ORIGINAL ARTICLE

Frequency of Birth Defects and Associated Risk Factors

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ABSTRACT

Aim: To determine frequency of congenital anomalies in new born & associated maternal risk factors.
Methods: The cross sectional observational study was conducted at Department of Obstetrics and Gynecology Unit A of Khyber Teaching Hospital Peshawar from 1st June 2007 to 30th June 2009. All the women giving birth to babies with congenital anomalies during this period were included. Demographic details, associated risk factors and the type of congenital anomalies in babies were recorded. Diagnosis of congenital anomalies was based on u/S and clinical evaluation of new born.
Results: Maternal age ranged from 17-39 years and mean age was 24.63±5.97 years. 26.72% were nullipara and 21.55% mother were grand multipara with mean parity was 3.8. Major risk factor for congenital anomalies was consanguineous marriage (46.5%) which was marked high. Other frequent factors were previous h/o of miscarriages in 17% and past h/o congenital anomalies in 10.34% cases.
Conclusion: Neural tube defects were the most prevalent anomaly detected and early prenatal diagnosis is helpful in decreasing the indirect prevalence of perinatal mortality by offering early termination. The commonest associated factor was consanguineous marriage the frequency of which may be reduced by creating awareness regarding the avoidance of consanguineous marriages.
Keywords: Birth defects, Risk factors, Neural tube defect

INTRODUCTION

The birth defects also termed as congenital anomalies remains one of the leading causes of morbidity and mortality of neonates and infants in developing countries like ours[1,2]. In our country it accounts for about 6-9% of perinatal deaths[3,4]. Birth defects are defined as the macroscopic or microscopic abnormalities of body structure which are diagnosed in either antenatal or postnatal period. These anomalies can occur as single or multiple defects.[5] Isolated congenital anomaly is the structural defect, which can be detected as one localized defect in the morphogenesis, while multiple birth defects results from two or more different morphogenic abnormalities that occur during the emberyogenesis[6].

Evaluation before conception and counselling is recommend as preventive measures even before the pregnancy begins like for example daily intake of folic acid before conception reduces the rate of malformations[7]. About 65–75% cases of congenital malformations result from multiple causes not just a single factor is involved.[8] Single or multiple gene defects[2], hereditary predisposition[3] and diabetes mellitus[9-11] are commonly responsible for birth defects.

Early diagnoses of birth defects through frequent ultrasonography, maternal serum markers and techniques like chorionic villous sampling during pregnancy and early termination of pregnancy has reduced the incidence of congenital anomalies and Its associated morbidity and mortality[12] in developing countries

Ultrasonography can detect 70-80% of the birth defects if performed during early pregnancy.[13] Developing countries like ours are lacking these facilities at the primary health care level as a result more pregnant patients with congenitally malformed fetuses presents in the last trimester of pregnancy to tertiary health centres like Khyber Teaching Hospital Peshawar so the need was felt to conduct this study.

PATIENTS AND METHODS

This cross-sectional descriptive study was carried out in Obstetrics and Gynaecology Unit A, Khyber Teaching Hospital from 1st June 2007 till 30th June 2009 for duration of 2 years. The data were collected from the antenatal clinic of the Gynaecology and Obstetrics, OPD and the labour room both booked as well as non-booked patients for the study. Patients once identified antenatally or postnatally were admitted for further workup, which included a detailed history and examination. History was specifically directed towards finding out of known risk factors associated with congenital anomalies in newborn viz history of diabetes mellitus, infection during pregnancy, exposure to medicines, radiation exposure, cousin marriage, past history of abortion or congenital anomalies and any family history of diabetes mellitus or congenital anomalies. Routine

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investigations such as blood grouping, full blood count, urine routine examination and random blood sugar were carried out in all the patients along with obstetrical ultrasound. The data were collected on a predesigned proforma with all the investigations and findings. Data analysis was done by SPSS version 16.

RESULTS

During the study period of 2 years 6297 deliveries occurred and 116 neonates had congenital anomalies giving a prevalence of 18.4/1000 births or 1.8%. Among those, 80% patients were non booked (less than 3 visits in outpatient department). Maternal age ranged from 17-39 years and mean age was 24.63±5.97 years. 60 women (51.72%) were between 21-30 years of age and name of them were above 40 years. 31(26.72%) were nullipara and (21.55%) mother were grand multipara. Mean parity was 3.8 (Table 1).

Table 1: Type of congenital anomalies (n=116)

Anomalies	No.	%
Central Nervous System	**58**	**50.0**
Hydrocepalus	27	23.3
Anencephaly	18	15.5
Encephalocele	-	-
Meningocele	7	6.0
Meningomyelocele	3	2.6
Spina bifida	8	6.8
Microcephaly	2	1.7
Musculoskeletal System	14	12.1
Talipes equinovaurs	8	6.8
Syndactyly	2	1.7
Polyactyly	2	1.7
Achondroplasia	-	-
Osteogenesis imperfect	-	-
Congenital dislocation of hip joint	-	
Arthrogyposis multiplex	2	1.7
Facial Deformity	**4**	**3.4**
Gestational System	**9**	**7.7**
Fetal ascutes	3	2.6
Omphalocoele	6	5.2
Oesophageal atresia	-	-
Duodenal atresia	-	-
Miscellaneous	**9**	**7.7**

The comment congenital anomalies were neural tube defects seen in 58 patients (50%) amongst than hydrocephalus (27) was predominant followed by anen-cephalous (18 cases), while other neural tubo defects were lesser common including 8 cases of spina-bifida, 7 cases of nemegocele 3 cases of meningo mydocele and 2 cases of microcephalic 31 babies (18%) had multiple congenital anomalies all associated with neural tubo defect. Second comments system affected was of musculoskeletal system including 14 cases (12%) and talepo equinovarus was the most frequent differently with 8 cases, among them in 7 cases talips equinovarus was associated. Other defects include hydrops fatalies in 9 cases, facial deformities 4 cases, gastrointestinal (9 cases). Table 2 depicts relevant risk factors associated with congenital anomalies of fetus. Major risk factor for congenital anomalies was consanguineous marriage (46.5%) which was marked high. Other frequent factors were previous history of miscarriages in 17% cases and past history of congenital anomalies in 10.34% cases.

Table 2: Associated risk factors for congenital anomalies (n=116)

Risk factors	No.	%
Past H/o abortion	25	21.5
Past H/o S.B./NND	7	6.0
Past H/o congenitally abnormal babies	12	15.8
H/o fever / infection	5	6.6
H/o drug intake in pregnancy	2	5.3
H/o diabetes mellitus	8	6.8
H/o smoking	-	-
H/o irradiation in pregnancy	1	0.8
H/o consanguineous marriage	-	-
Family H/o congenitally abnormal babies	4	3.4
Family H/o diabetes	4	3.4

DISCUSSION

The frequency of birth defects in our study was 18.4/1000 deliveries or 1.8% which is in correspondence with other studies[14,15], whereas this figure was reported as 87 per 1000[16] United Arab Emeritus while in a study carried out in Aga Khan University Medical Centre in Karachi the frequency was 28/1000 birth.[17] This difference in results may be due to different characteristics of the study population, availability of diagnostic facilities and the duration of postnatal follow up.

In our study the maternal age was commonly in the range of 21 to 30 years and none was above 40 years which is in contrast to other studies where 32% of mothers were aged 35 years and above younger age group may be the reason for not detecting any case of Down syndrome in this study[14].

The only significant risk factor in this study was prevalence of consanguineous marriage (46.5%). This is in comparison to other reported studies of 44%[15] but it is in contrast to a study from Agha Khan University hospital, which claimed only 18.2% cases associated with consanguinity which does not correlate with other studies.[17] This is a preventable and definite associated risk factor which can be resolved by creating awareness and providing marital and genetic counselling.

About 6(6.89%) mothers were diabetic almost similar to the results of others studies (5.26%)[18] incidence of birth defects are more among diabetic mothers as compared to general population, which can be reduced by strict glycemic control around the time of conception and during the period of emberyogenesis. It is also reported that pre-gestational diabetes mellitus is also a significant risk factor[19].

As in other studies, neural tube defect was the commonest anomaly found in this study (50%), which is comparable to a study from Karachi showing 63%[20]. Neural tube defect was also reported as most common birth defects from India as 4-15/1000 live births and in United States as 1/2000 births[21]. Ensuring maternal folic acid supplementation during peri-conceptional period can lower the incidence of these anomalies[15,22].

Different skeletal system abnormalities either in isolation or in association with other anomalies were found in 14(12.06%) cases which is comparable to other studies[23]. Again genetic counselling and early antenatal detection would help in lowering the incidence in newborns by earlier pregnancy termination the before the period of viability. A contrast of heart defects was found which may be due to under diagnosis because of lack of availability of sophisticated diagnostic tools and loss of neonates to follow. Increased rate of detection of cardiac anomalies is possible by incorporating four chamber views of the heart. Thus by creating awareness among the studied population regarding the avoidance of consanguineous marriage, peri-conceptional use of folic acid supplementation, screening of high risk cases and early prenatal detection by routine use of ultrasonography around 16-20 weeks and offering termination of pregnancy in cases of lethal anomalies the perinatal morbidity and mortality.

CONCLUSION

NTD were the most prevalent anomaly detected and early prenatal diagnosis is helpful is decreasing the indirect prevalence of perinatal mortality by offering early termination. The commonest associated factor was consanguineous marriage the frequency of which may be reduced by creating awareness regarding the avoidance of consanguineous marriages.

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