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Incidence of neural tube defects and other external congenital anomalies among newborn

Abstract

Background: Congenital malformations are becoming an important cause of neonatal death due to reduction of mortality by other common causes of death owing to the improvement of neonatal care. Routine physical examination of every newborn is important for multiple purposes including exploration of external congenital anomalies.

Objective: To determine the incidence of Neural Tube Defects (NTDs) and other external congenital anomalies that can be detected by routine physical examination only.

Methods: It was an observational cross sectional study. This study was conducted in the department of Pediatrics and Obstetrics of Khulna Medical College Hospital from June 2016 to May 2017. All the delivered newborn in Obstetrics ward and the neonates in the pediatrics ward were included in the study. The babies were examined immediately after delivery or admission. A detailed history including potential risk factors was taken and a thorough physical examination was performed. All the relevant information was noted in a predesigned questionnaire.

Results: A total of 4,462 neonates were studied in pediatrics and obstetrics ward. Thirty four (0.76%) cases of congenital anomalies were found during one year. Neural tube defects were observed in 4 (0.09%) cases only. No other risk factor except folic acid intake was related with them. Congenital anomalies most frequently involved the musculoskeletal system (29.4%).

Conclusion: Routine physical examination of newborn can identify a significant number of congenital anomalies. Non-intake of folic acid during preconceptional and antenatal period is the single-most risk factor for NTDs.

Keywords: Neural tube defects, Congenital anomalies, Neonate

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Introduction

One of the important purposes of the routine examination of the newborn is to detect any congenital anomalies that may impair normal development of the baby.[1] There are many internal anomalies like renal, cardiac, metabolic and GIT anomalies. The major constraint to detect such internal anomalies is lack of diagnostic facilities and expert manpower. The prevalence of congenital anomalies is underestimated in the developing world due to lack of reliability of medical records and health statistics.[2] A few reported estimates of the prevalence at birth of congenital anomalies came from registries of congenital malformations in newborns which show that the prevalence of recognizable malformations among newborn is between 2-3% that is similar to that found in the industrialized world.[3]

Congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India.[4] Patients with multiple congenital anomalies present a relatively infrequent but tremendously difficult challenge to the pediatrician. Among all the congenital anomalies most important preventable anomalies is the neural tube defects (NTDs). Some trial suggested that perhaps all of spina bifida and anencephaly can be prevented with folic acid. Even though it is estimated that only 10% of the folic acid preventable spina bifida and anencephaly is actually being prevented.[5] The proportion of perinatal deaths due to congenital malformations is increasing as a result of reduction of mortality due to other causes owing to the improvement in perinatal and neonatal care. In the coming decades, this is going to be a leading cause of morbidity and mortality in centers providing good neonatal care.[6,7]

Aim of this study was to determine the incidence of neural tube defects and other external congenital anomalies in the newborn presented to a tertiary care hospital and also to explore the implication of potential risk factors related to it.

Materials and methods

This observational and descriptive study was carried out in the department of Pediatrics and Obstetrics in Khulna Medical College hospital, Khulna. The study period extended from June 2016 to May 2017. All the babies born in the Obstetrics ward were examined thoroughly within 24 hours to find out any external anomalies. Babies admitted in the newborn unit of pediatric department were also examined who did not come from Obstetric ward of this institute. All the babies with congenital anomalies were enrolled for detail study. Antenatal history for any risk factor was taken into account.

Two doctors were inducted to collect the data (one each for Pediatrics and Obstetrics ward) and to note it into the data sheet which were scrutinized by a senior pediatrician at a weekly interval. The questionnaire was prepared to note the general information including the risk factors of congenital anomalies like maternal gestational Diabetes Mellitus (GDM), hypertension, antenatal and preconceptional ingestion of folic acid, consanguinity, drug ingestion etc. The babies were followed up till discharge or death.

Results

A total of 4,462 babies were examined for congenital anomalies

during the period of one year. The babies examined in pediatric department were 1,359 and that in Obstetric department were 3,103 (Table 1). Total number of congenital anomalies was 34 (0.76%). Among the 34 cases, 25 (74%) were male and 9 (26%) were female. Male- Female ratio was 2.8:1.

In this study NTDs were found in 4 (0.09%) cases. Among the 4 neural tube defects, 3 (75%) were male. Maternal age and other risk factor association are described in Table 2. Non-intake of folic acid during perinatal and antenatal period was observed in all 4 cases of NTDs.

Distribution of anomalies according to system involvement is presented in Table 3. Anomalies related to musculoskeletal system was most frequent (10; 29.4%). Club foot was the highest in frequency among the musculoskeletal system (80%). Next most commonly involved system was alimentary system (23.5%). No contributory factor was looked after for congental anomalies other than NTDs.

Table 1 Incidence of Congenital Anomaly

Ward	Total number of Baby	Number of Cong. anomaly	% of Cong. anomaly	NTD No.	% of NTD
Pediatrics	1359	26	1.91	2	0.15
Obstetrics	3103	08	0.26	2	0.06
Total	4462	34	0.76	4	0.09

Table 2 Personal profile & risk factors related to NTD patients

Sl no	Sex	Maternal age (Yrs)	Maternal illness	Consanguinity	Antenatal Folic acid	H/O NTD	Type of NTD
1	M	19	Nil	Nil	Nil	Nil	Meningocele
2	M	28	Nil	Nil	Nil	Nil	Anencephaly
3	F	30	Nil	Nil	Nil	Nil	Anencephaly
4	M	26	Nil	Nil	Nil	Nil	Anencephaly

Table 3 Congenital anomalies related to system involved

System	Types of anomaly	No. of patient	% of Total
Nervous system	Anencephaly	3	
	Hydrocephalus	2	
	Meningocele	1	
	Total	6	17.7
Musculoskeletal System	Club foot	8	
	Syndactyly-Hand	1	
	Flexon contracture of hand	1	
	Total	10	29.4
Alimentary System	Cleft lip +/- Cleft palate	5	
	Omphalocele	2	
	Inguinal hernia	1	
	Total	8	23.5
Ear and Eye	Microtia ± Anotia	2	
	Microphthalmia	2	
	Exophthalmia	2	
	Hypertelorism	1	
	Total	7	20.6
Others	Webbed neck, Low set ear, Dextrocardia	1	
	Webbed neck, Wide spread nipple	1	
	Rudimentary external genitalia	1	
	Total	3	8.8
Grand Total		34	100

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Discussion

This study was an attempt to find out the external congenital anomalies which were revealed by physical examination only. Our findings were close to that of Singh et al from India who reported a frequency of 1.5% and Golalipour et al from Iran (1.01%).[8,9] The low rate of our study was due to exclusion of still born which was included in other studies. Cardiac murmur was also excluded in our study because it needs further investigation for confirmation. Frequency of congenital anomalies were more in male in our study which is found in other studies in a variable degree.[10-12]

The incidence of NTD in this study was 0.09% (1 in 2,500 live birth) with male predominance. The worldwide incidence of NTDs is estimated to be around 1 per 1,000 livebirths.[13] However, there are remarkable variations in the incidence of NTDs and other CNS defects. In certain regions of China, the incidence of NTD is 1 in 100 live births, being the highest, whereas in the Scandinavian countries, it is only 1 in 5,000 livebirths.[14,15] In the United States, the incidence of NTDs was estimated as 1 per 1,000 deliveries, anencephaly as 0.6-0.8 per 1,000 live births, and open spina bifida as 0.5-0.8 per 1,000 livebirths.[16] Among the 4 cases of NTDs in present study, none of the mother had taken folic acid during preconceptional and antenatal period. However causal relationship could not be established as it has not been compared with the control group.

Among the systems involved, musculoskeletal system had highest incidence (29%) followed by alimentary system (23%) which is comparable with other studies. One study from India and another from Iran found musculoskeletal anomalies as highest in order (30.60% and 30.10% respectively).[17,18] Asindi et al. from India and Ekewre et al. from Nigeria found alimentary system, nervous system and cardiovascular system as the most commonly affected parts in descending order of frequency in their series.[19,20] There were variations in the frequency of organ system involvement. It depends partly due to inclusion criteria and modality of investigation applied.

This study is limited by few factors. Presumably, a small number of cases has been missed in obstetrics ward due to delivery at late night and also junior doctors might have overlooked some positive findings. The implication of risk factors could not be ascertained as the factors were not noted for all the babies except the NTD cases.

Conclusion

Further large scale multicenter study is needed to quantify the national incidence of congenital anomalies with particular emphasis to folic acid-preventable NTDs. Estimation of national incidence would be immensely helpful in adopting the preventive measures for childhood mortality and morbidity.

Acknowledgments

None.

Conflicts of interest

None.

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