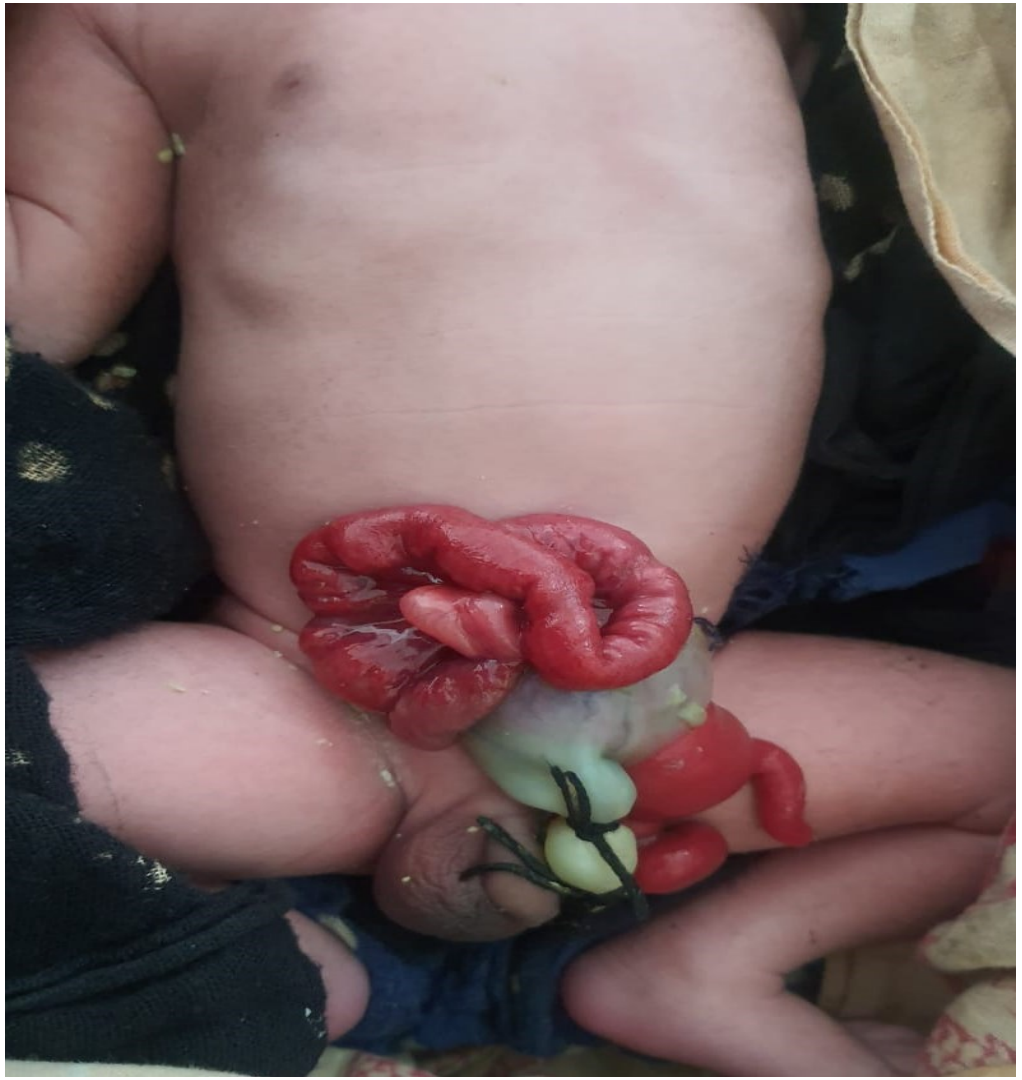
- Genitourinary Abnormalities: 4
- Congenital TORCH Infection : 3
- Gastroschisis: 3
- Tracheo- Esophageal Fistula: 3
- Congential Diapragmatic Hernia: 3
- Omphalocele: 1

Down Syndrome

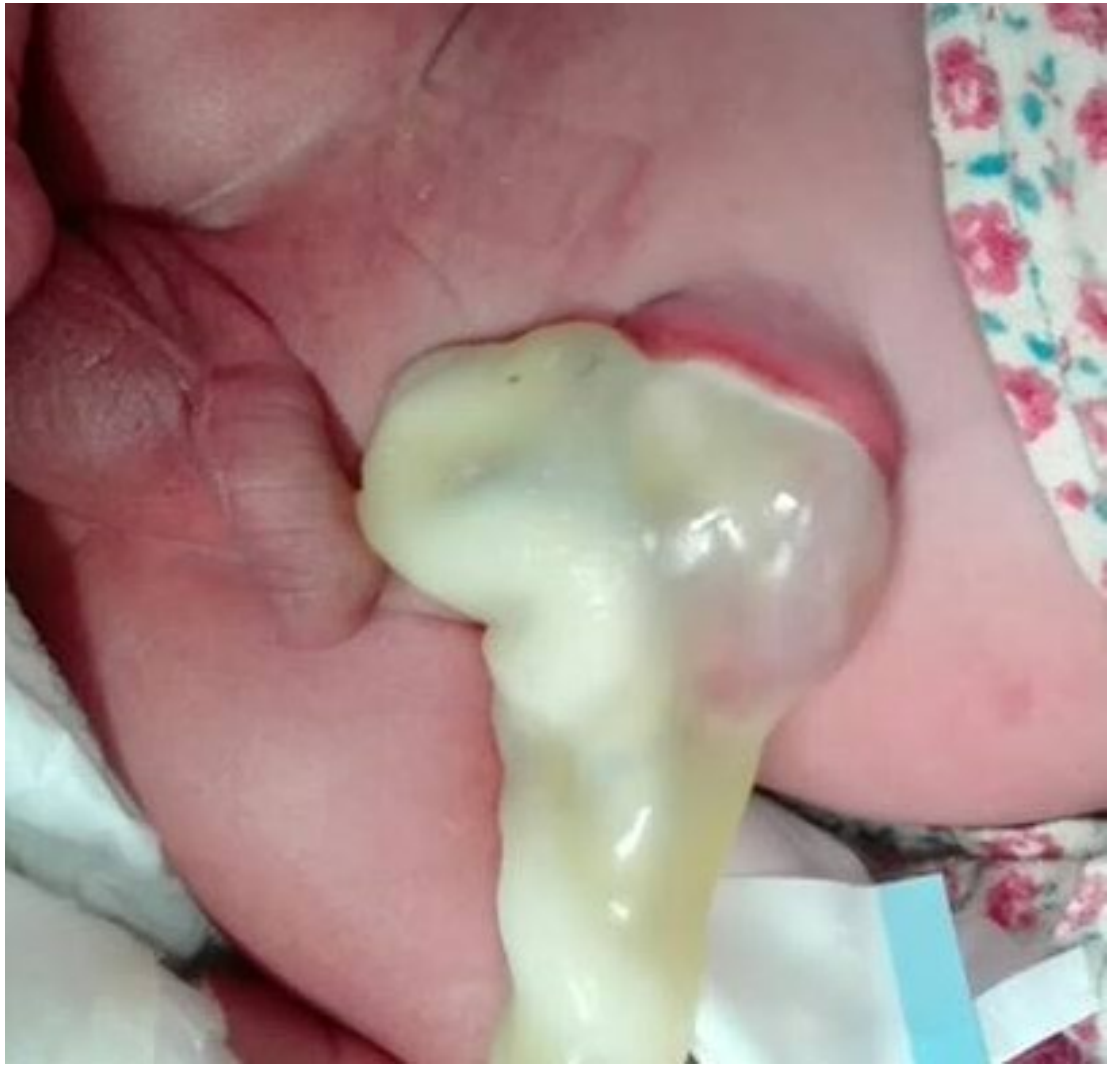
- Most chromosomal disorder
- Associated with increased maternal age
- Down Syndrome Society, Nepal.
  - Equal rights and opportunities for people with Down Syndrome and other Intellectual Disabilities

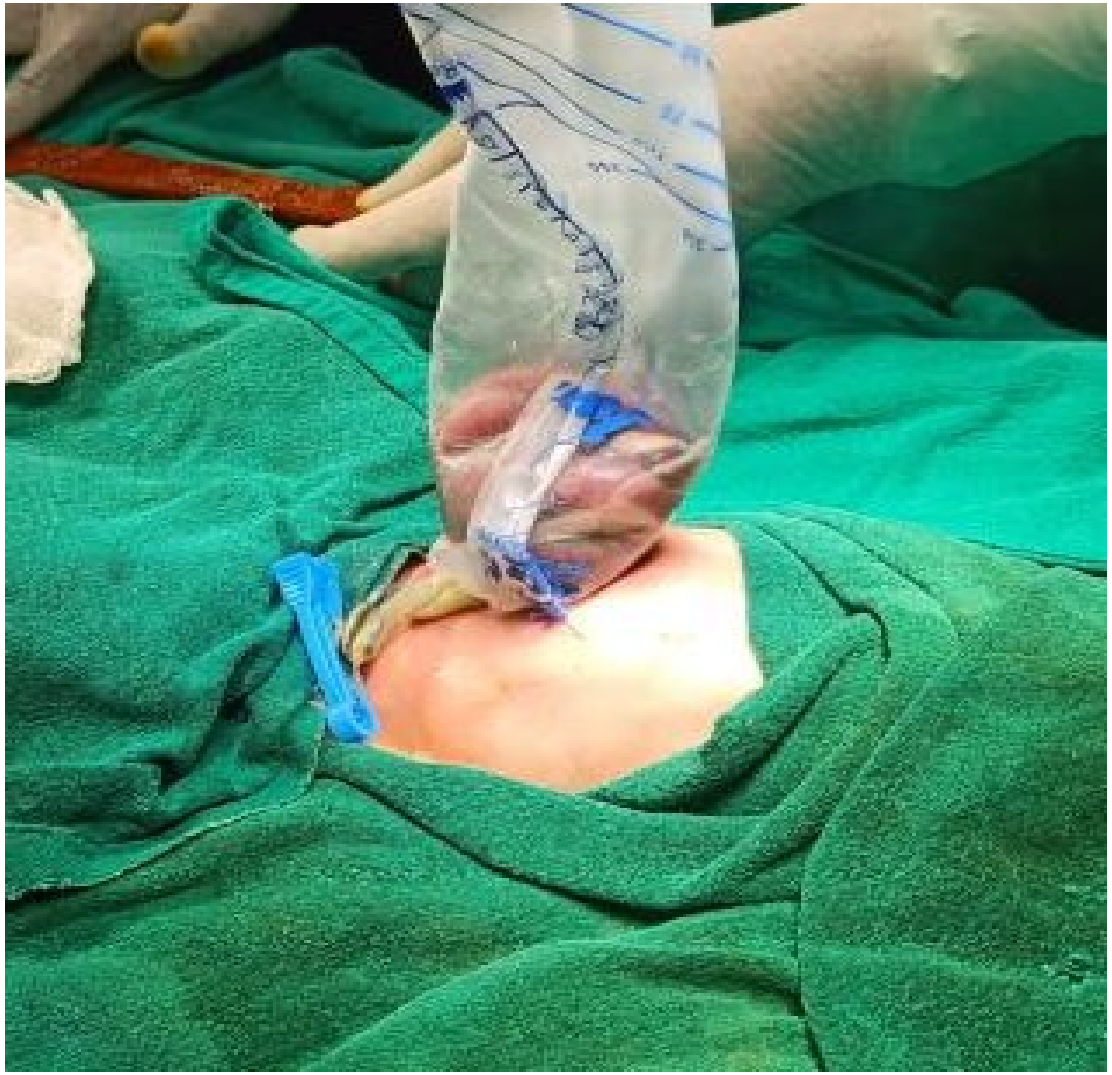
Gastrochisis



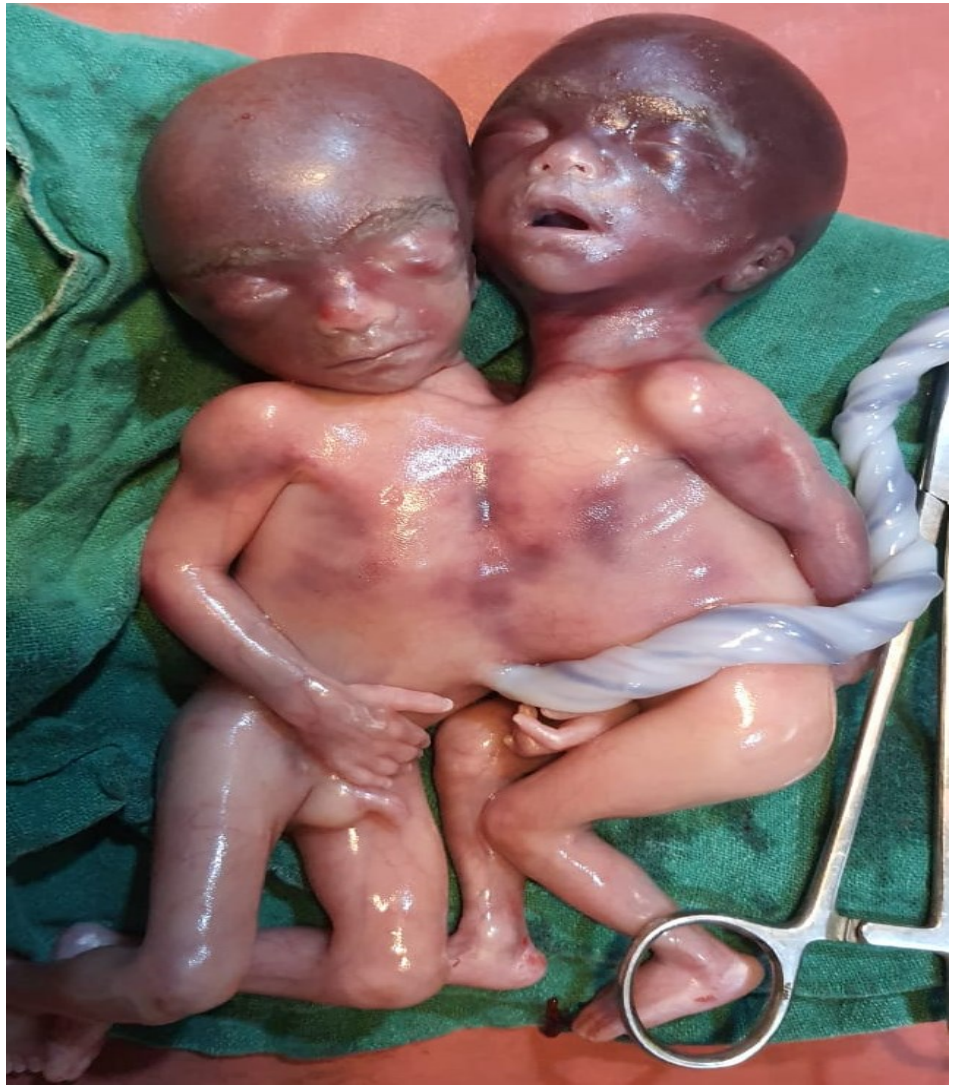


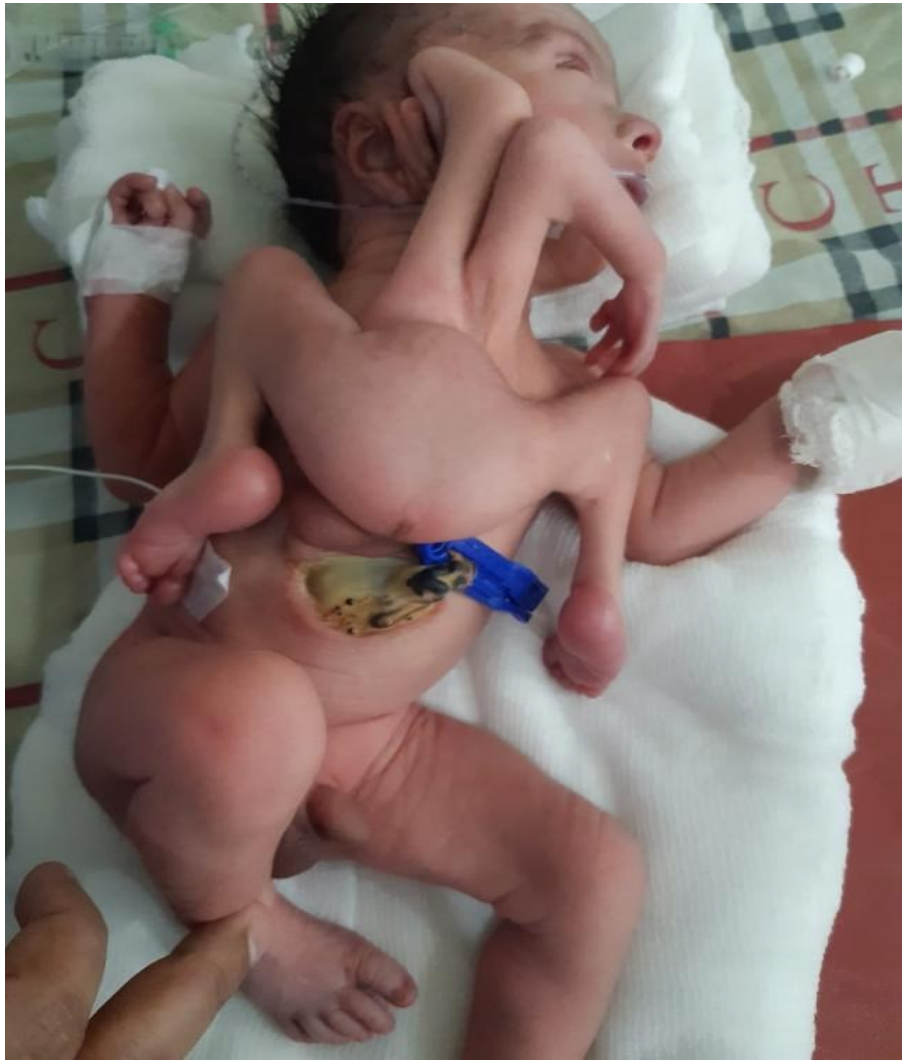
Omphalocele



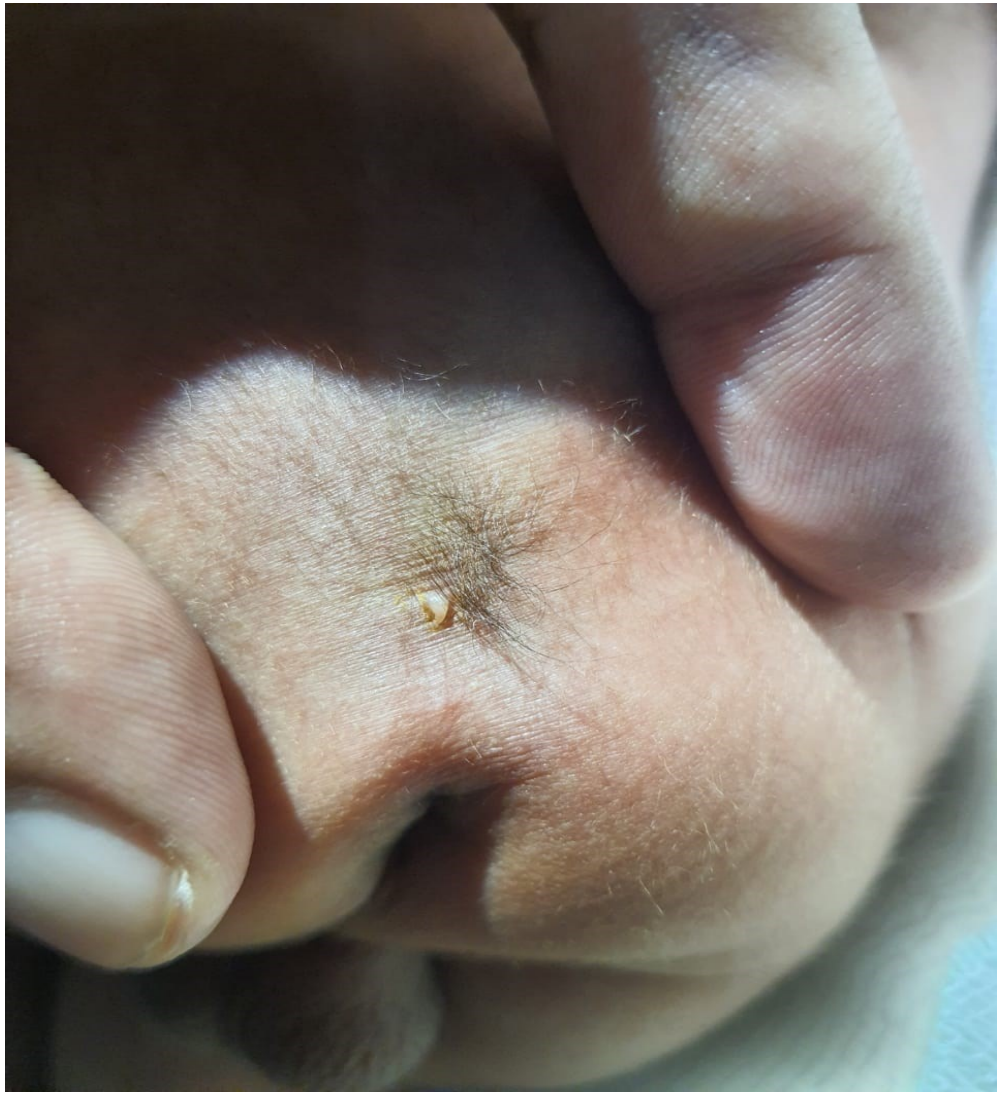


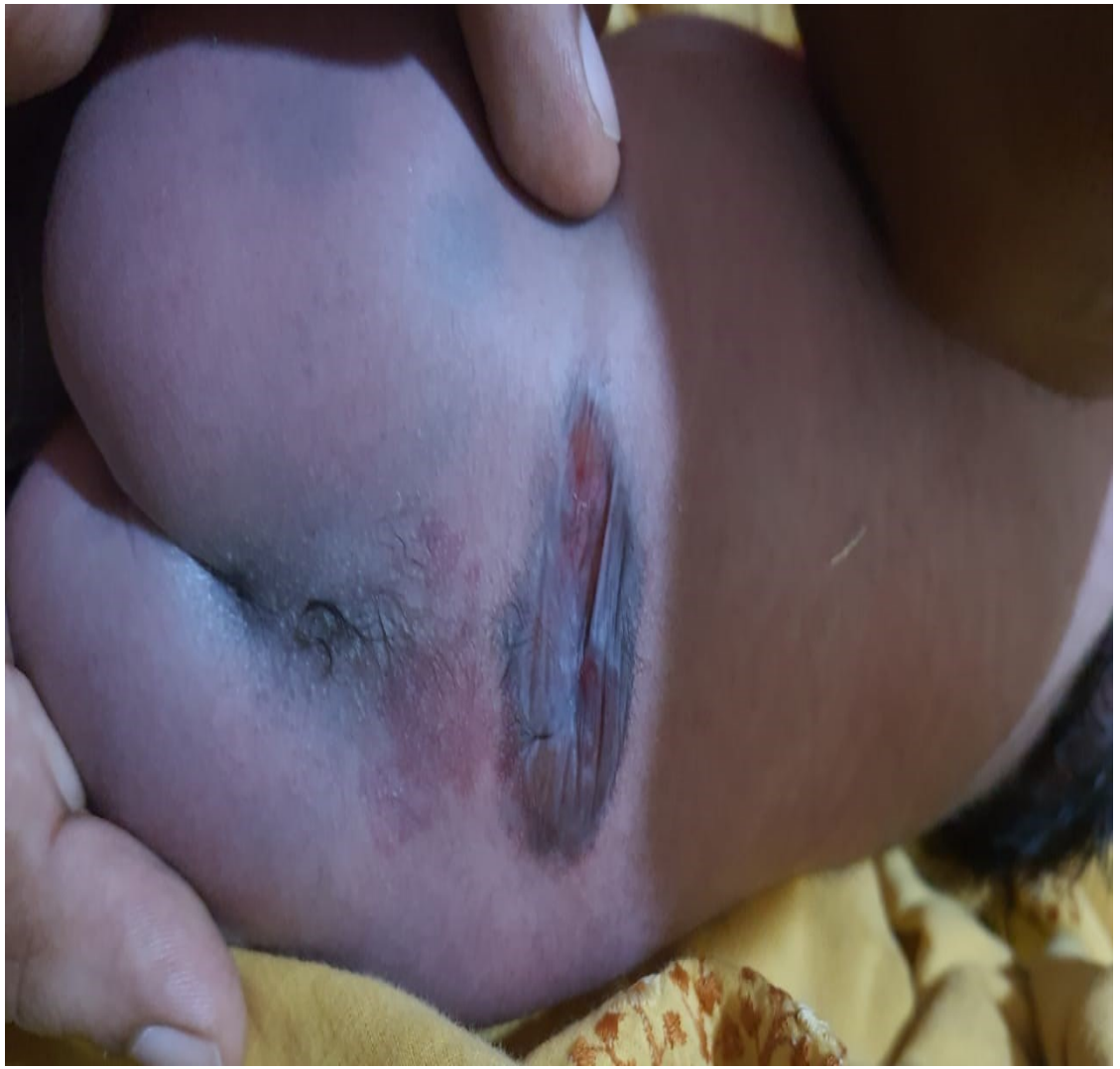
Conjoint Twin

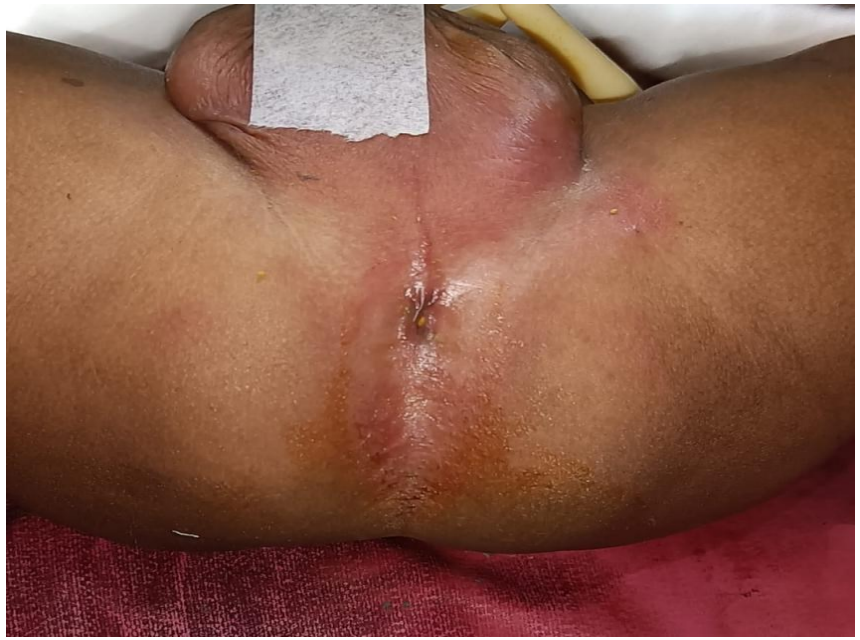




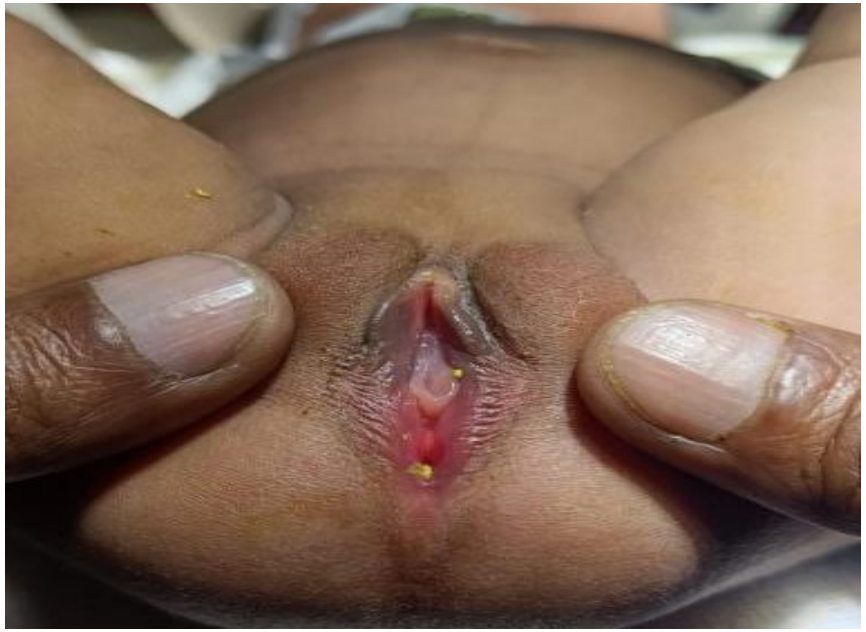
Neural tube Defect

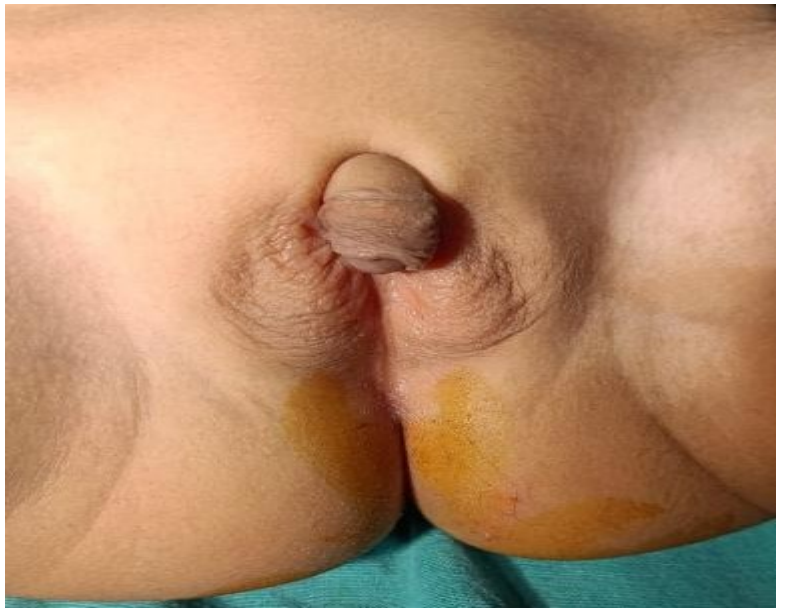


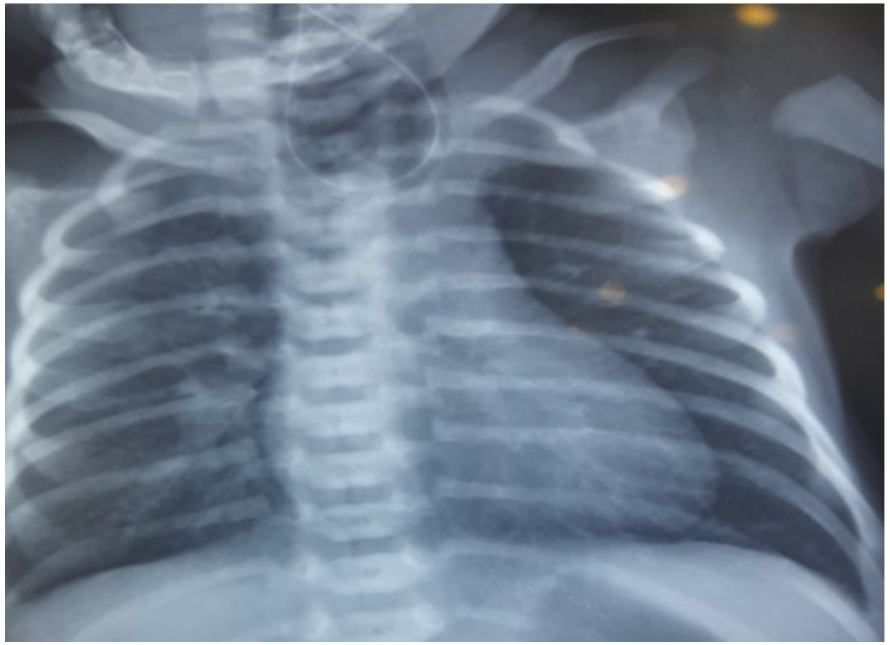




Genitourinary Abnormalities

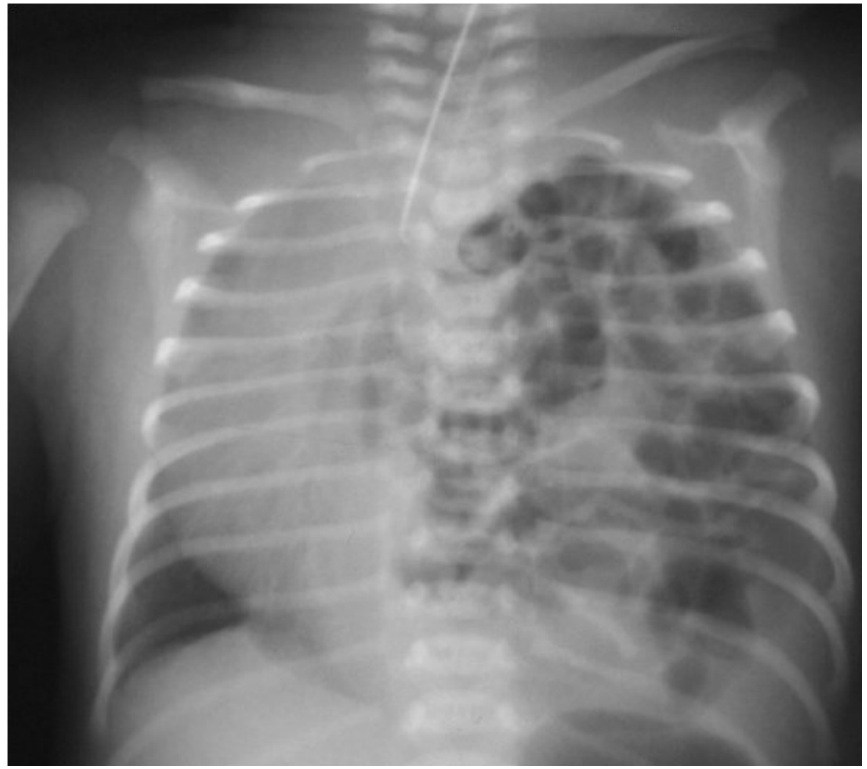


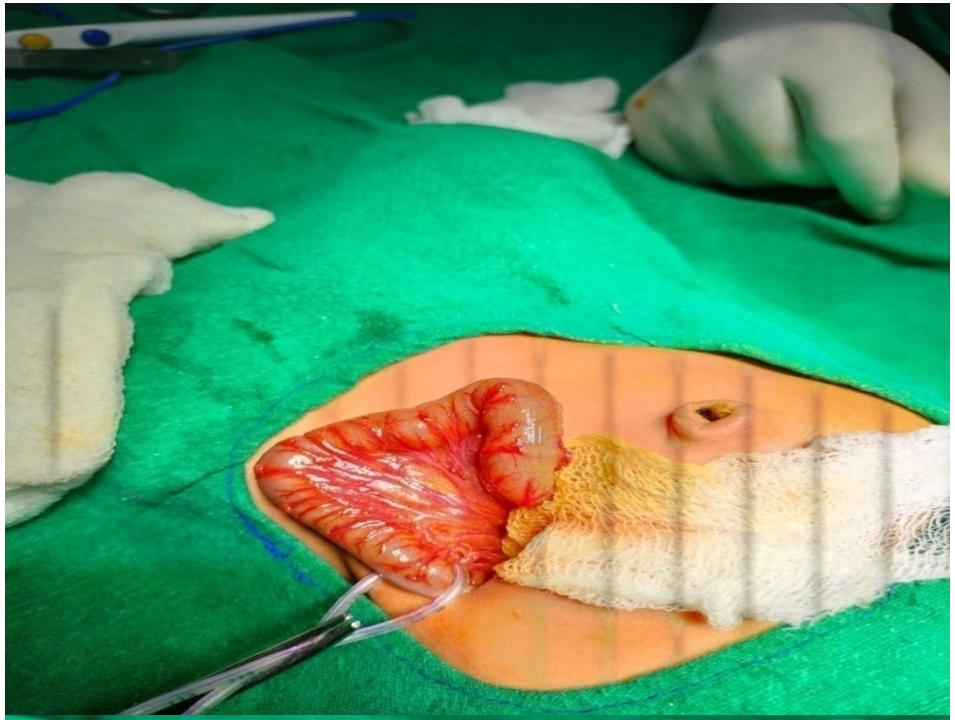


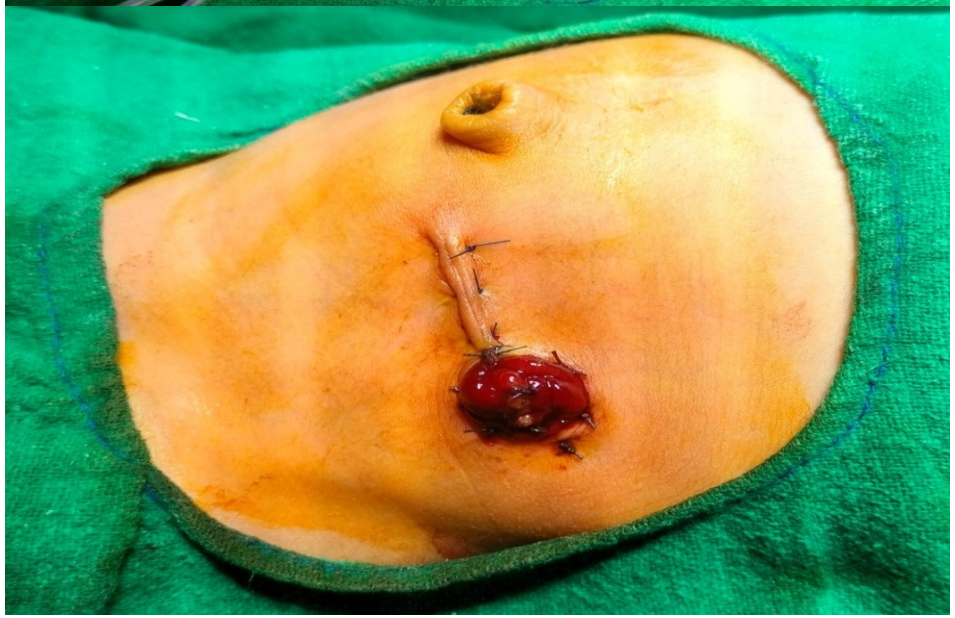


Tracheoesophageal Fistula

Diaphragmatic Hernia







GI Anomalies

Nobel Diagnostic Center Pvt. Ltd.
Nobel Medical College & Teaching Hospital (P.) Ltd.

Kanchanbari,
Biratnagar-4 (Nepal)
Ph. No.: 021-460736, 461735

Name: B/O Laxmi Shrestha 6D/M
Ref By:
I.D No: 79088613

Histo No. 310/023
Date: 2079/11/09

Histopathology report

Clinical History:

Specimen:
A. Rectosigmoid tissue
B. Descending colon (mid)
C. Colostomy site

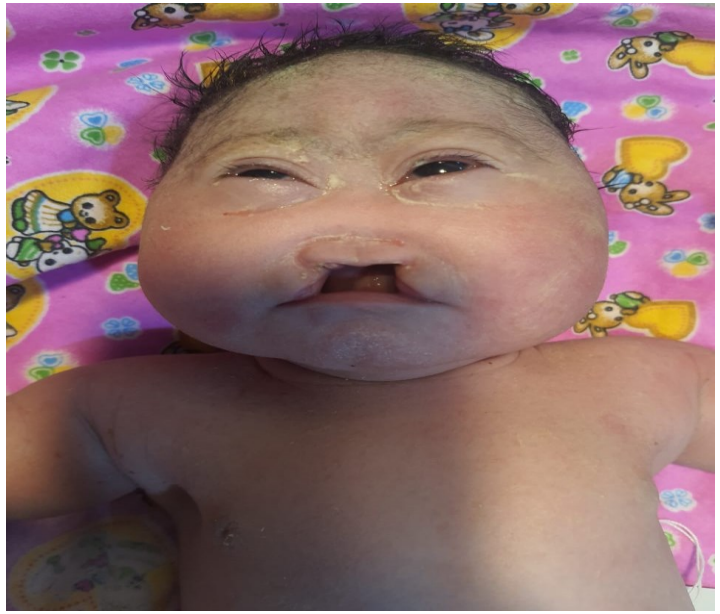
Gross:
A: Received specimen labeled as "Rectosigmoid tissue" is single irregular tissue piece measuring 0.2X0.1 cm, pale white color. A/E in A
B: Received specimen labeled as "Descending colon" is single irregular tissue piece, measuring 0.2X0.2 cm, pale white in color with areas of hemorrhages. A/E in B
C: Received specimen labeled as "Colostomy site" is single irregular tissue piece measuring 0.3X0.2 cm, pale white in color. A/E in C

Microscopy:

A. The section from rectosigmoid tissue shows hypertrophy of muscles and nerve bundles. There is complete absence of ganglionic cells in the section examined. Malignancy is not seen.
B. The section from descending colon shows hypertrophy of muscles and nerve bundles. There is complete absence of ganglionic cells in the section examined. Malignancy is not seen.
C. The section from Colostomy site shows tissue lined by columnar cells with basally placed nuclei. The submucosal layer shows chronic inflammatory infiltrates along with hemorrhages. Muscular layer is hypertrophied.

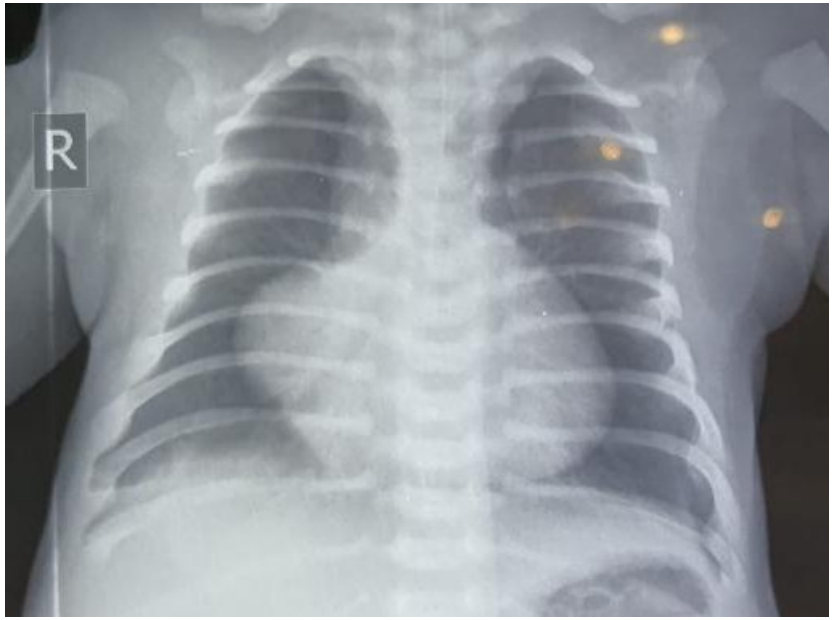
Impression: Consistent with Hirschsprung Disease.

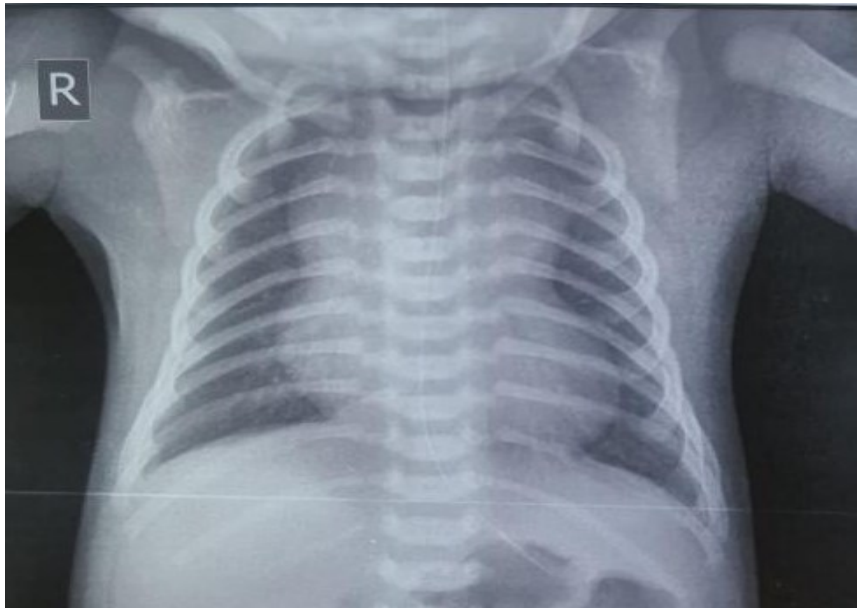
Consultant Pathologist
Dr Rashmita Bhandari
M.D (pathology)
NMC NO:18455

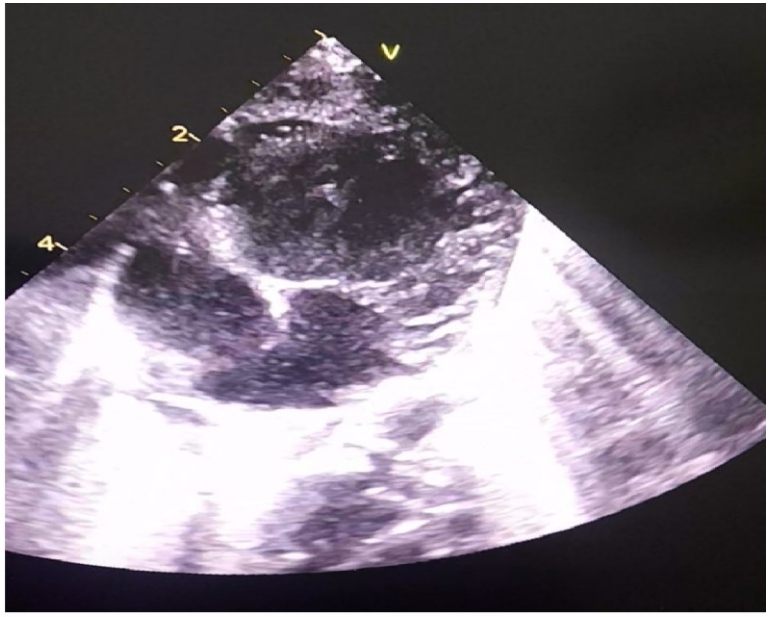


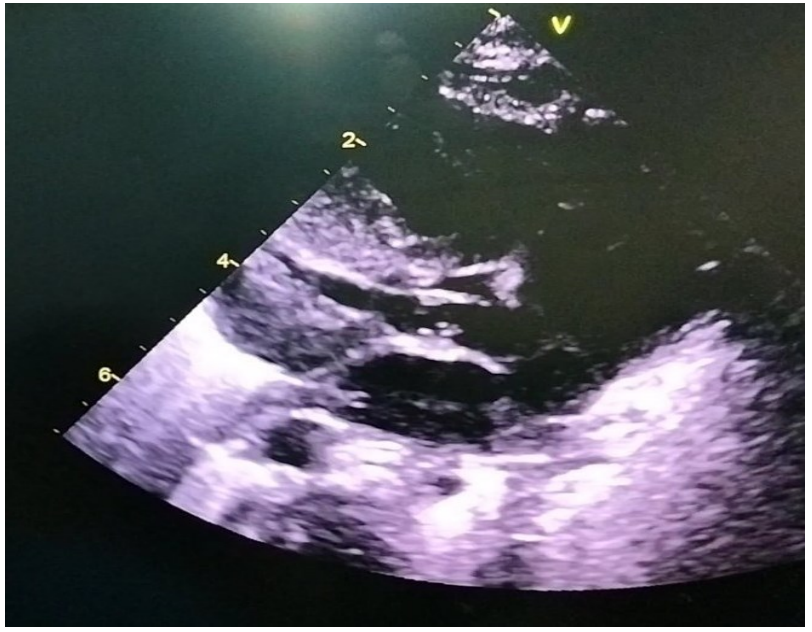
Cleft Lip and Palate

Congenital Heart Disease









Role of Paediatrician

- Antenatal Counselling
- Early Identification and Timely Referral
- Anticipation for need of Resuscitation
- Pregnancy History
- Thorough Head to toe examination
- Laboratory Studies

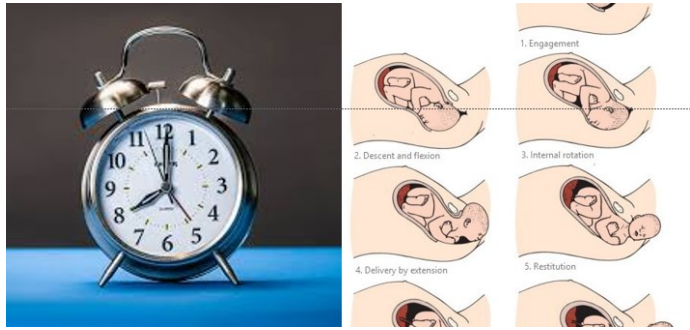
<u>Low Resource Setting</u>

- All Babies should be examined properly
- Thorough Examination (Head to Toe Examination including spine)
- Cyanosis
- Pulse Oximetry Monitoring
- All Orifices Should be checked

Department of Neurosurgery

Dr Prakash Kafle

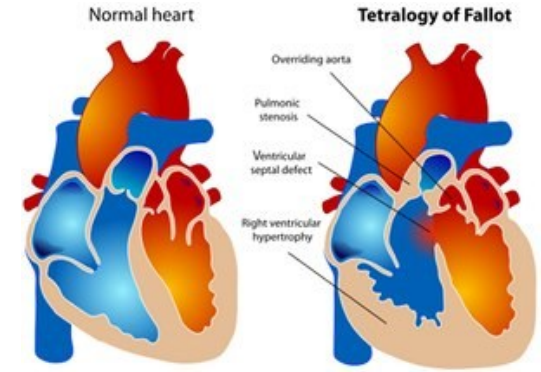
CNS Birth Defects

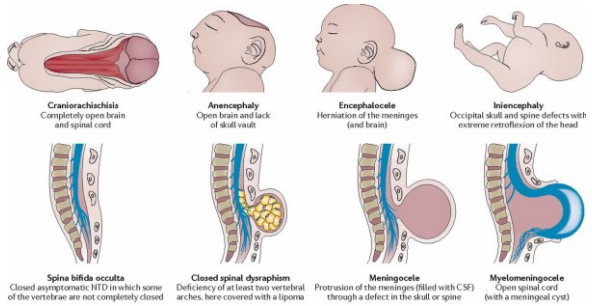


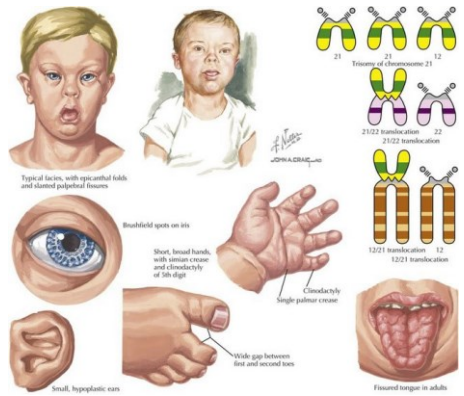
- Presents at birth
- Mild to severe form
- Surgery
  - Address the symptoms
  - Correct the deformity
  - Maximize cognitive and motor functions
  - Prevent the development of neurological deficits

Most common & severe BD

- Heart defects
- Neural tube defects
- Down syndrome





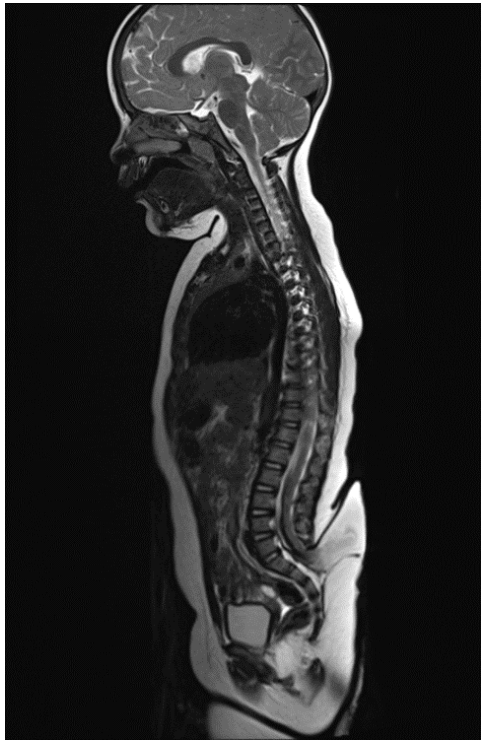


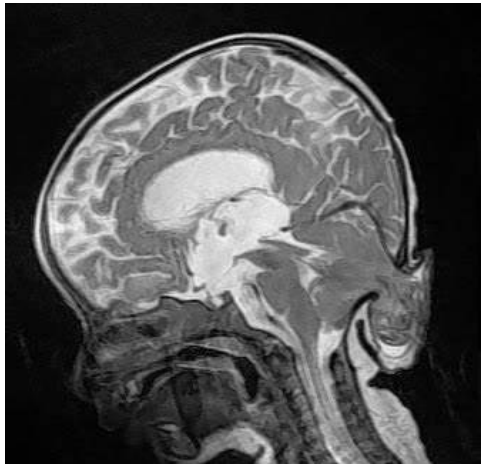
Commonly encountered-CNS- BD

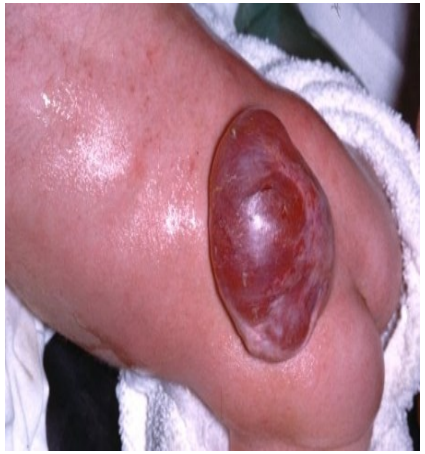
- Neural Tube defects
  - Encephaloceles
  - Mylomeningocele
  - Lipomylomeningocele
  - Meningocele
  - Mylocele
  - Anencephaly and its spectrum
- Arachnoid Cysts
- Chiari Malformations
- Aqueduct Stenosis

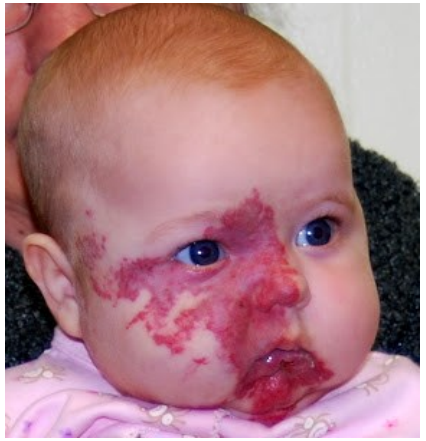
- Sturge-Weber Syndrome
- Prosencephaly
- Microcephaly
- Megalencephaly
- Lissencephaly
- Cortical micro dysgenesis
- Agenesis of the corpus callosum
- Aplasia/Hypoplasia of Cerebellum
- Dandy walker malformation
- Joubert syndrome

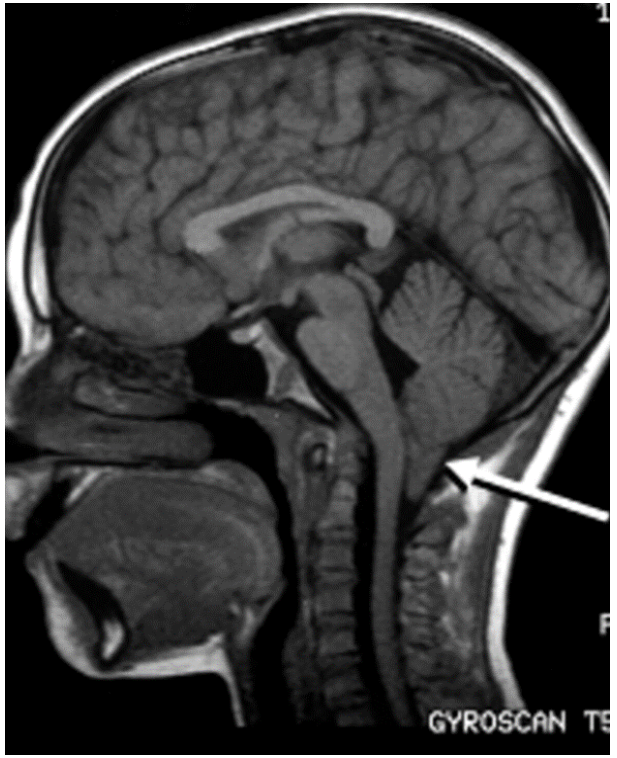
Illustrative Images

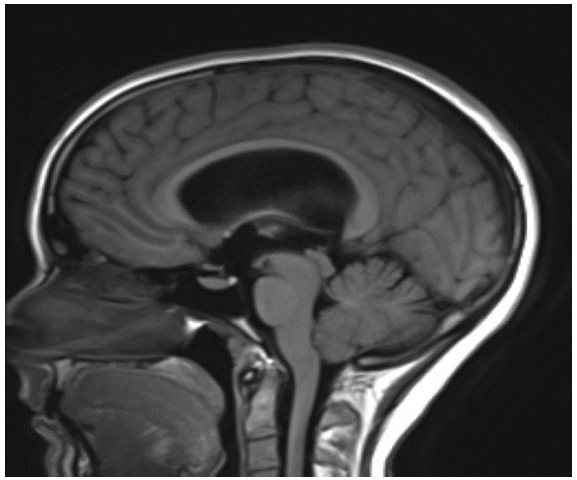


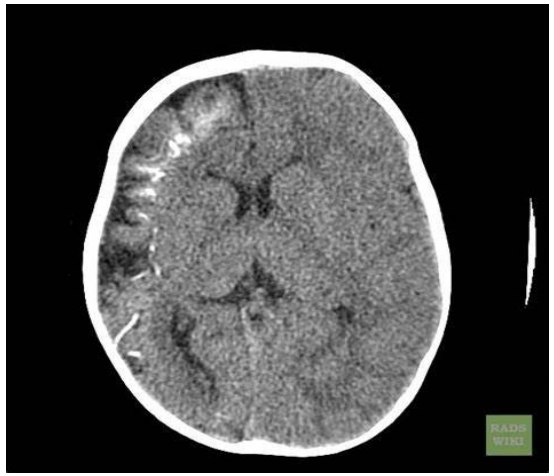


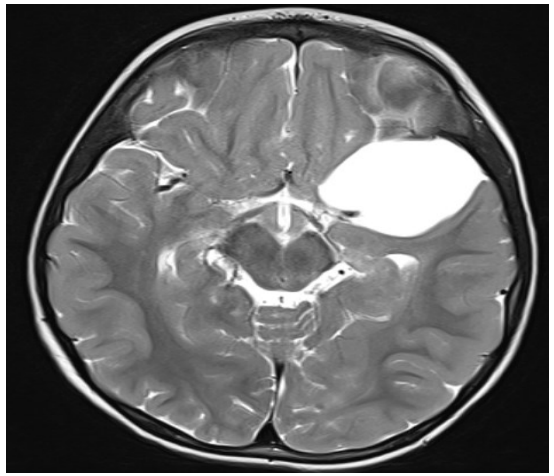




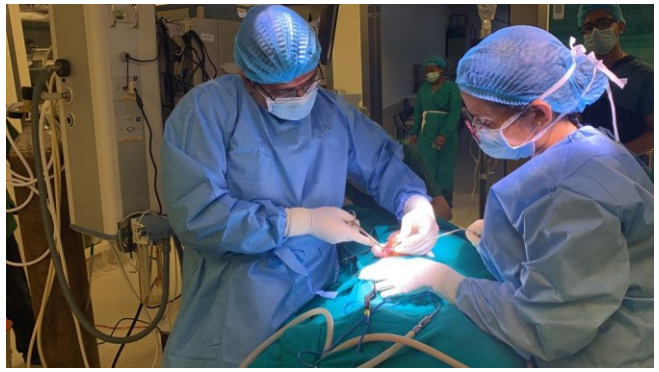


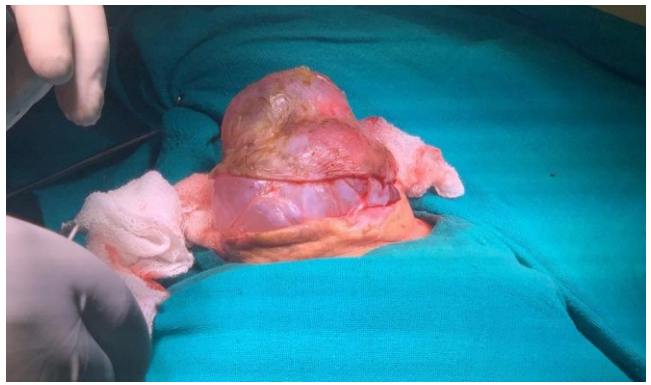


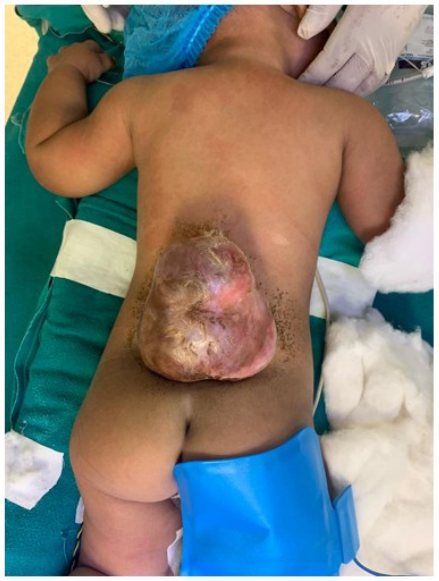


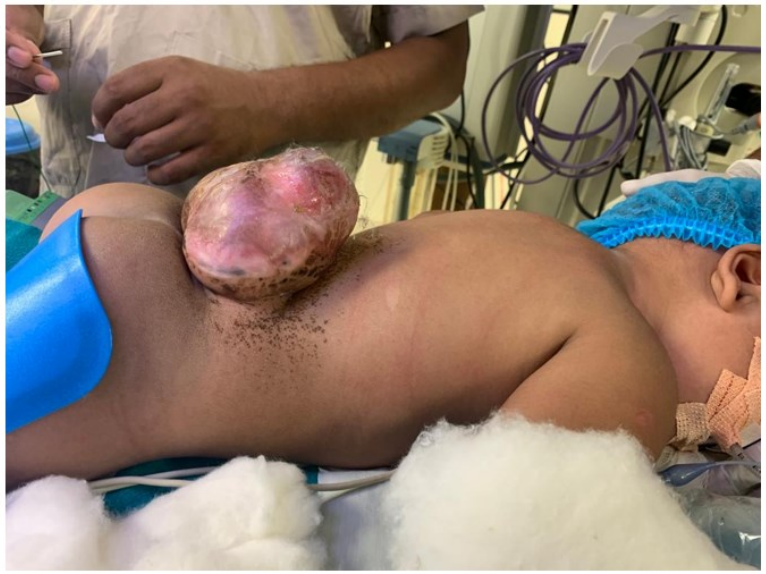


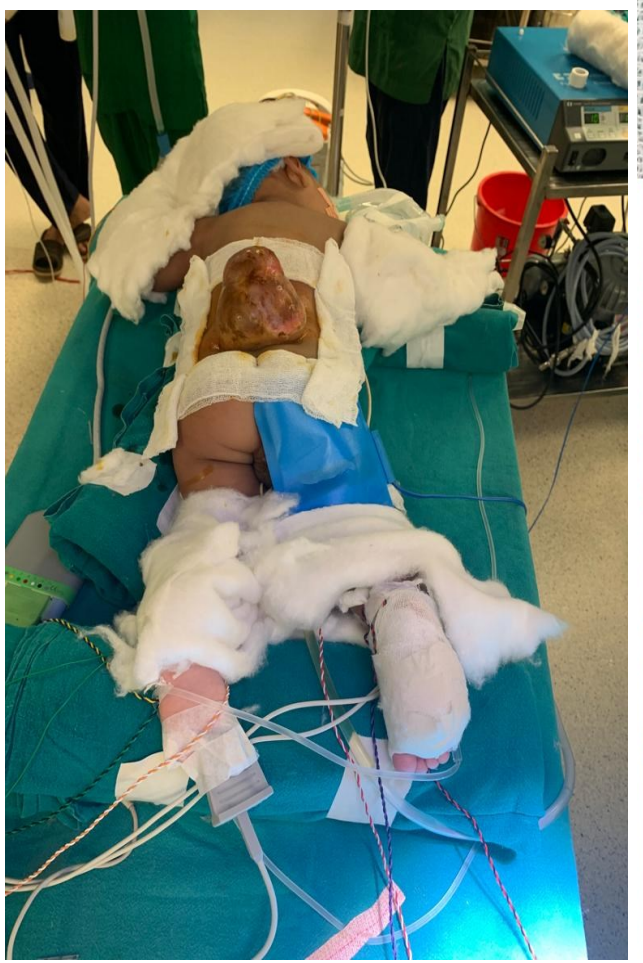
Illustrative cases

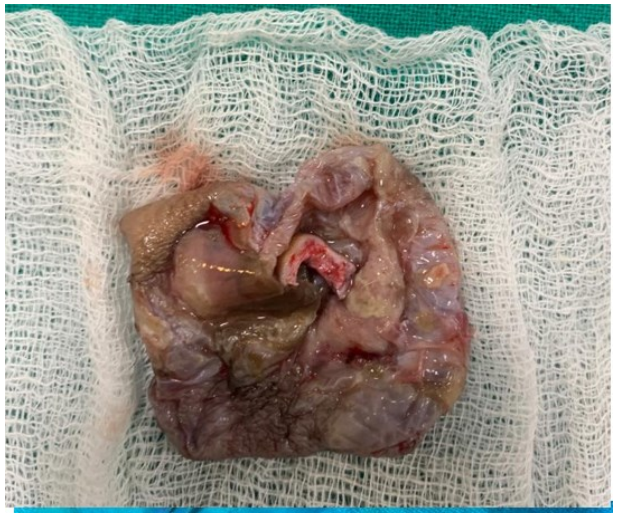


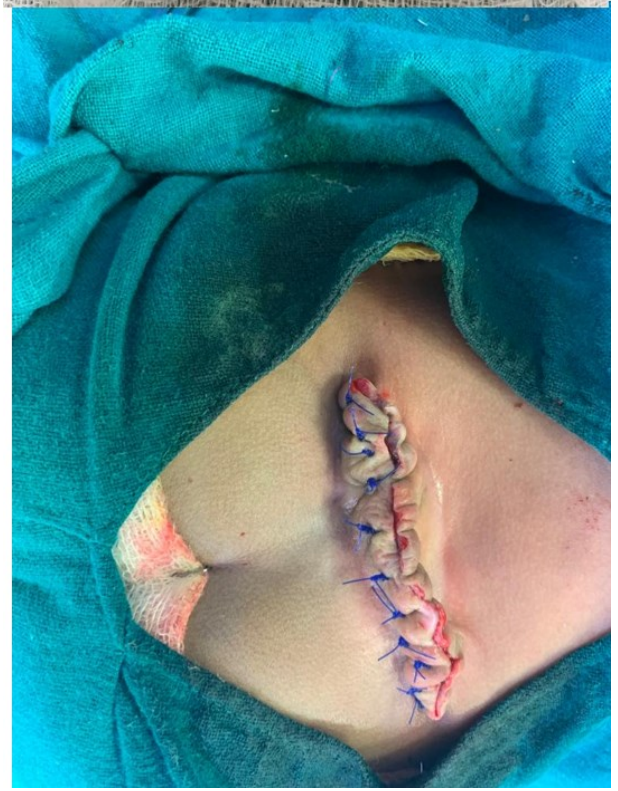


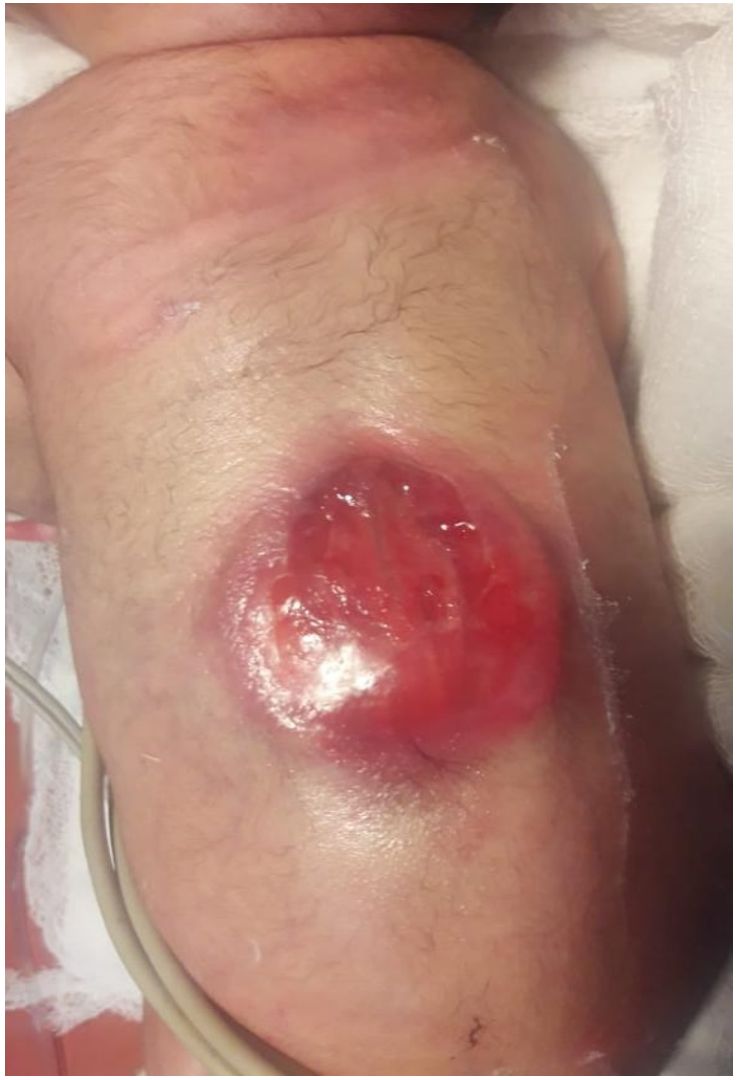




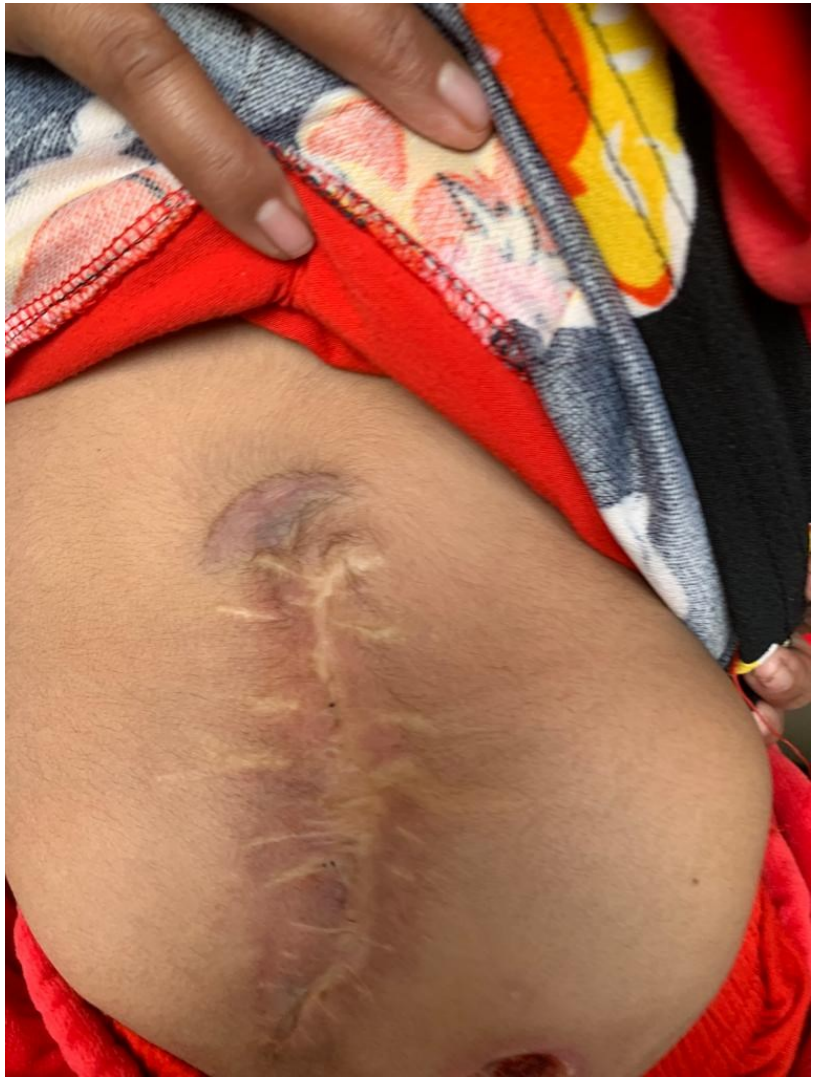




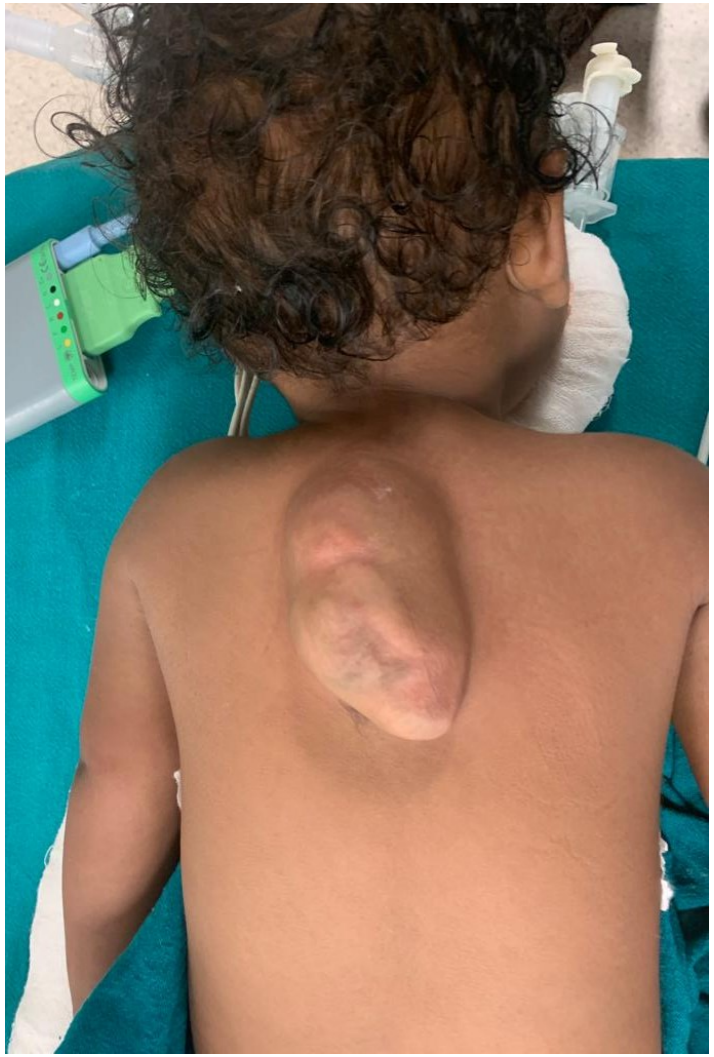


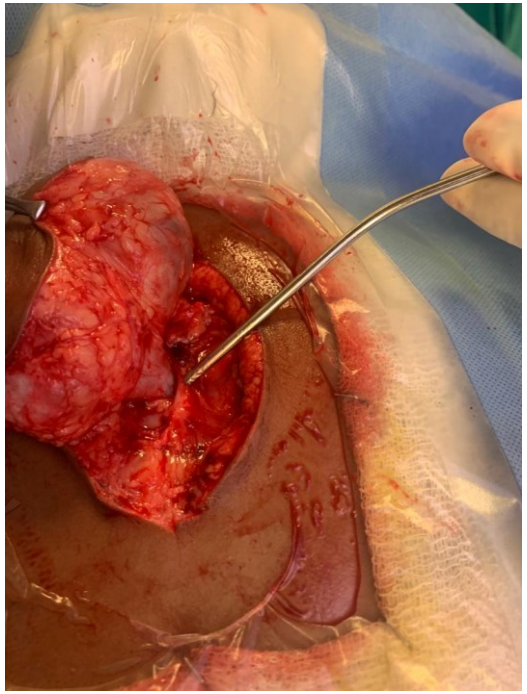


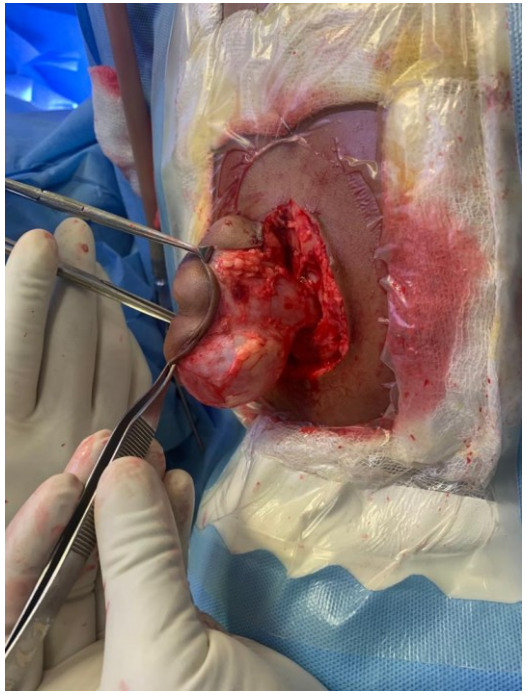
F/U after 3 years

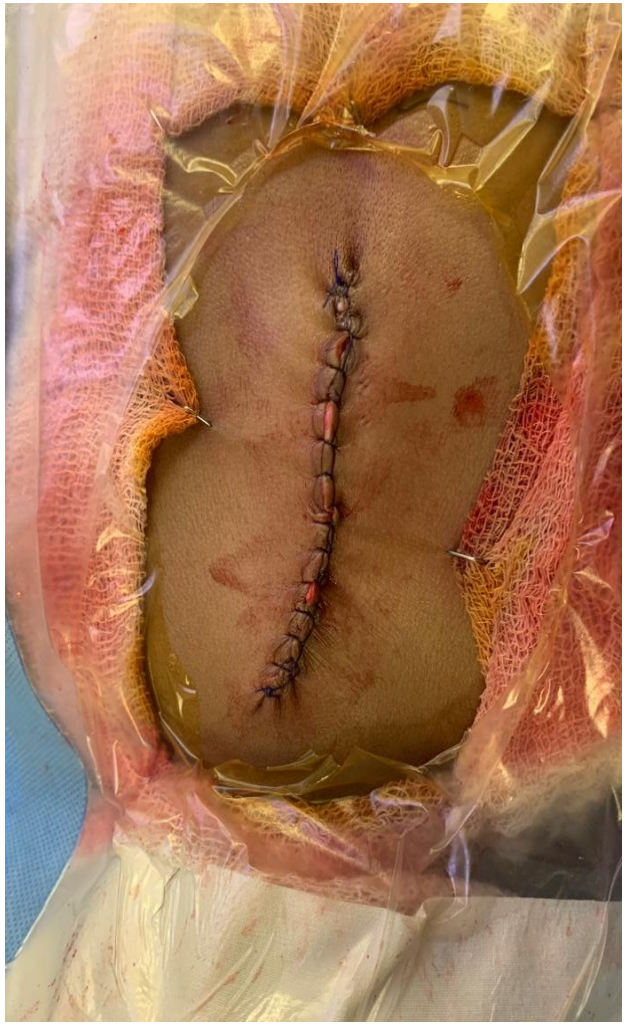


Cervico-doral MMC

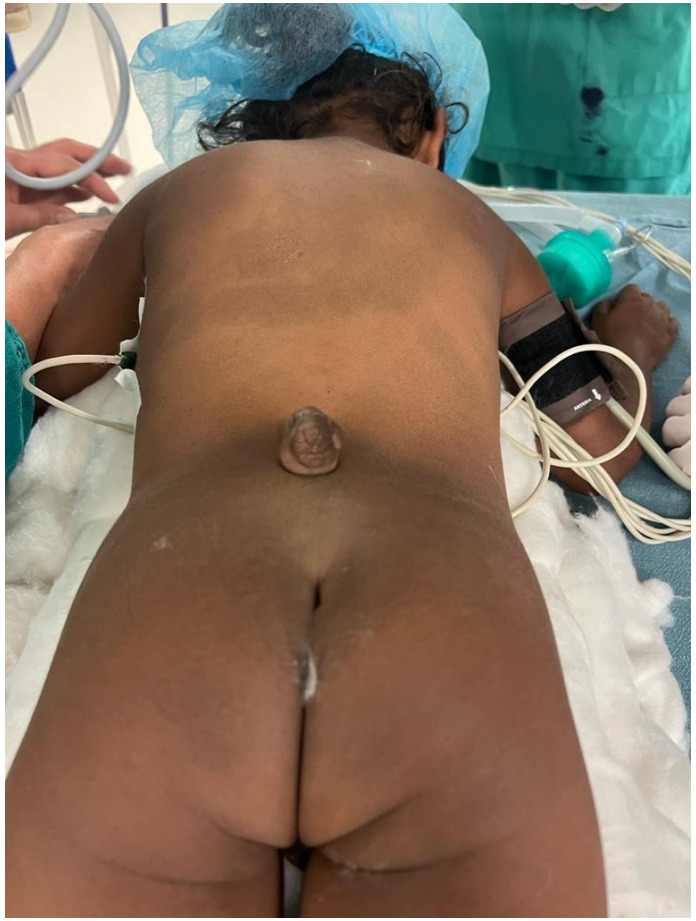


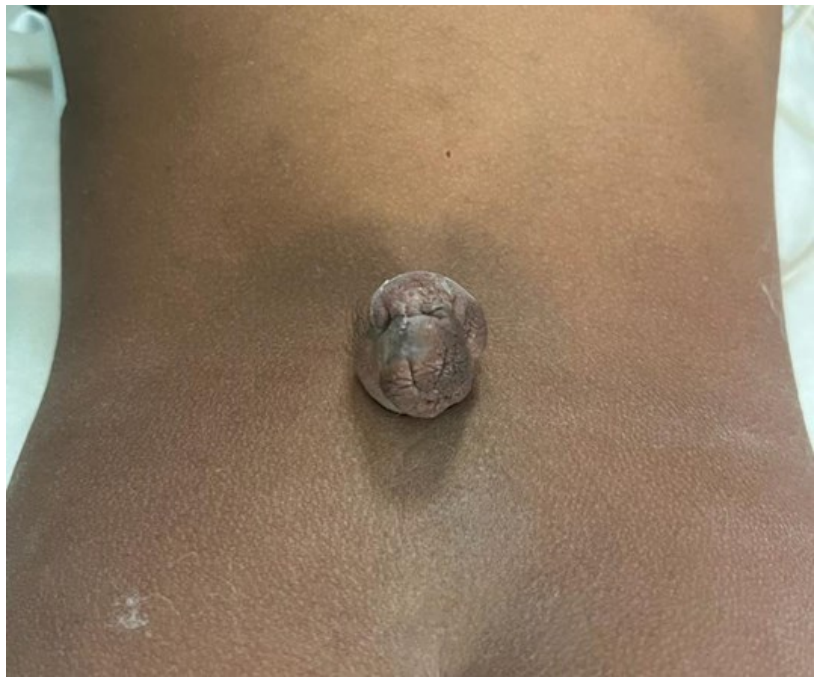


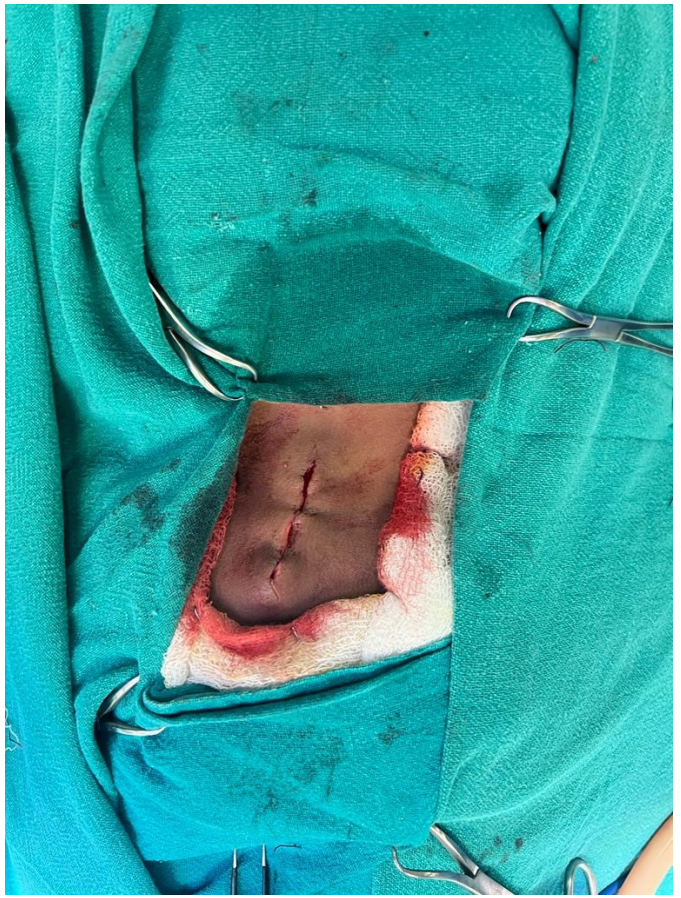




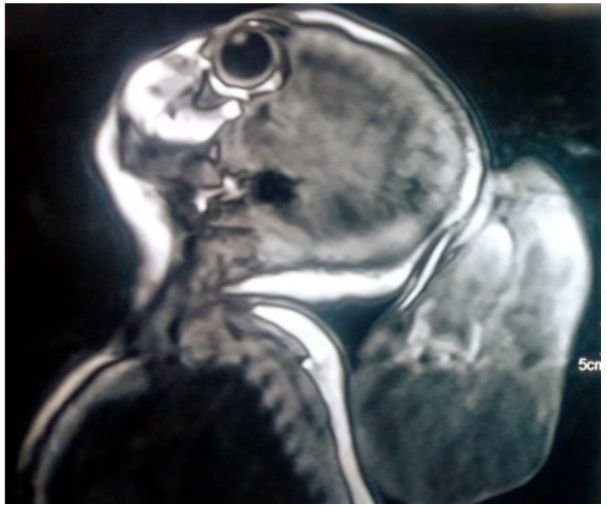
Lumbar-MMC

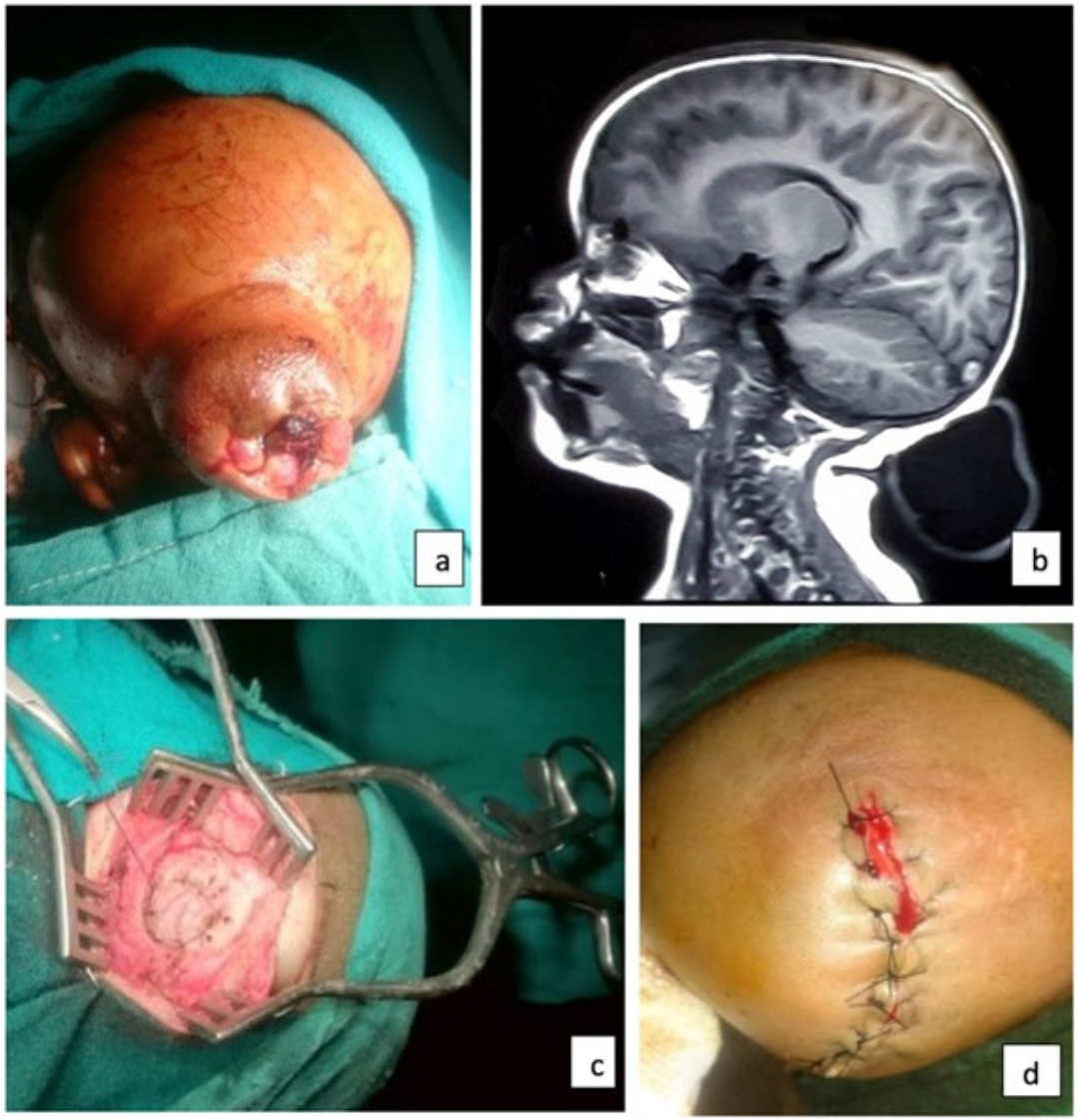


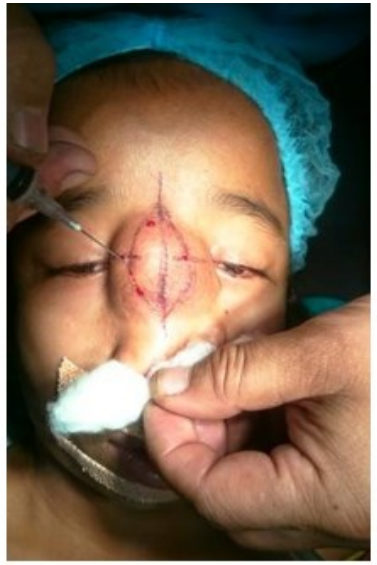


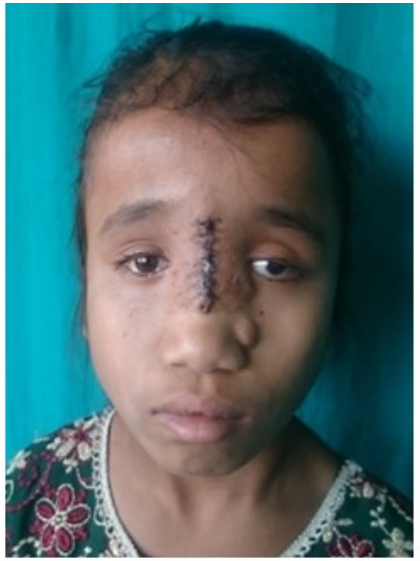


Encephalocele

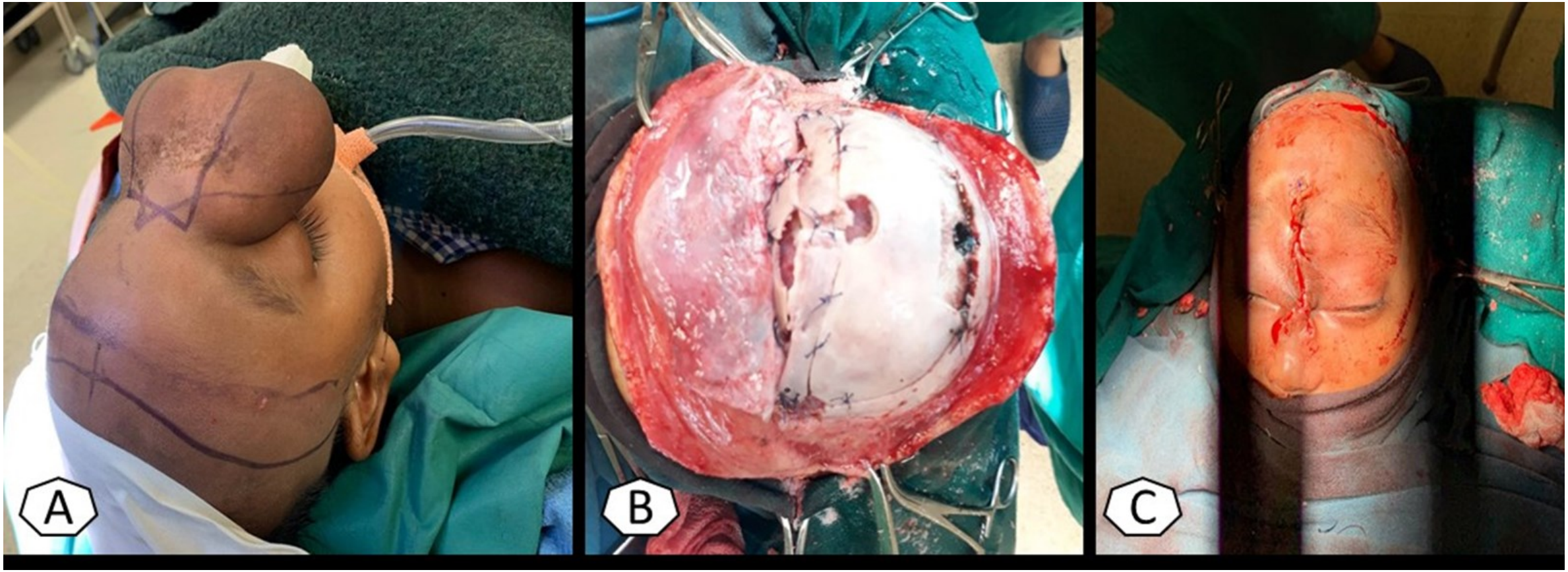




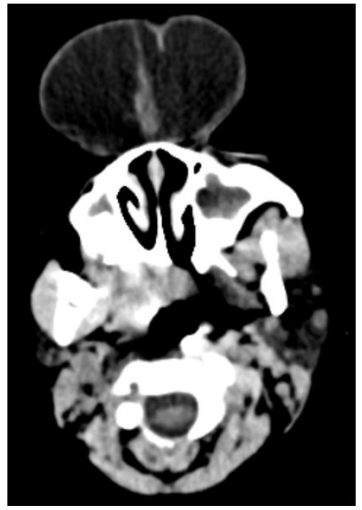
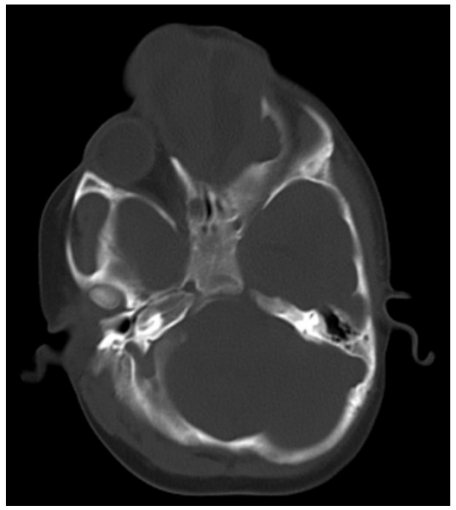


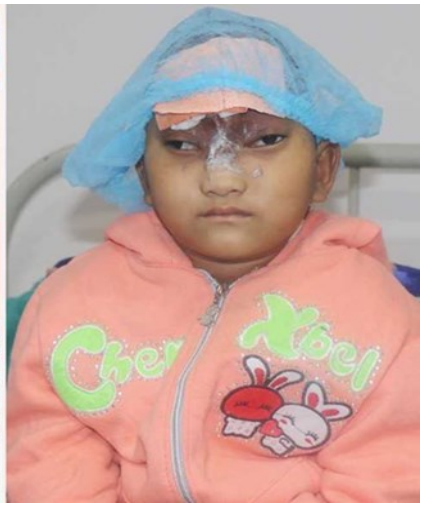
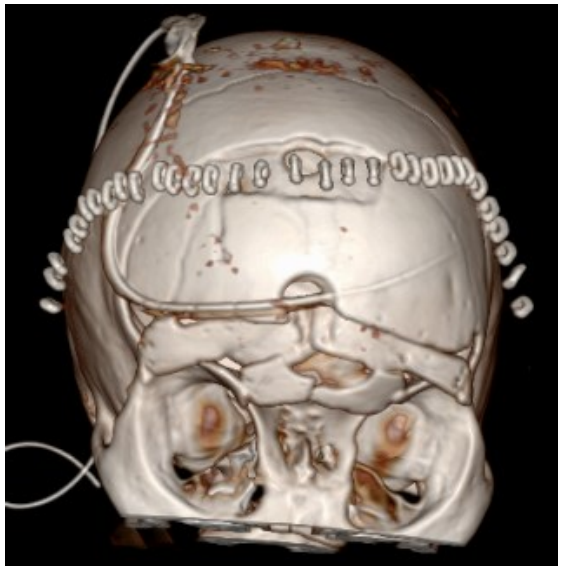
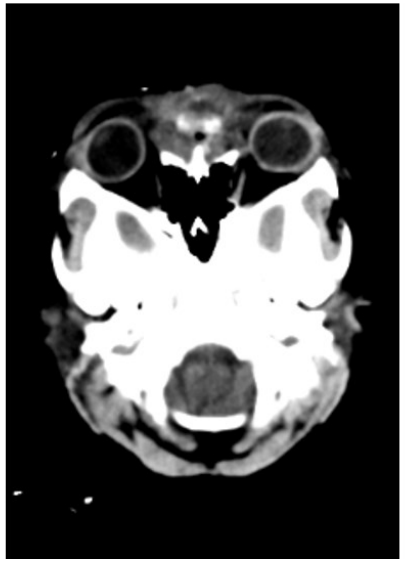
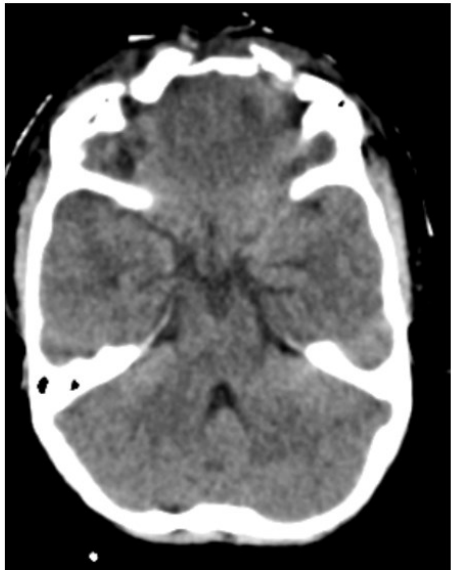
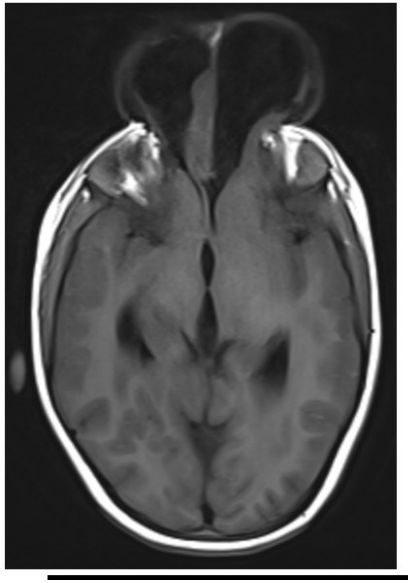
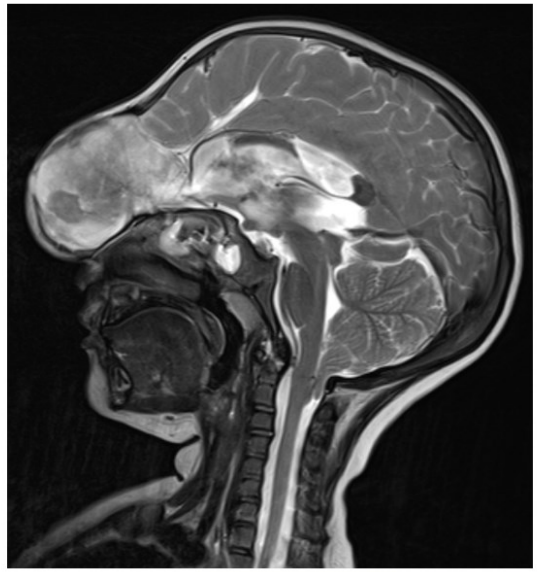
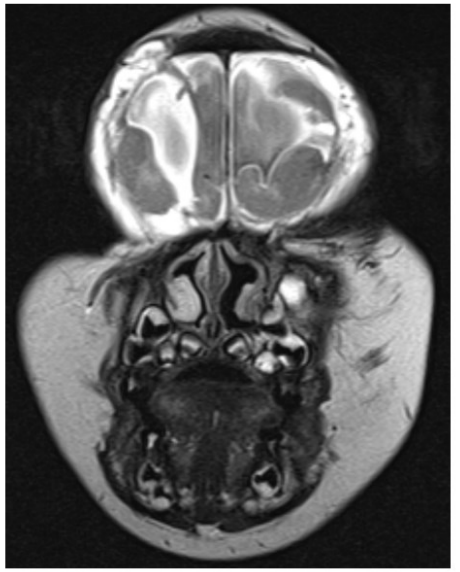


Frontonasal Encephalocele

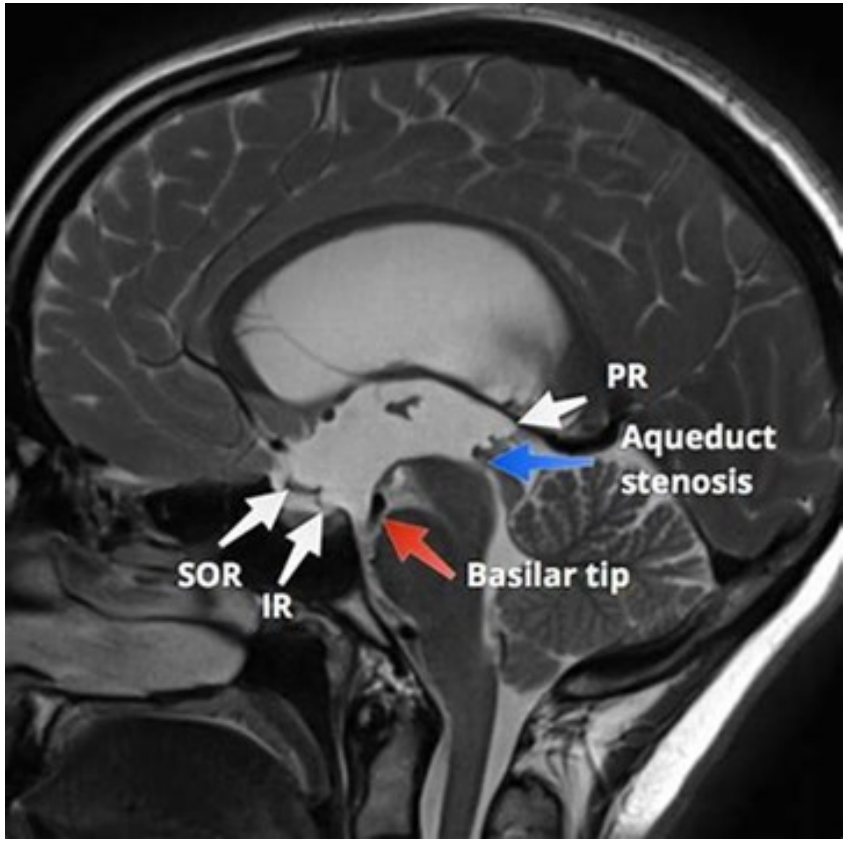


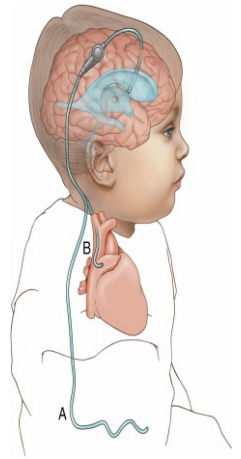
Radio imaging

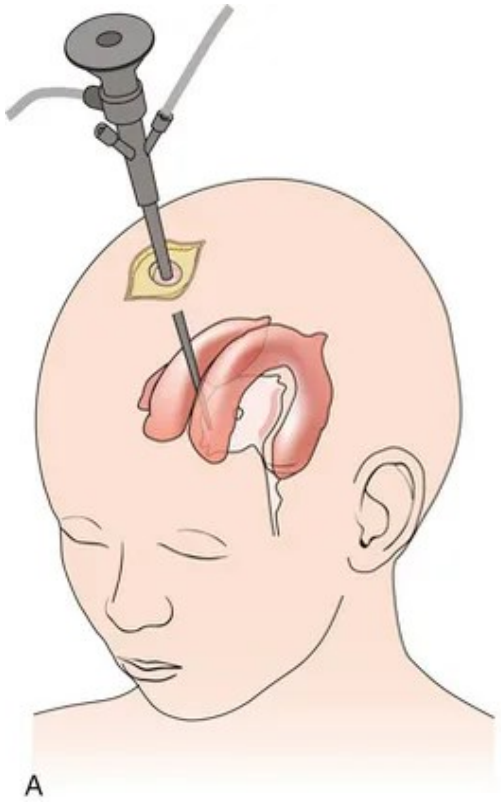



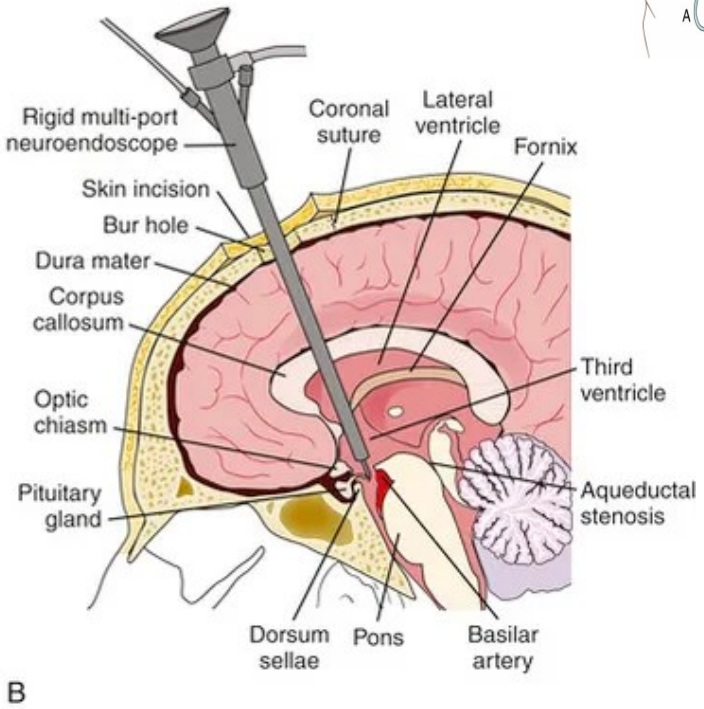








Hydrocephalus and ETV

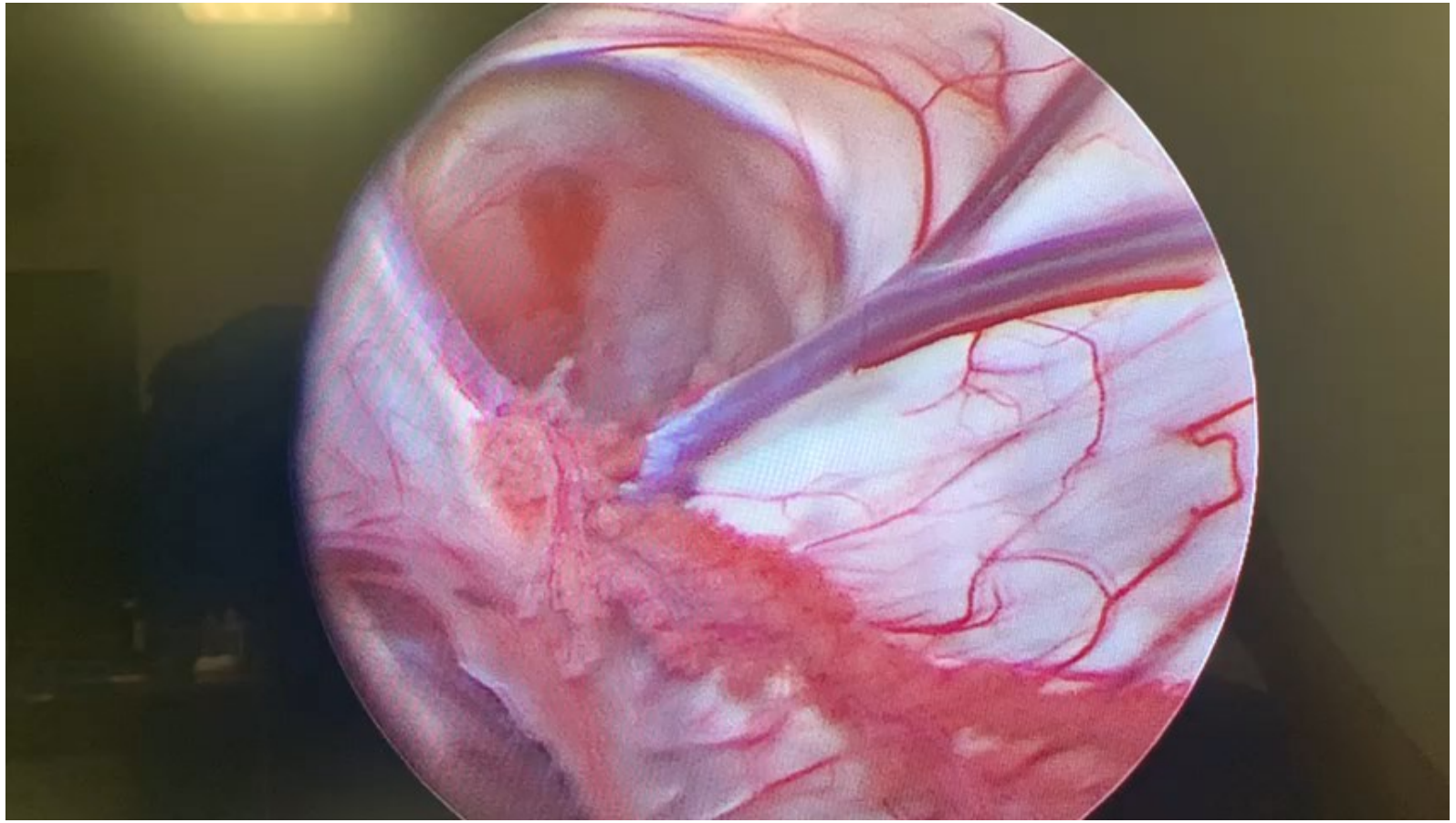








Illustrative ETV for Aqueduct Stenosis



https://youtu.be/3dHl-V51nDw (Dr. Prakash Kafle)

Glimpse of the recent publication(NINS)

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Original Article

Surgical Management and Early Outcome of Encephalocele

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Abstract

Background
There are limited studies pertaining to management of encephalocele in Nepal. So the present study seems justifiable to bridge the gap in the literature on encephalocele from Nepal on its clinical profile and early outcome. This study aims to characterize the clinical profile, management and outcome of largest series of encephalocele at tertiary care center in Nepal.

Materials and Methods
A retrospective analysis of encephalocele managed surgically at two tertiary care centers between 2015 and 2020 was performed.

Results
Total of 25 cases was surgically managed in the present study. The median age of study population was 2.5 months. There were 11 male and 14 female with male to female ratio of 1:1.26. Occipital encephalocele was the most common variant. Lump in the head (n=11) was the commonest clinical presentation followed by hypertelorism (n=10). One patient presented with cleft lip and one had CSF discharge in a case of occipital encephalocele. Bony defect was the common radiological findings. Excision and repair was the most common mode of surgery leading to good outcome. Mortality rate was 4% with morbidity of 20%.

Conclusion
Early surgical excision and tight dural closure with repair of bony defect is the standard treatment with relatively good outcome.

Keywords: *Cerebrospinal Fluid, Encephalocele, Neural tube defect, Occipital Mass*

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Joubert Syndrome: A Case Report

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Joubert syndrome (JS) isa rare autosomal recessive neurodevelopmental disorder involving cerebellarvermis and brainstem,marked byagenesis of cerebellar vermis, ataxia, hypotonia,oculomotor apraxia, neonatal breathingproblems and mental retardation. Magnetic Resonance Imaging (MRI) revealsthe characteristic *Molar tooth* sign of midbrain and *Batwing* appearance of rostral fourthventricle.

Key Words: Joubertsyndrome, hypotonia, vermianagenesis, molar tooth sign, bat wing appearance

Joubert syndrome is a rare autosomal recessive genetic neurodevelopmental disorderthat affects the area of brain that controls balance and coordination.[6]The common typical clinical manifestations are abnormal respiratory pattern (hyper apnea), occulomotor findings, low muscle tone (hypotonia), lack of muscle control (ataxia), developmental retardation with evidence of neuropathologic abnormalities of cerebellum and brainstem.[6]Historically, it was first described by Dr Marie Joubert, French Neurologist in 1969. Shedescribed four siblings with cognitive impairment, ataxia, episodic tachypnea, eye movement abnormalities, and cerebellar vermian agenesis in a large French-Canadian family with consanguinity traced 11 generations to a common ancestor. This disorderis characterized by congenital malformation of the hindbrain and a broad spectrum of other phenotypic

Spinal Dysraphism: Common Entity in Pediatric Neurosurgery

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Introduction: Neural tube defects are among the most common congenital malformations and a major cause of health problems in surviving children, especially in developing countries. Although the incidence of spinal dysraphism has significantly decreased over the last few decades, all over the world; however, the incidence is much higher in developing countries with poor socioeconomic status. The social and economic impact of this disease is not well documented; however, up to 75% of adult survivors may be dependent on parents or other care providers.

Aims and Objectives: The aim of this study is to review the demographic profile, clinical pretentions, surgical management and short term outcome of patients presenting with spinal dysraphism

Methodology: This is a prospective observational study of cases of spinal dysraphism managed surgically over the period of 2 years from March 2014 to February 2016 in Department of Neurosurgery at Tribhuvan University Teaching Hospital (TUTH), Kathmandu Nepal.

Results: Out of total 97 cases, there was male preponderance. In about 40% of population there was no history of proper ANC visit and most of them were from low economic status. Lump on the back was the commonest clinical findings. Lumbar Myelomeningocele was the commonest anatomical location of dysraphism. More than one third of patients needed CSF diversion postoperatively.

Conclusion: Myelomeningocele is a common NTDs. Open dysraphism may not always present as a lump. Delay in seeking medical attention may be due to illiteracy. None of the mother had taken folic acid prior to conception.

Key Words: hydrocephalus, myelomeningocele, spinal dysraphism

The term Spina dysraphism refers to a group of congenital anomalies of spine in which midline structure fails to fuse.[8] They are commonly known as neural tube defects (NTDs). It is among the most common congenital malformations and a major cause of health problems in surviving children.[7] Among them myelomeningocele represents the most serious form of dysraphism, a so called apert or open form involving the

Summary

- CNS –BD is usually Syndromic
- Multidisciplinary team is a must
- Most NTD with Neurological impairment don’t return to Normal
- No metanalyses on genetic studies
- Studies are required to establish the genetic and environmental risk factor identifications
- Prevention remains the best strategy in the management of NTDs

world birth defects day march 3

World Birth Defect day

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