Original article

Pattern and risk factors of birth defects among the newborns: A hospital based study

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Abstract :

Introduction : Birth defects are one of the important causes of perinatal mortality and morbidity.

Methods : A cross sectional descriptive study was carried out in In-patient Department of Obstetrics and Gynaecology in Dhaka National Medical College Hospital between January 2008 and December 2008.

Objectives : The objectives of the study was to determine the frequency of identifiable birth defects, to observe the types of birth defects found among the newborns and to estimate the risk factors present among the parents of birth defect babies.

Results : During this study period 2990 deliveries took place. The frequency of birth defect babies was found 2% (n=60). Seventy seven percent of the babies were born at term and 23% of them were born prematurely. Babies were born live and still birth 88.33% and 11.67% respectively. Birth defects were predominant in male (55%) than female (45%) babies. Among the mothers 17%, 32% 38% and 13% were of age <20, (20-25), (26-30) and (31-40) years respectively. Educational status revealed 88% of the mothers and 83% fathers were literate. Thirteen percent of the population came from poor socioeconomic condition. Others were from middle class (54%) and affluent (33%) society. No risk factors were identified in 57% of the population. Diabetes or GDM (15%), no intake of Folic acid and Zinc (13%), maternal age >35 years (7%), maternal infection (3%), history of previous birth defects babies (3%), Consanguineous marriage (2%) was found as risk factors. Most common system involved was musculoskeletal system (35%), followed by genitourinary system (24%), central nervous system (19%), and face (15%). Birth defect was found in cardio-vascular and gastrointestinal system was 3% and 3% respectively. Functional defect (Down's syndrome) was found as 2%.

Key word: Birth defect, Pattern, System involved, Risk factors

Introduction :

Birth defect is a widely used term for a congenital malformation, i.e. a congenital, physical anomaly which is recognizable at birth. According to CDC, most birth defects are believed to be caused by a complex combination of factors including genetics, environment, and behaviours. Though many birth defects have no known cause.[1] Birth defect began to emerge as one of the major childhood health problems.[2] It is a dominant cause of infant morbidity and mortality.[3] It has been seen that 12.3-32% of neonatal deaths were considered to be secondary to congenital anomalies [4].

Incidence of birth defect varies from country to country. It was reported to be as low as 1.07% in Japan and as high as 4.3% in Taiwan [5] whereas the birth prevalence of anomalies was 2%in England, 1.49% in South Africa [6] and 3.65% in India.[7]The reason for the regional difference of congenital anomalies might be attributed to the many factors, such as: maternal risk factors, environmental exposures, ecological, economical, ethnic and other factors.[8,9]. Risks factors like infectious agents, chemical compounds, radiation, use of medication, maternal metabolic diseases, multiple births, prematurity, occupational exposure are associated with higher congenital disorder.[10,11] Furthermore, low schooling and low socioeconomic status in the population are other factors which are highly relevant.[12] An environmental exposure can have a preconceptional mutagenic action or a post-conceptional

teratogenic action.[13]Deficiency of folic acid and other nutrients such as zinc and vitamin B1 in the periconceptional period are established risk factor for neural tube defects.[14,15] Birth defects are increasing in pregnancies at age more than 35 years and abnormal intrauterine environment.[10]

Consanguineous marriages regarded as important factor contributing to increased congenital malformations, recessive gene may thus come to light for the first time in an in bred descendant after have been hidden for generations. [5] Routine screening for fetal abnormalities is very successful, there are limitations to the abilities of both the technique and the operators to detect every anomaly.[16] Whereas, treatment and rehabilitation of these morbid children is difficult, and sometimes recovery is impossible. [17]

Materials and Methods :

This was a cross sectional, descriptive study carried out in the Department of Obstetrics and Gynaecology at Dhaka National Medical College between January 2008 and December 2008. The objectives of the study was to determine the frequency of birth defects, to observe the types of birth defects found among the newborns and to estimate the risk factors present among the parents of birth defect babies.

No sampling technique was applied in this study. All women admitted for delivery in this department was recruited in this study purposively. Data was collected from the mothers who delivered birth defect babies by face to face interview regarding socio-demographic factors, duration of pregnancy, maternal illness like diabetes, hypertension, hypothyroidism, maternal infection (with rubella, toxoplasmosis, cytomegalo virus, herpes simplex), smoking habit, antenatal check up, drug intake, supplementation of folic acid and zinc. Laboratory investigations for blood sugar, VDRL, thyroid function test, antibody of TORCH infection and ultrasonography for the detection or confirmation of congenital anomaly of the foetus was done. Thorough examination of the babies after birth was performed by the investigators. Follow up of the babies were done till discharge from hospital or death. All data was screened and analysed.

Results :

During this study period 2990 deliveries took place. The frequency of birth defect babies was found 2% (n=60). Total birth defect babies were 68, as five babies had multiple birth defects. Seventy seven percent of the babies were born at term and 23% were born prematurely. Babies were born live and still birth 88.33% and 11.67% respectively. Birth defects were predominant in male (55%) than female (45%) babies. Among the mothers 17%, 32% 38% and 13% were of age <20, (20-25), (26-30) and (30-40) years respectively. Fathers were of

age (22-25), (26-30), (31-40) and (40 & above) in 13%, 30%, 40% and 17% cases. Educational status revealed 88% of the mothers and 83% fathers were literate. Thirteen percent of the population came from poor socioeconomic condition. Others were from middle class (54%) and affluent (33%) society. Among the mothers of birth defect babies no risk factors were identified in 57% of the population. Diabetes or GDM (15%), no intake of Folic acid and Zinc (13%), maternal age >30years (7%), maternal infection (3%), history of previous birth defects babies (3%), Consanguineous marriage (2%). The most commonly involved system of the birth defects was musculoskeletal system (35%), followed by genitourinary system (24%), central nervous system (19%), and face (15%). Birth defect was found in cardio-vascular and gastrointestinal system was 3% and 3% respectively. Functional defect (Down's syndrome) was found as 2%.

Table I: Socio-demographic characteristics of the mothers of birth defect babies:

Maternal age (in years)	N	%
<20	10	17
20-25	19	32
26-30	23	38
≥31	8	13
	60	100
Maternal education		
(level of education)		
Illiterate	7	12
Upto primary	8	13
VI-secondary	30	50
HSC and above	15	25
	60	100
Monthly family income (in BDT)		
Within 5000	8	13
>5000-10,000	16	27
>10,000-20,000	16	27
>20,000	20	33
	60	100

Table II: Characteristics of the birth defect babies

Gestational age (in weeks)	N	%
<37 weeks	14	23
37 -41 weeks	46	77
	60	100
Sex		
Male	27	45
Female	33	55
	60	100
Condition at birth		
Live	53	88.33
Still birth	7	11.67
	60	100

Table III: Distribution of birth defects babies according to anatomical system

System	Type of birth defects	frequency	Percentage
Musculoskeletal (n=24)	Club foot	8	11.76
	Polydactyly in hand	4	5.88
	Polydactyly in foot	1	1.47
	Syndactyly	3	4.41
	Cong. Amputation of great toe	1	1.47
	Absence of fingers	1	1.47
	Absence of distal two phalanges in 4 fingers	1	1.47
	Sirenomelia	1	1.47
	Conjoint twin	1	1.47
	Achandroplasia	1	1.47
Genitourinay (n=16)	Undescended testes	3	4.41
	Hypospadiasis	3	4.41
	Imperforated hymen	2	2.94
	Hydronephrosis	5	7.35
	Polycystic kidney	3	4.41
CNS (n=13)	Anencephaly	2	2.94
	Spinabifida	3	4.41
	Hydrocephalous	3	4.41
	Encephalocoele	1	1.47
	Meningicoele	3	4.41
	Sacrococcygeal terratoma	1	1.47
Face (n=10)	Cleft lip	8	11.76
	Cleft palate	2	2.94
CVS (n=2)	Septal defect	2	2.94
GIT (n=2)	Omphalocoele	1	1.47
	Gastrochiasis	1	1.47
Functional (n=1)	Down's syndrome	1	1.47
Total		68	100

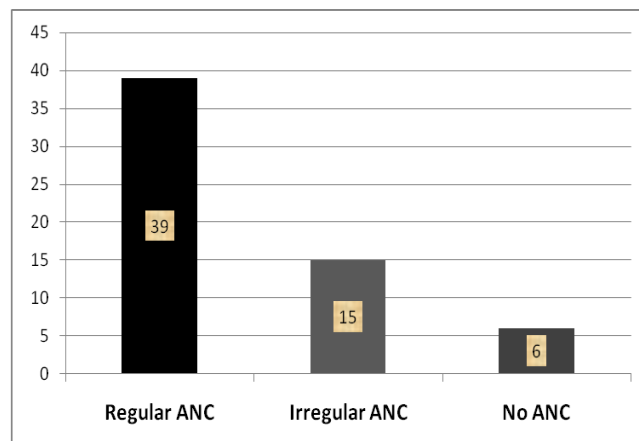**Figure 1: Distribution of mothers according to Antenatal check up:**

Figure 2: Distribution of mothers according to risk factors
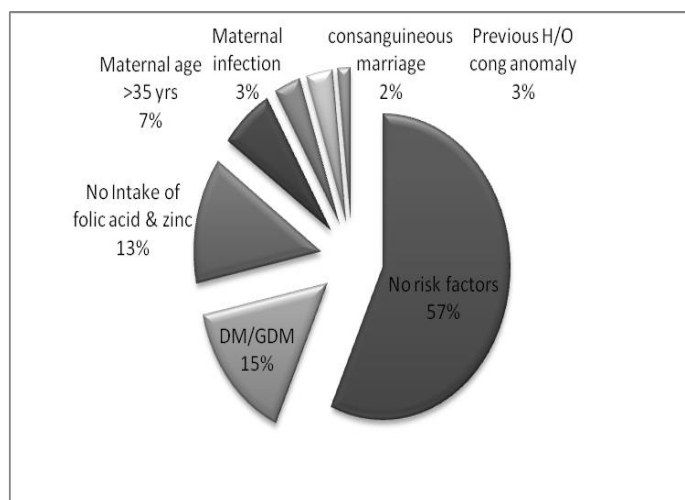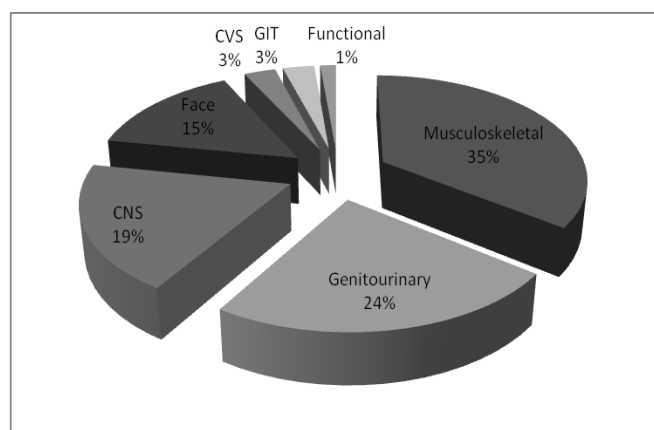

Figure 3: Distribution of birth defects according to System involved

Discussion :

The total occurrence of birth defect babies in this study was found 60 (2%) in a total of 2990 babies born in the department of Obstetrics and Gynaecology of DNMCH. This is in conformity with several studies.[18-23] On the other hand, other authors found higher frequency (3% or more) of birth defects in their studies.[24-26] Actually the prevalence of birth defect varies from country to country depending on racial, economic, ecological and ethnic factors.[8,9]

In this study the percentage of abnormalities increased significantly at age (25-30) years (38%) than at age of above 30 years (13%). Similarly another author found significant percentage (34.98%) of birth defect babies at younger maternal age (20- 24) years.[27] Though we know that incidence of congenital anomaly is more with advanced age of mother due to aged chromosome.

Current study found 50% and 25% of the population got secondary and higher secondary education respectively. Moreover, the population was economically solvent except 13% (poor). These two factors reflect the lower frequency (2%) of birth defects in this study. According to Sylvana G and coworker schooling and socioeconomic status in the population are important factors which are highly relevant.[12]

This study found 65% of the population received antenatal check up regularly. Study also found those who had major congenital anomaly sought no antenatal check up and were not screened for anomaly. Another author found only 8% mothers received regular antenatal check up and got frequency of birth defect is much higher (3.68%)[28] than ours (2%).

Regular antenatal check up may help early diagnosis and termination of fetuses incompatible with life thereby reduces the number of congenital anomaly baby born.

Current study found congenital anomaly more in female babies (55%). But several authors of other studies found male predominance in birth defects in their studies.[25,28,29] In a study Khanum et al found females as the predominant group.[30]

In this study birth defect observed single system involvement constituted 91.67% of the cases compared to 8.33% of the multiple system involvement which is almost similar to the findings of another study carried out at BSMMU. They found single system involvement in 88.33% cases and multiple system involvement in 11.67%.[28] The study of K Chowdhury and co-workers got a bit higher percentage (15.15%) of babies with multiple system involvement.[29]

More than one system anomaly is usual, but in our study along with other authors multisystem involvement was found less in percentage. In depth investigation might reveal the defects of other system which was not apparent.

Among the system distribution of birth defects musculoskeletal system (35%) was the most commonly involved system in this study. Genitourinary system (24%) was involved next to musculoskeletal system. Other authors also found musculoskeletal system as the most commonly involved system.[25,29, 30] Other systems involved in our study were central nervous system (19%), face (15%), cardio-vascular (3%) and gastrointestinal (3%) system. K Chowdhury found defect in gastrointestinal and genitourinary system as second and third in order.[29] Some Authors found predominant involvement in central nervous system [18,19,24] and cardiovascular system [23]. Actually defects of musculoskeletal system are the easiest to detect by physical examination. The low prevalence of cardiovascular defects may be due to the deficiency of in depth investigation. Moreover many cardiovascular defects are not obvious during the first 48 hours of birth and by this time many babies left hospital without detection of cardiac defect.

Risk factor of congenital anomaly was not found in 48% cases as detailed investigation was not possible to carry out in our study. Even though in 57 % cases risk factors were found. Gestational Diabetes Mellitus or Diabetes Mellitus was found in 15% mother. This study found risk factors like, no intake of folic acid & zinc (13%), increased maternal age (7%), maternal infection with cytomegalo virus and rubella virus (3%), history of previous birth defects babies (3%) and consanguineous marriage (2%). Different studies showed infrequent ante natal check up,[28] deficiency of folic acid and zinc,[14,15] consanguineous marriage,[5] maternal chronic illness, gestational illness, maternal infection with TORCH are strongly relevant with of birth defects.

Conclusion :

This was a hospital based study. To get a real picture of birth defects a country wise research is needed. Birth defect is a major childhood health problem. Treatment and rehabilitation of the morbid birth defect babies is difficult and recovery in many cases is impossible. Therefore, prevention programs can be approached if the etiology of birth defect is known. Programme should be extended by Government, NGO and private organisations towards pre-conceptional folic acid, folic acid and zinc supplementation during the first trimester of pregnancy, Rubella vaccination to each girl. Pre-conceptional control of diabetes mellitus and genetic counseling and anomaly scanning is need for at risk mothers. Awareness to be build against teratogenic drugs, smoking and alcohol intake

during pregnancy. Consanguineous marriage should be avoided.

Women should complete their reproductive lives within 30 years of age. On the other hand, termination of pregnancy in lethal and severe birth defects should perform. Early detection followed by effective treatment of deformities is necessary.

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