

BIRTH DEFECTS

**A cross sectional study
AT**

**Nobel Medical College, Kathmandu
University**

Department of Obstetrics and Gynaecology

Presenter: Dr Munjal Yadav, Lecturer

BIRTH DEFECTS

Key Facts

1. According to WHO report, about **303 thousand** newborn deaths occur **within 4 weeks** of birth **every year** worldwide from congenital anomalies.
2. Congenital anomalies associated with-
 - Low Birth Weight
 - Prematurity
 - Multiparity
 - Consanguinity and
 - Caesarean delivery

Key Facts

3. Common defects-

- Neural tube defects
- Down syndrome

4. Common cause (to identify the exact cause)

- Genetic
- Infectious
- Nutritional
- Environmental factors

Key Facts

5. Global prevalence of serious birth defect
 - 3-6%
6. Nepal prevalence
 - 0.52% (95% CI: 44.0–61.0)
 - 7% of all neonatal deaths (NDHS)

Introduction

- **Synonyms:** Congenital anomalies/birth defects/ congenital disorders/ congenital malformations
- **Definition:** Structural or functional anomalies
 - That occur during intrauterine life and
 - Can be identified
 - prenatally
 - at birth, or
 - later in infancy

Selected Major Birth Defects

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

Selected external minor defects

Absent nails
Accessory tragus
Anterior anus (ectopic anus)
Auricular tag or pit
Bifid uvula or cleft uvula
Branchial tag or pit
Camptodactyly
Cup ear
Cutis aplasia (if large, this is a major anomaly)
Ear lobe crease
Ear lobe notch
Ear pit or tag
Extra nipples (supernumerary nipples)
Facial asymmetry
Hydrocele
Hypoplastic fingernails toenails
Iris coloboma

Lop ear
Micrognathia
Natal teeth
Plagiocephaly
Polydactyly type B tag, involves hand and foot
Preauricular appendage, tag or lobule
Redundant neck folds
Rocker-bottom feet
Single crease, fifth finger
Single transverse palmar crease
Single umbilical artery
Small penis (micropenis)
Syndactyly involving second and third toes
Tongue-tie (ankyloglossia)
Umbilical hernia
Undescended testicle
Webbed neck (pterygium colli)

Cross Sectional Study

Methodology

- Design: Descriptive cross sectional study
- Selection criteria
 - Hospital delivery
 - All modes of delivery
 - Congenital anomalies in prenatal USG
 - Congenital malformation at birth
- Duration of study: 1 year (May 2019 - May 2020)

Results

- Total number of birth defects = 104
 - Male 56 + Female 44 + 4 Ambiguous
- Incidence = 1.25%
- Mean age = 25.88 years
- Mode: 60% in 20-30 years
- Unbooked = 87
- Identified after admission/delivery = 44

Relation of consanguinity and birth defects (in 12 Muslim women)

Congenital Disorders	Frequency
Anencephaly	5
Diaphragmatic hernia	1
Hydrocephalus	1
Hydrops Fetalis	1
Club foot	1
Phocomelia	2
Undescended testis	1

Major system involved (N=100)

Major system involved	%
Cleft lip/palate	6
CNS	37
Digestive	10
Genitourinary	7
Hydrops fetalis	9
Lymphatic	3
MSK	13
Renal	9
Respiratory	4
Others	2

Types of birth defects

Significant findings	Frequency
Hydrocephalus	13
Ambiguous genitalia	4
Anencephaly	17
Anorectal atresia	2
Aqueductal stenosis	1
Hydronephrosis	5
Hypospadias	1
Meningomyelocele	1
Omphalocele	6

Types of birth defects

Significant findings	Frequency
Klippel-Feil Syndrome	1
Cleft lip/palate	9
Club foot	7
Congenital Cystic Adenomatoid Malformation	1
Congenital High Airway Obstruction Syndrome	1
Corpus callosum agenesis	2
Cyclops	1

Types of birth defects

Significant findings	frequency
Cystic Hygroma	3
Diaphragmatic hernia	2
Hydrocele	1
Polydactyly	2
PUV with Hydronephrosis	1
PUV with UB Diverticulum	2
Renal Agenesis	1
Skeletal Dysplasia	2
Spina Bifida	1
Undescended testis	1
Hydrops Fetalis	10

Results

- The vast majority of the cases ($n=76$) were diagnosed only during the third trimester.
- Almost more than half ($n=58$) of the pregnant female didn't use folic acid.
- Among congenital anomalies related to the central nervous system, 35.13% didn't have a history of use of folic acid.

Results

- Regarding Medical history, among them 4 were overt diabetic, 1 case was VDRL positive and 2 cases were hypertensive.
- Pregnancy was terminated through normal vaginal delivery in 75 of them whereas 20 had to go through cesarean section. One even had to go through a hysterectomy.

Mode of termination of pregnancy

Mode of termination	Frequency(n)
CS	19
Vaginal Delivery	78
VBAC	2
Caesarean Hysterectomy	1
Total	100

Some cases

Hydrocephalus



Cystic Hygroma



Hydrocephalus



Omphalocele



Phocomelia

- Underwent C-section for fetal distress.
- Unbooked referred case with thick MSL
- No h/o Antenatal visits



Encephalocele



Meningocele



Ambiguous genitalia



Body Stalk Anomaly



Skeletal Dysplasia



Hydrops fetalis



Cleft lip and Palate



Discussion

- Exact magnitude of birth defect related events is still unrevealed.
- Prevalence high in Nepal but true magnitude is still Unknown
- Current study aim is to establish the prevalence and pattern of congenital anomalies through obstetric Ultrasonography in unselected population visiting NOMCTH.

Prevention

- Preconceptional
- Antenatal period
 - First trimester
 - Second Trimester
- Postnatal period
 - Screening of newborn

Conclusion

- Early detection of major anomalies will offer to the parents the option of an earlier, safer and psychologically less traumatic termination of pregnancy.
- Further helps in planning interventions and necessary management.
- Health care awareness

Take Home Message

I care....

Because birth defects affect all of us

AND

**Because raising awareness can help
babies around the world.**

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Thank You