J. Dhaka National Med. Coll. Hos. 2019; 25 (01): 06-10

Original Article

Study of Major Congenital Anomalies in Neonates born at Dhaka National Medical Institute Hospital

Rashidul Karim[1], Sultan Uddin[2], Md. Shafiqur Rahman[3], Humayra Sultana[4], Syeda Farzana Rahat[5]

[1]Professor (c.c.), Department of Paediatric, Dhaka National Medical College, [2]Professor and Head, Department of Paediatric, Dhaka National Medical College, [3]Associate Professor, Department of Paediatric, Dhaka National Medical College [4]Assistant Professor, Department of Paediatric, Dhaka National Medical College, [5]Assistant Professor (c.c.), Department of Paediatric, Dhaka National Medical College

Abstract

Background: Congenital malformations are major contributors of neonatal mortality or life long disability. Major malformation accounts for 15% of neonatal death. It is a priority health problem in newborn. The objective was to study opted to know the frequency, the pattern of congenital anomalies, associated risk factors, various system involved and immediate outcome of congenital malformations in newborns.

Methods: This prospective hospital based observational study was carried out in the department of Paediatrics, Dhaka National Medical College and Hospital, Dhaka for a period of 3 years from 1st January, 2016 to 31st December, 2018. All congenital anomalous babies during the study period either detected before birth by ultrasonography of mother or detected at birth were included in this study. Diagnosis of congenital anomalies was based on clinical evaluation of newborn babies by the pediatrician and other appropriate investigations such as radiography, ultrasonography, echocardiography and chromosomal analysis etc.

Results: The anomalies in this study were divided into major and minor anomaly. During the study period 68 newborns with major congenital anomalies were included. Major anomalies identified involved the gastro-intestinal (GE) system (30.87) was found to be the commonest type of anomaly. Cleft lip and cleft palate (14.70%) was the most common anomaly seen in the gastro-intestinal system. Of the 68 major anomalous babies, 8 (11.76%) babies had multiple anomalies. Out of 68 newborn, 26.47% were still births and 73.53% were live births. Among the anomalous babies 67.65% were male and 32.35% were female. Of them birth weight less than 2.5 kg were 39.71% and weighing 2.5 kg or more were 60.29%. Out of total 68 mothers with major congenital anomalous babies, 58.82% of multiparas, more than half of the mothers (76.47%) aged <35 years, 52.94% of babies delivered <37 weeks of gestational age were found. 23 (33.82%) mothers had history of significant maternal illness, history of previous abortion 29 (42.65%), gave the history of previous congenital anomalous babies (10.29%) and also 54.41% of mothers were the history of irregular/absent antenatal checkup.

Conclusion: Congenital anomalies is a priority health problem in newborns. This study has highlighted the prevalence and types of congenital anomalies seen in our locality. Results of the study can be used to predict future incidence of anomalies and to increase public awareness about congenital anomalies to take preventive measures.

Keywords: Major congenital anomalies, Neonate.

Introduction:

Congenital anomalies are also known as birth defects, congenital disorders or congenital malformations. Congenital anomaly is an internal or external structural defect that is identifiable at birth.[1] According to WHO, congenital anomalies are defined as structural or functional anomalies, including metabolic disorders which are present at the time of birth.[2] Congenital anomalies account for 11% of neonatal deaths globally and 9% in India.[3]

Congenital anomalies can be classified as major and minor anomalies. Major defects are structural abnormalities that have cosmetic or medical consequences which may require surgical intervention for correction; examples include cleft palate. Minor anomalies are those with no medical or cosmetic

significance they are more useful for recognition of specific syndromes though isolated anomalies may occur sporadically.[2] A major congenital anomaly affect 2-3% of newborn babies of the approximately 35,000 children born in each year.[4] A study done at AIIMS show that congenital malformations contributed to 13.4% of perinatal deaths as compared to a decade back. Major malformations accounts for 15% neonatal death.[5]

Congenital anomalies are an important cause of neonatal morality both in developed developing countries. It is not only a leading cause of fetal loss, but also contributes significantly to preterm birth, childhood and adult morbidity along with considerable repercussion on the mothers and their families.[6]

Birth defects represent defective morphogenesis during early fetal life. Maternal risk factors contributing to the occurrence of congenital anomalies include genetic and environmental factors and their interaction with each other that may results into malformation, deformation, disruption or dysplasia, that eventually cause congenital anomalies.[2]

Around 40% to 60% of congenital anomalies are of unknown etiology; 20% are attributed to a combination of heredity and other factors; 7.5% due to single gene mutations; 6% is caused by chromosomal abnormalities; and another 5% is due to maternal illness, such as diabetes or infection or use of anticonvulsant or other drugs.[7]

No national survey or hospital based statistics regarding congenital anomalies in Bangladesh is available till date.

This study has been undertaken which will serve as a reference point for actual picture of congenital anomalies in this tertiary hospital and it will also generate data of congenital anomalous among newborns that will help national registry in future. This study was done to know the frequency, pattern of major congenital anomalies and various presentations, which may help to develop strategies for prevention, counseling and management in our setting.

Material and Method:

This is a prospective hospital based observational study. This study was carried out in the Department of Paediatrics of a teaching Dhaka National Medical College & Hospital, Johnson Road, Dhaka, for a period of 3 years from 1st January, 2016 to 31st December, 2018.

The study was conducted in 68 newborns with major congenital anomalies. Newborns either full term or preterm with congenital anomalies were included in the study. All babies delivered at DNMCH including still births comprise the study material.

All neonates were thoroughly examined soon after birth for major and/or minor congenital malformations by an expert pediatrician. All congenital anomalies babies during the study period either detected before birth by ultrasonography of mother or detected at birth were included in this study. Neonatal data regarding gestational maturity, birth weight, sex, anomalies present in the neonate and outcomes were documented. As per the proforma made, relevant information regarding maternal age, parity, maternal disease, maternal drug intake, previous bad obstetric history, consanguinity, and maternal antenatal investigations including antenatal ultrasonography were obtained by reviewing the maternal and labour ward records and by interviewing the parents.

The newborns were examined and assessed systematically for the presence of congenital anomalies. Diagnosis of congenital anomalies was based on clinical evaluation of newborn babies by the pediatrician and other appropriate investigations such as radiography, ultrasonography, echocardiography and chromosomal analysis etc. Anomalies in the study population was classified as per European surveillance of congenital anomalies classification guidelines into major and minor anomalies. The spectrum of anomalies were analyzed in system wise manner. Immediate outcome of the baby, whether the baby was alive as dead, whether the baby needed immediate neonatal support or not was noted. Data was entered into excel data sheet and appropriate statistical analysis was performed.

Results:

Table–I: Shows that the predominant system involved was gastro-intestinal (GI) system (30.87%) followed by central nervous system (23.53%), Musculoskeletal system (16.17%), and urinary system (8.82%).

Cleft lip and cleft palate (14.70%) was the most common anomaly seen in the Gastro-intestinal system and like wise congenital hydrocephalus (14.70%) in central nervous system, craniosynostosis (8.82%) in Musculoskeletal system, Hydronephrosis (5.88%) in urinary system. Multiple congenital anomalies involved 11.76%, congenital Rubella Syndrome (4.41%) and collodian baby (1.47%) in Miscellaneous group.

Table–II: Show that, out of 68 subjects still births accounted for 26.47% and live births accounted for 73.53%. Among the anomalous babies 67.65% were male and 32.35% were female. Of them birth weight ≥ 2.5 kg accounted for 60.29% and weighing <2.5 kg (39.71%) babies were congenitally malformed. Out of 50 alive babies, 35 babies (70%) admitted in the neonatal ward and 15 babies (30%) were not admitted.

Table–III: Shows that different components of the obstetric history were explored. Regarding the parity of the mothers, 28 were primiparas and rest 40 were multiparas. Cases of congenital anomaly were found in 58.82% of multiparas, whereas in primiparas, the proportion was 41.18%. In the present study, 11 (16.18%) mothers had a history of consanguinity, whereas in non-consanguineous couples were 57 (83.82%). Among 68 subjects 10.29% gave the history of having congenital anomalous babies.

In this table also shows that 52.94% were with <37 weeks of gestation and 47.06% were with 37 weeks or more of gestation.

In the present study 23 (33.82%) mothers had a history of significant maternal illness. Among 68 subjects; 29 (42.65%) are the history of having abortion. It has been seen that more than half of the mothers were aged <35 years (76.47%) with only 23.53% of the mothers were over the age of 35 years. About 54.41% were the history of irregular/absent antenatal checkup and regular antenatal checkup were 45.59%.

Table–I: System wise distribution of congenital anomalies (n=68)

Anomalies	No.	%
Central Nervous System	**(23.53%)**	
Congenital Hydrocephalus	10	14.70%
Anencephaly	1	1.47%
Encephalocele	1	1.47%
Meningocele	2	2.94%
Meningomyelocele	2	2.94%
Urinary System	**(8.82%)**	
Hydronephrosis	4	5.88%
Polysystic kidney disease	2	2.94%
Gastrointestinal System	**(30.87%)**	
Gastroschisis	2	2.94%
Omphalocele	3	4.41%
Anorectal anomalies	6	8.82%
Cleft lip and cleft palate	10	14.70%
Musculoskeletal System	**(16.17%)**	
Achondroplasia	2	2.94%
Gross Bony defect (Absence of 1st and 2nd lumber vertebra)	1	1.47%
Craniosynostosis	6	8.82%
Congenital dislocation of hip joint	2	2.94%
Miscellaneous	**(17.64%)**	
Multiple congenital anomalies	8	11.76%
Collodian baby	1	1.47%
Congenital rubella syndrome	3	4.41%

Table – II: Immediate fetal out come

(Association of type of birth, gender, birth weight, admission of the babies with congenital anomalies)

State of the baby	Frequency	Percentage
Still birth	18	26.47%
Live birth	50	73.53%

Sex of the baby	Frequency	Percentage
Male	46	67.65%
Female	22	32.35%

Birth weight	Frequency	Percentage
<2.5 kg	27	39.71%
≥2.5 kg	41	60.29%

Admission in the neonatal ward (In case of live birth)	Frequency	Percentage
Admitted	35	70%
Not admitted	15	30%

Table–III: Obstetric history

(Association between congenital anomalies and maternal and perinatal risk factor)

Obstetric history	Frequency	Percentage
a. Parity		
Primi para	28	41.18%
Multi para	40	58.82%
b. History of Abortion		
None	39	57.35%
Once or more	29	42.65%
c. History of congenitalAbnormal babies		
None	61	89.71%
One or more	7	10.29%
d. Gestational age (week)		
<37 weeks	36	52.94%
≥37 weeks	32	47.06%
e. History of consanguinity		
Present	11	16.18%
Absent	57	83.82%
f. History of Maternal illness		
Present	23	33.82%
Absent	45	66.18%
g. History of Antenatal check-up		
Regular	31	45.59%
Irregular/Absent	37	54.41%
h. History of Maternal age		
<35 years	52	76.47%
>35 years	16	23.53%

Discussion:

Significance of congenital malformation lies not only in their contribution to neonatal and perinatal mortality but, also in causing disabilities and handicaps in infant and children.[1] It is a priority health problem in newborns. During the study period, 68 newborns with major congenital anomalies were included.

In the present study, the predominant system involved in the major anomalia is gastrointestinal (GI) system (30.87%) followed by central nervous system (23.53%), musculoskeletal system (16.17%) and urinary system (8.82%).

In the current study, Cleft lip and Cleft palate (14.70%) was the most common anomaly seen in the gastrointestinal system which is comparable to a study done by Sarkar S et al (6.6%).[6]

Neural tube defects second commonest anomaly in this study (23.53%). Congenital Hydrocephalus was most common Neural tube defect found in this study (14.70%). Other defects being Meningocele, Meningomyelocele, Encephalocele, Anencephaly (Table-I).

Similar results was found by K Fatema et al[7] where she has shown Neural tube defect was the commonest type of anomaly. Among the most frequent Neural tube defect was hydrocephalus. Ensuring folic acid supplementation during preconception period can lower the frequent of these anomalies.[7]

In the present study, multiple defects were present 11.76% of the babies (8 to 68 babies with anomalies). Similar results were found by K Fatema et al[7] where she has shown 11.67% of the neonates with multiple defects. In our study, results is lower that observed by other studies like in India, Mishra and Bhaveja[8] found multiple anomalies are 37.6% of anomalies who had reported 45.2% in his study.

However this could be due to the fact that some of the associations and diseases could not be confirmed because of lack of further workup, early death and logistic reasons.

Cardiac malformation in this study was absent, may be due to under diagnosis because of lack of availability of sophisticated diagnostic technique and neonatal follow up.

In this study, low birth weight (LBW) (<2.5kg) associated with risk of congenital malformtions. This highlights the fact that the presence of congenital anomaly itself hampers the growth of a developing foetus.[10]

Present study has documented higher incidence of malformation in male babies (67.65%) than female babies (32.35%). Other studies like K Fatema et al[7] and Aman T et al[9] has also documented similar results.

In our study, out of 68 subjects, still births accounted for 26.47% and live births accounted for 73.53%. In other study like K Fatema et al[7] found higher percentage of congenital malformation in still birth. Usually major malformations are incompatible with life, this may the reason of high incidence of congenital malformation in still born babies.[10] In this study, lower percentage of still births than live births could be due to total babies born in the department of Obstetrics and Gynaecology of DNMCH during the study period.

Previous studies have reported significantly higher incidence of congenital malformation among the multiparas.[10] Our results is consistent with this finding (58.82%).

Consanguineous marriages (when parents are related by blood) are reported to play a major role in the occurance of congenital malformations.[11] In the present study, non-consanguineous couples were more (83.82%). In our study, majority of mothers with congenital anomalous fetuses belong to gestational age <37 weeks (52.94%) as seen in other study.[7]

Regular antenatal check up may help early diagnosis and termination of fetuses incompatible with life. Present study has reported antenatal visit in majority of mothers were irregular or absent (54.41%).

Maternal age is also a risk factor for abnormal intrauterine fetal development. Advanced maternal age increases the risk of chromosomal abnormalities, including Down's Syndrome.[1] Swain's study has also documented highest incidence of malformations in babies of mothers more than 35 years of age.[12]

But in our study, more than half of mothers were aged <35 years (76.47%) with only 23.53% of the mothers were over the age of 35 years. Prenatal risk factors associated with occurrence of anomalies is also well established from previous studies.

Risk factors that are identified included Maternal illness e.g. maternal infections such as syphilis and rubella etc, maternal anaemia, malnutrition, maternal diabetes, maternal poor socioeconomic status, maternal exposure to certain pesticides and other chemicals, as well as certain medications, alcohol, tobacoo and radiation during pregnancy, may increase the risk having a fetus or neonate affected by congenital anomalies.[12] In this study, 23 (33.82%) Mothers with congenital anomalies babies had history of significant maternal illness such as diabetes, fever, anaemia, rubella,

UTI, malnutrition, hypertension etc. This is because could not give proper history and irregular/absent antenatal visit.

Congenital anomaly contributed a significant proportion of infant mortality and morbidity as well as fetal mortality.

This study is encountered only among newborn with major congenital anomalies. From this study some clue may derived regarding the frequency and distribution pattern of major congenital malformation among Bangladeshi populations. Despite the high risk of recurrence of congenital malformations, there are no well accepted preventive measures in developing countries. It indicates that strong preventive measures for congenital anomalies in this region are needed.

Conclusion:

Congenital anomalies are an important causes of infant and childhood deaths, chronic illness and disability as well as fetal mortality. Mortality of infants born with congenital anomalies varies with the type of anomaly. This study has highlighted types of major congenital anomalies seen in our locality. To draw significant conclusions it is recommended that all neonates should be examined with scruting for overt as well as occult congenital anomalies.

Regular antenatal visits and prenatal diagnosis are recommended for prevention, early intervention, even planned termination, when needed will reduced perinatal morbidity and mortality. Antenatal diagnosis, genetic counselling, better diagnostic and management facilities should be provided to improve the outcome.

References:

1. Prajapati JV, Kacha RA et al. Study of congenital malformation in Neonates born at tertiary care hospital. National Journal of Community Medicine, March 2015; 6: 30 – 34.
2. UNICEF. Neonatal Health. Available at Unicef.in/whatwedo/2/Neonatal-Health.
3. Tenali AS, Kamalakannan SK, Jayaraman KK. Spectrum of Congenital anomalies of neonates in a tertiary care hospital in southern India. International Journal of Contemporary Pediatrics, March-April 2018; 5 (2): 1 – 6.
4. Unimon K Devassy, Danasegaran M, Kumar SS et al. Congenital anomalies among Newborns. Bali Medical Journal (BaliMJ) 2015; 4 (1): 21 – 23.
5. Thaddanee R, Patel HS, Thakor N. A study on incidence of congenital anomalies in newborns and their association with maternal factor: a prospective study. International Journal of contemporary pediatrics, April – June 2016; 3 (2): 579 – 582.
6. Sarkar S, Patra C, Dasgupta MK, Nayek K and Karmakar PR. Prevalence of congenital anomalies in Neonates and associated risk factors in a tertiary care hospital in Eastern India.
7. K Fatema, F Begum, N Akter, SMM Zaman. Major Congenital Malformation among the Newborns in BSMMU Hospital. Bangladesh Medical Journal 2011; 40 (1): 7 – 12.
8. Mishra PC, Baveza R. Congenital malformation in newborns: A prospective study. Indian Pediatr 1989; 26: 32 – 5.
9. Aman Taskade, Krishna, Vilhelar et al. Congenital malformation at birth in central India. A rural medical college based data. Indian Journal of human genetics, September 2010; 16: 159 – 163.
10. Mohanty C, Mishra OP, Das BK, Bhatia BD, Singh G. Congenital malformations in newborn: A study of 10,874 consecutive births. J Anat Soc Indian 1989; 38: 101 – 11.
11. Hudging L, Cassidy SB. Congenital anomalies. In: Martin RJ, Fanaroff AA, Walsh MC, editors. Neonatal Perinatal Medicine. 8th ed. Philadelphia: Mosby – Elsevier, 2006; PP 561 – 81.
12. Swain S, Agrawal A, Bhatia BD. Congenital malformation at birth. Indian Pediatrics 1994; 31: 1187 – 1191.