



ST PAULS HOSPITAL MILLENIUM MEDICAL COLLEGE

MAGNITUDE OF CONGENITAL MALFORMATIONS AMONG NEONATES
ADMITTED TO SPHMMC, A ONE YEAR REVIEW

BY SHALOM TSEGAYE

PUBLIC HEALTH ADISOR: MISS MESRACH AYALEW

CLINICAL ADVISOR: DR ABATE AND DR GIZESH

A STUENT RESEARCH PAPER TO BE SUBMITTED TO THE
DEPARTMENT OF PUBLIC HEALTH, SPHMMC. IN PARTIAL
FULFILLMENT FOR THE REQUIRMENTS FO DEGREE OF DOCTOR OF
MEDICINE

SEPTEMBER 2017

ADDIS ABEBA, ETHIOPIA



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1. Acknowledgment

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2. Abstract

Background: Congenital anomalies are defined as structural or functional anomalies that are present at birth. Congenital anomalies can affect any part of the body or any physiologic system. Worldwide the prevalence of congenital anomalies is 3-4%.

Objectives: The study was aimed at determining the magnitude of congenital anomalies and the associated factors in St Paul's hospital millennium medical college neonatal intensive care unit.

Methods: The study was carried out in SPHMMC NICIU, a cross sectional retrospective review of charts of patients diagnosed with congenital anomalies on HMIS at NICU over a year period. The data was cleared and entered into SPSS, chi square was used to analyze associations between the variables.

Result: There was a total of three thousand three hundred and fifteen neonatal admissions to the NICU over the one year study period. 94 CA were picked. Fifty two (55.3%) were male and forty two (44.7%) were female. Most of the mothers (83%) were in the age group of 20-35. 70.2% of the mothers were from region 4, 28.7% from region 14, and 1.1% from region 3. 71.3% of the mothers had ANC follow-up and also received iron supplements. 45.7% of CA occurred in para 1 mothers, 39.4% in parity of 2-4 and 1.1 % in above 4 parity. Most of the neonates (80.9%) with CA were in the gestational age between 35-40 weeks. The percentage of neonatal anomalies in SPHMMC was 2.83%. The most common anomaly was NTD(32.1%) followed by musculoskeletal and orofacial.

Conclusion: The prevalence and pattern of CA is not well researched in our country. This impedes policies and society to address this issues. However, the 2.83% at SPHHMMC is an indicator of a problem. NTD was the most common CA. this should create a root based changes starting from folic acid supplementation in reproductive aged women, creation of awareness on NTD and effect of folic acid supplementation, increases on awareness on antenatal care for mothers.

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4. Introduction

The development of the embryo is a complex process from the time of fertilization to the formation of all cells, tissues and organs. In pregnancy, there is a critical period of organ formation during embryogenesis. Interference during this period due to intrinsic and extrinsic factors lead to birth defects or congenital malformations. [1]

Congenital anomalies are defined as structural or functional anomalies that are present at birth. Congenital anomalies can affect any part of the body or any physiologic system.

Some of the congenital anomalies might be life threatening, may impair function or may interfere with cosmetic values. Accordingly, malformations can be classified as major and minor. Minor malformations have mostly cosmetic significance. They rarely are medically significant or require surgical intervention. They represent part of the normal variation in the general population. Examples of minor anomalies include ear tags, clinodactyly (incurving of the fifth finger). Major malformations are those that have medical and/or social implications. These often require surgical repair.

The long term disability caused by congenital anomalies may have a significant impact not only on a child's wellbeing and development in addition to families and society at large. Congenital malformations are one of the cause of infant mortality worldwide. On 2015, WHO estimated that worldwide out of the 2.86 million deaths during the neonatal period, 303,000 deaths were due to congenital anomalies. [2]

Maternal illness has also been associate with congenital malformations. Pregestational diabetes mellitus is associated with a two to three-fold increase in risk of congenital anomalies, including congenital heart disease and spina bifida, and, less commonly, caudal regression and focal femoral hypoplasia.

Maternal drug ingestion, both medical and recreational, can cause adverse fetal and neonatal outcomes. Women are sometimes prescribed medications that are contraindicated in pregnancy because of evidence that the risk of birth defects outweighs any potential benefits of the medication. These women should be counseled about the adverse effects of these medications and the importance of consistent use of effective contraception. Some common teratogenic medications

include: Angiotensin converting enzyme inhibitors, Anticonvulsant agents, antineoplastic agents

Congenital malformation is one cause of neonatal mortality and morbidity. On 2015, WHO estimated that worldwide out of the 2.86 million deaths during the neonatal period, 303,000 deaths were due to congenital anomalies. About 43% of congenital malformations have associated factors identified.[3]

Many types of congenital anomalies present to the pediatric department. These patients usually have prolonged hospital stay due to the severity of their illness and existing co-morbidities. There is scarcity of research on this topic in Africa, especially Ethiopia. As such it is difficult to determine the burden of conditions. It also hinders the health care system to address this issues.

Worldwide the prevalence of congenital malformation is 3-4%. In a retrospective study done in central and northwest Ethiopia from 2010-2014, 319,776 medical records of children aged 0–17years were analyzed. Of these, 1.9% of children were diagnosed as having birth defects. The majority (58.5%) of the children were male and 41.5% female. [4]

Among the participants of the study the proportion of birth defects ranged as follows: orofacial (34.2%), neural tube (30.8%), upper and lower limb (12.8%), cardiovascular system (10.3%), digestive system and abdominal wall (4.8%), unspecified congenital malformations (2.5%), Down syndrome (2%), genitourinary system (2%), head, face, and neck defects (0.4%), and others (0.3%).

Among the orofacial clefts the proportion of cleft lip, cleft lip and palate, and cleft palate was 70.2, 21.2, and 8.3% respectively. The frequency of orofacial clefts by laterality showed that unilateral cleftlip was the most frequent (63.2%) and bilateral cleft palate the least frequent (0.7%).

In a study conducted in Tikur Anbessa hospital, charts of 115 children with neural tube defect was reviewed retrospectively. The most common defect was meningomyelocele 64.4%. It was followed by meningocele 18.3% and encephalocele 13%. [5]

In a cross-sectional study done on all the newborns who were born during 9 months period from January to December 2009 in Lebanon hospital, the prevalence of congenital anomalies was 2.4%. Cardiovascular system defects and limbs anomalies each accounting for 16.6%.[6]

The cause of congenital anomalies is unknown in 43% of the cases. Other causes of congenital anomalies are genetic and nongenetic. Genetic abnormalities include chromosomal disorders (eg, Down syndrome); single gene (monogenic) disorders, including those that are autosomal recessive (eg, cystic fibrosis), autosomal dominant (eg, Marfan syndrome). Nongenetic etiologies include environmental factors like teratogens or drugs like anticonvulsants, alcohol abuse, cigarette smoking. Factors like previous siblings with congenital anomalies, maternal folate deficiency, maternal diabetes are also associated.

The study of birth defects and their etiology is termed teratology. The word teratogen is derived from the Greek teratos, meaning monster. A teratogen may be defined as any agent that acts during embryonic or fetal development to produce a permanent alteration of form or function. Thus, a teratogen may be a drug or other chemical substance, a physical or environmental factor such as heat or radiation, a maternal metabolite such as diabetes, a genetic abnormality, or an infection.

Drugs like anticonvulsant, acetylcholinesterase inhibitors and antineoplastic are unsafe in pregnancy. None of the anticonvulsant drugs are considered truly “safe” in pregnancy. This is because most medications used to treat epilepsy have been proven or suspected to confer an increased risk for fetal malformations. Both major and minor malformations are more common in fetuses exposed to antiepileptic drugs (AEDs) in utero compared with offspring of untreated women with epilepsy and women without epilepsy. The overall risk of major malformations is 4 to 6 percent in AED use. CA associated with AEDs are neural tube, congenital heart and urinary tract defects, skeletal abnormalities, and cleft palate.[7]

Alcohol Ethyl alcohol is a potent and prevalent teratogen. Alcohol is one of the leading cause of preventable birth defects in the United States. The alcohol effect on a fetus is known as fetal alcohol syndrome. Clinical Characteristics of Fetal alcohol syndrome include dysmorphic facial features (Small palpebral fissures, thin vermilion border and smooth philtrum), pre- or postnatal growth impairment, and central nervous system abnormalities. Other alcohol-related major and minor birth defects include cardiac and renal anomalies, orthopedic problems, and abnormalities of the eyes and ears.

Folic acid is known to be necessary for growth and function of human cells as it is crucial for biosynthesis and methylation of deoxyribonucleic acid (DNA) and ribonucleic acid (RNA). This is important for cell division, differentiation and regulation of gene expression especially when there is rapid cell division like

during embryogenesis. Folic acid is crucial for normal brain and spinal cord development during the first 4 weeks of gestation. Periconceptional use of folic acid has been proved to cause significant reduction of the risk for neural tube defects and other congenital anomalies like orofacial clefts, congenital heart diseases, urinary tract, limb and digestive system anomalies.

Exposure to infectious agents can result in a variety of problems in the fetus and neonate, including malformations, congenital infection, short and long-term disability, and death. The pathogenesis of the fetal defects is usually direct invasion of fetal tissues leading to damage from inflammation and cell death. Agents known to be toxic to the fetus or embryo are toxoplasmosis, rubella, cytomegalovirus, herpes, and syphilis (the so-called TORCH infections). [8]

Cigarette smoke contains a complex mixture of nicotine, cotinine, cyanide, thiocyanate, carbon monoxide, cadmium, lead, and various hydrocarbons. In addition to being fetotoxic, many of these substances have vasoactive effects or reduce oxygen levels. It has increased risk for cardiac anomalies has also been reported. A study using data from the National Vital Statistics System of more than 6 million births found an association between maternal smoking and hydrocephaly, microcephaly, omphalocele, gastroschisis, cleft lip and palate, and hand abnormalities.

These research will determine the prevalence and pattern of congenital anomalies in St Paul hospital millennium medical college. It also determines the factors associated with congenital anomalies. Since St. Paul's hospital millennium medical college is tertiary hospital with a catchment population of 5 million, this research can shed the light on the national level. This can help to develop strategies improves detection, management and rehabilitation of patients. This includes medical and surgical interventions as well as programs that increases the awareness of parents. It could also assist in constructing special needs for children with this conditions.

5. Methods

A cross sectional study design will be used to assess the magnitude of CA among all neonates admitted the NICU in SPHMMC over a year period from January 1, 2016- December 31, 2016.

Study Area

6.1 Data collection

Data was collected from hospital records from January 1, 2016 – December 31 2016. Neonates with CA were picked from NICU registry (HMIS). The registry includes a total of 14 different diagnosis. Upon admission of every neonate to the NICU, a diagnosis is established and recorded to the registry. There was a total of 3,315 neonates admitted in the study period. Out of which, 94 had CA. Retrospective chart review of this 94 neonates was done. A structured check list was used to identify patient information. The check list included the maternal sociodemographic variables (of age, sex, region), maternal ANC follow-up, iron supplementation, parity, pregnancy, neonatal GA, weight, diagnosis of CA.

6.2 Data processing and analysis

Data was entered to SPSS. Data was cleaned and entered. Chi square was used to analyze the association of dependent and independent variables with the presence of congenital malformations. The 95% confidence intervals were determined and associated factors with p-value of less than 0.05 was considered significant.

6. Ethical consideration

All information regarding the patient will remain confidential. Patient records will be kept such that the identity of the patient will not be disclosed. Patient information was entered with identification code.

7. Result

There was a total of three thousand three hundred and fifteen neonatal admissions to the NICU over the one year study period. Out of these, Ninety four were diagnosed with congenital anomalies. Fifty two (55.3%) were male and forty two (44.7%) were female.

Most of the mothers (83%) were in the age group of 20-35. 70.2% of the mothers were from region 4, 28.7% from region 14, and 1.1% from region 3. 71.3% of the mothers had ANC follow-up and also received iron supplements. 45.7% of CA occurred in para 1 mothers, 39.4% in parity of 2-4 and 1.1 % in above 4 parity. 95.7% of pregnancies were singleton, 3.2% were twins and 1.1% were triplets. Most of the neonates (80.9%) with CA were in the gestational age between 35-40 weeks.

Table.1 Variables frequency and percentage

Variables		Frequency	Percentage
Sex	Male	52	55.30%
	Female	42	44.70%
Gestational age	25-30wks	1	1.10%
	30-35wks	17	18.10%
	35-40wks	76	80.90%
Mother's age	<20	9	9.60%
	20-35	78	83%
	>35	7	7.40%
ANC	YES	67	71.30%

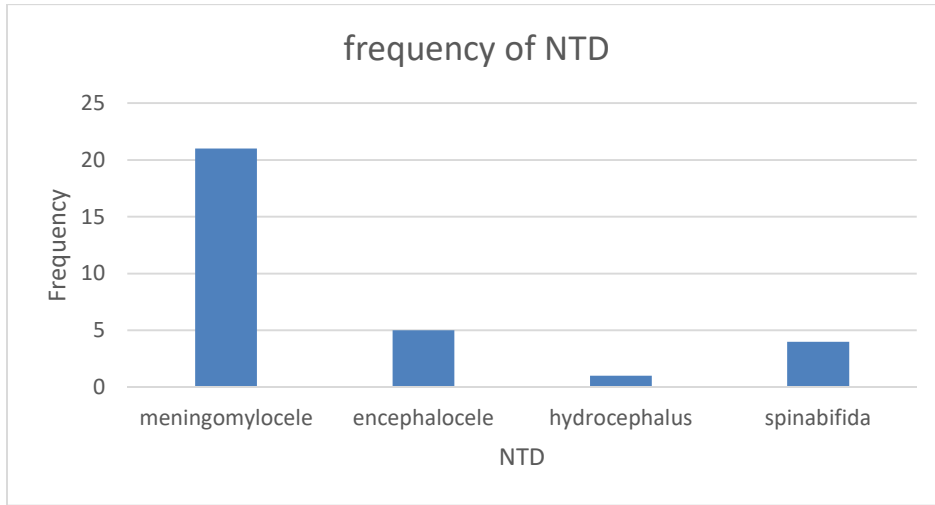
	NO	27	28.70%
Iron	YES	66	70.20%
	NO	28	29.80%
Pregnancy	Singleton	90	95.70%
	Twins	3	3.20%
	Triplet	1	1.10%
Parity	One	43	45.70%
	two-four	37	39.40%
	>four	14	14.90%
Neonatal Weight	1000-1500kg	5	5.30%
	1501-2000kg	9	9.60%
	2001-2500kg	25	26.60%
	2501-3000kg	32	34%
	3001-3500kg	19	20.20%
	3501-4000kg	4	4.30%

Distribution of congenital anomalies

NTD

32.9% of the CA were NTD. Out of which, 67.7% of neonates had meningocele, 16% had encephalocele, 3.22% had hydrocephalus and 12.9% had spina bifida.

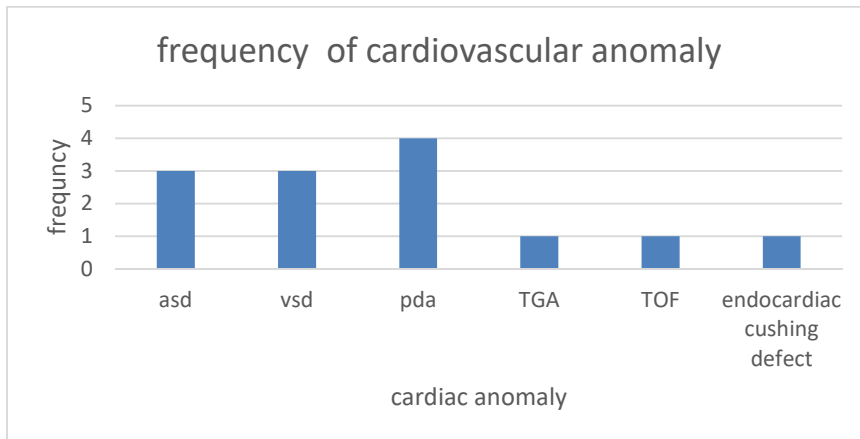
Graph 1: frequency of NTD



Cardiac anomaly

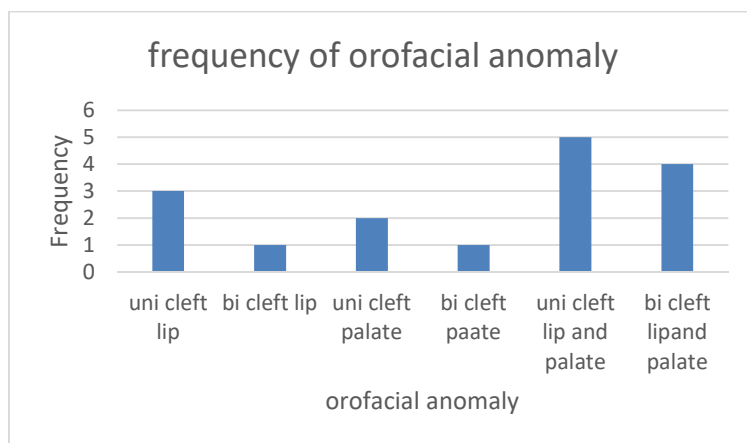
13.8% of neonates had cardiac anomalies. Out of which, 23% was ASD, another 23% was VSD, 30.7% was PDA and TGA, TOF and endocardial cushioning defect each were 7.7%.

Graph:2 frequency of cardiovascular anomaly



Orofacial anomaly

17% of the neonates had Orofacial anomaly. 18.75% had unilateral cleft lip, 6.25% had bilateral cleft lip, 12.5% had unilateral cleft palate, 6.25% had bilateral cleft lip, 31.25% had unilateral cleft lip and palate and 25% had bilateral cleft lip and palate.



Respiratory anomaly

- 2.1% of the neonates had Respiratory anomaly. TEF was the only respiratory anomaly picked with frequency of 2.

GI Anomaly

- 14.9% of the neonates had GI anomalies. Of which 21.4% had HUS, 42.9% had imperforate anus, 14.2% had duodenal atresia and 21.4% had omphalocele.

Table 2: frequency of GI anomaly

GI anomaly	frequency
HUS	3
Imperforate	6

anus	
duodenal atresia	2
omphalocele	3

GUS anomaly

- 5.3% of neonates had GUS anomalies of which 80% had hypospadias and 20% had PCKD.

GUS anomaly	frequency
Hypospadia	4
PCKD	1

Musculoskeletal anomaly

19.1% of the neonates had MSS CA of which 5.5% had unilateral clubfoot, 77.8% had bilateral clubfoot and 16.7% had polydactyl.

MSS anomaly	frequency
Uni club foot	1
Bi club foot	14
Polydactyl	3

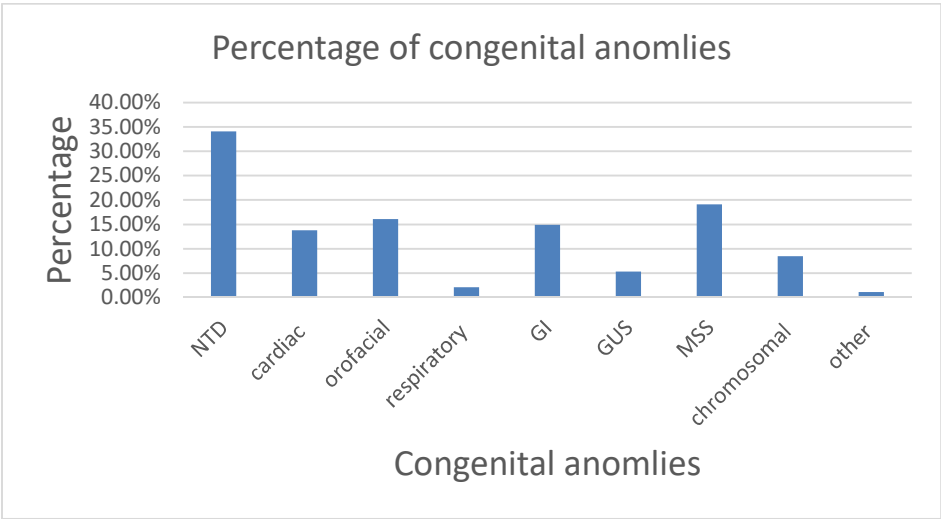
Chromosomal anomaly

- 8.5% of the neonates had chromosomal disorders of which 62.5% had downs syndrome, 25% had patu, 12.5% had noonan syndrome.

chromosomal disorder	frequency
Downs syndrome	5
Patu	2
Noonan syndrome	1

- There was one neonate with cystic hygroma, with 1%.

Distribution of congenital anomalies



Significant P value was found between weight and cardiovascular anomaly, sex and Gastrointestinal anomaly, orofacial and region, gestational age and genitourinary system anomaly and mothers age and chromosomal anomaly.

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Discussion

The percentage of neonatal anomalies in SPHMMC is 2.83% There was a total of three thousand three hundred and fifteen neonatal admissions to the NICU over the one year study period This finding is close to the prevalence of CA in research done in central and northwest Ethiopia by Taye et al. The prevalence of birth defect was 1.9 % with the most prevalent congenital anomaly being orofacial (34.2%) followed by NTD(30.8%) and musculoskeletal anomalies(12%).

This percentage is also comparable to the prevalence of CA done in pediatrics department of Gonder collage hospital (2.16%). The prevalence of CA is as well comparable with 2.4% in research done in Lebanon by Francine et al. The commonest detected anomaly was cardiovascular(16.6%) followed by musculoskeletal anomalies(16.6%).

There were more male neonates with CA(55.3%)as compared to female neonates(44.7%). Similarly, more male children (58.5%) were observed with CA than female ones (41.5%) in a study in northwest and central Ethiopia. Similarly, Mashuda et al, from Tanzania reported that 54.6% of the male and 44.9% of the female children were observed with congenital anomalies.

The most common CA is NTD in SPHMMC (32.1%). This percentage is slightly higher than NTD prevalence (30.6%) in studies done in central and northwest Ethiopia by Taye et al. Similar prevalence of NTD was found in research done at Tanzania. This relatively higher percentage could be due to the large catchment area of SPHMMC in Ethiopia. This also shows the folic acid deficiency in mothers, lack of awareness about prenatal folic acid supplementation.

The second most common anomaly in SPHMMC was musculoskeletal anomalies(19%).

Conclusion

The prevalence and pattern of CA is not well researched in our country. This impedes policies and society to address this issues. However, the 2.83% at SPHMMC is an indicator of a problem. NTD was the most common CA. this should create a root based changes starting from folic acid supplementation in reproductive aged women, creation of awareness on NTD and effect of folic acid supplementation, increases on awareness on antenatal care for mothers.

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