FREQUENCY AND PATTERN OF DISTRIBUTION OF ANTENATALLY DIAGNOSED CONGENITAL ANOMALIES AND THE ASSOCIATED RISK FACTORS

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ABSTRACT

Objective: To find the frequency and pattern of distribution of antenatally diagnosed congenital anomalies and the associated risk factors.

Methodology: This descriptive study was carried out at the department of Obstetrics and Gynecology, Lady Reading Hospital Peshawar from March 2012 to April 2013. Women with ultrasound report of congenitally abnormal fetus irrespective of the gestational age were included. They were evaluated for the presence of risk factors including periconceptional use of folic acid, maternal medical disorders e.g diabetes, epilepsy and history of smoking, maternal and paternal ages, consanguineous marriages and family history of anomalies.

Results: A total of 62 women were included in the study. Fifty seven (91.9%) were having isolated anomalies while 5 (8.1%) presented with complex anomalies. Central nervous system (CNS) was the most commonly involved system (79%). Lack of folic acid use and consanguineous marriages were two most important risk factors; however their correlation with congenital anomalies was not significant. Forty eight (77.4%) women have never used folic acid and consanguineous relation was present in 52 (83.9%). Other risk factors like maternal medical disorder e.g. diabetes, epilepsy, maternal and paternal ages, family history and maternal smoking were non-significant.

Conclusion: Lack of periconceptional use of folic acid and consanguineous marriages were two most important risk factors. Awareness among the general population and improvement in the antenatal care can help in the early detection and management of congenital anomalies.

Key Words: Congenital anomalies, Consanguinity, Folic acid.

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INTRODUCTION

Congenital anomalies are an important cause of perinatal mortality and morbidity. According to WHO the congenital anomalies can be defined as structural or functional anomalies including metabolic disorders which are present at the time of birth[1]. The exact prevalence of congenital anomalies varies in different areas and among different population. Congenital anomalies affect approximately 1 in 33 infants and results in approximately 3.2 million birth defect related disabilities every year[2]. The proportion of perinatal deaths due to congenital malformation is increasing as a result of reduction of mortality due to other causes owing to improvement in perinatal and neonatal care[3]. In Pakistan about 6-9% perinatal deaths are attributed to congenital malformation[4].

Various risk factors have been identified as contributing factors to these defects which include genetic factors, maternal age, maternal drug intake like anti epileptics, ACE inhibitors etc, radiation exposure, maternal illnesses e.g. diabetes, infection e.g. toxoplasmosis, rubella etc, smoking, folic acid deficiency and consanguinity[5,6]. Some of these risk factors can be avoided.

Antenatal screening can help in early detection of many of these anomalies[7]. Early detection can be helpful in deciding about termination of pregnancy or any therapeutic intervention[8].

To decrease the incidence of various congenital anomalies and their prevalence in the society, it is important that the pattern of distribution, prevalence and

associated risk factors are identified for every country and even for every region[9]. Therefore this study was carried out to find the pattern of different congenital anomalies and the associated risk factors in our local set up. This can help us to modify these risk factors and in the long run can help to decrease the incidence of these anomalies in our own society.

METHODOLOGY

This descriptive study was carried out at the department of Obstetrics and Gynecology, Lady Reading Hospital Peshawar, from March 2012 to April 2013. Women admitted in the labour room with the ultrasound report of congenitally abnormal baby irrespective of gestational age were included in the study. Children in whom the congenital anomalies were diagnosed after delivery were excluded from the study.

After admission, verbal consent was obtained from all the women included in the study to answer a semi structured proforma. The type of birth defect was classified using International Classification of Disease (ICD) 10. Congenital anomalies were divided according to the system involved (central nervous system (CNS), gastrointestinal (GIT), renal, musculoskeletal, face and neck). The fetus was diagnosed as having either isolated (only one system involved) or complex anomaly (two or more system involved). Detailed history was obtained especially regarding the risk factors including maternal medical disorders e.g. diabetes, epilepsy etc, smoking, periconceptional use of folic acid, consanguinity, drug history, maternal and paternal ages and family history of congenital anomalies. Both maternal and paternal ages were ascertained using information from identity card. Three categories of marriages were included i.e., 1st cousin, 2nd cousin and non-consanguineous relationship. All the relevant data was entered in a pre designed semi structured Performa and descriptive statistics were calculated.

RESULTS

During the study period, the total number of deliveries was 4389. A total of 62 antenatal women with ultrasound report of congenitally abnormal fetus were included in the study, making the pre delivery congenital anomalies frequency to be 1.4% of the total deliveries.

Demographic details of the sample are given in Table 1. Most of the patients presented in late pregnancy.

Fifty seven (91.9%) fetuses were having isolated anomalies while in 5 (8.1%) cases multiple organs/systems were involved. Central nervous system anomalies were the most common with 49 (79%) cases and the most common subtype was hydrocephalus 16(25.8%). Renal system was involved in 4 (6.5%) cases while there were two (3.2%) fetuses each having anomalies of the gastrointestinal and skeletal system (Table 2).

Among the different risk factors which were evaluated during the study it was found that consanguinity and lack of folic acid use during pregnancy/periconceptional period were the most common (Table 3). Consanguineous relationship was present in 52 (83.9%) cases, of which 31(50%) were 1st degree relatives and 21(33.9%) were second degree relatives.

DISCUSSION

In our study the incidence of pre delivery congenital anomalies detection was 1.4% of the total deliveries. Raza et al has reported 4.1% incidence of congenital anomalies in the infants[10]. Fifty seven (91.9%) were having isolated anomalies while in 5(8.1%) cases multiple systems were involved. Central nervous system was the most commonly involved system (79%), and hydrocephalus (25.8%) was the most common CNS anomaly. Khan et al in their study have reported 40% CNS anomalies with hydrocephalus as the most common CNS anomaly, skeletal system was involved in 40% cases and genitourinary system in 18%[11]. Similarly Fatema et al have also reported a high incidence of CNS anomalies (46.67%) with hydrocephaly as the most common CNS anomaly (33.3%). Urinary system was involved in 23.3%cases, GIT 6.68%, skeletal system in 5% cases, and 11.6% were having multiple anomalies[12].

In our study 24(38.7%) women presented in the 2nd trimester and 38(61.3%) in 3rd trimester. There was none who presented in the 1st trimester. Fatema et al in their study have also reported that majority (46.67%) of respondents belonged to gestational period between 34-36 weeks with average gestational age of 33.25 weeks[12]. Padma et al has also reported late detection of congenital anomalies, majority of their sample presented between 29-32 weeks gestation[13]. Although pre gestational diabetes is a significant risk factor for the fetus and associated with 2-3 fold increase in anomalies[14], but in our sample diabetes was present in only 5(8.1%) cases. Fatema et al has also reported a low incidence of diabetes in their study (3.33%)[12]. On the other hand Fauzia et al has reported a high incidence of diabetes (25%) in the mothers delivering congenitally abnormal babies[15]. In a local study conducted by Raza et al has documented diabetes in 2.4% and hypertension in 13.3% cases[10].

Another important risk factor in our study was lack of periconceptional use of folic acid. Folic acid was used by only 14 (22.6%) women while 48 (77.4%) have never used it during pregnancy. Neural tube defects were the most common anomalies associated with folic acid defi-

Table 1: Demographic details of the sample (n=62)

Variables		Frequency (%)
Parity	Nullipara	21 (33.4%)
	Multipara	28 (45.2%)
	Grandmultipara	13 (21%)
Period of Gestation	1st Trimester	0
	2nd Trimester	24 (38.7%)
	3rd Trimester	38 (61.3%)

Table 2: Type of Anomaly (n=62)

Type of Anomaly			Frequency
Isolated Anomalies, [57 (91.9%)]	CNS Anomalies, [49 (79%)]	Hydrocephalus	16
		Anencephaly	14
		Meningomyelocele	4
		Encephalocele	6
		Hydroceph+meningomylocele	4
		Anenceph+spinabifida	2
		Microcephalous	1
		Acrania	1
		Dandy walker malformation	1
	Renal Anomalies, [4 (6.5%)]	Polyscystic kidneys	2
		Dysplastic· Multicystic kidneys	2
	GIT Anomalies, [2 (3.2%)]	Omphalocele	1
		Gastroschisis	1
	Skeletal Anomalies, [2 (3.2%)]	Achondroplasia	2
Complex Anomalies, [5 (8.1%)]		Mickel Gruber syndrome	2
		Hydroceph, Omphalocele	1
		Esophageal atresia, Renal anomlies	1
		Facial abnormalities, Talipes, Esophageal atresia, Renal agenesis	1

Table 3: Risk factors (n=62)

Risk factors	Yes (%)	No (%)
Consanguinity	52 (83.9%)	10 (16.1%)
Maternal age >35years	7 (11.3%)	55 (88.7%)
Paternal age >40years	7 (11.3%)	55 (88.7%)
Folic acid intake	14 (22.6%)	48 (77.4%)
Maternal medical disorders	5 (8.1%)	57 (91.9%)
History of anomalies in the previous pregnancies	3 (4.8%)	59 (95.2%)
Smoking	0	62 (100%)

ciency (39 cases). And those who used it, they started it after the pregnancy test was positive at their 1st antenatal visit, and none of them used it in the periconceptional period. Our study was consistent with that conducted by Shawky et al who has reported that 27.5% of the mothers have used folic acid during pregnancy which was significantly lower than the control group[16]. Raza et al in their study has also documented that 63.5% of mothers haven't taken folic acid during pregnancy[10]. Meta analysis has showed that folate fortification had a significant impact in reducing neural tube defects (RR 0.57)[17]. Similarly, Blencowe et al in their meta-analysis has shown a 70% reduction in the recurrence of neural tube defects while primary prevention was 62%[18].

Consanguinity was also an important risk factor for congenital anomalies in our study. Tayabi et al has shown a significant correlation between consanguineous marriages and occurrence of congenital anomalies, p=0.0018[19]. Sheridan et al in their study have reported that consanguinity was associated with a doubling of risk for congenital anomalies. In this multiethnic study 31% of all the anomalies in children of Pakistani origin could be attributed to consanguinity[20]. Similarly other studies have also reported increased incidence of congenital anomalies due to homozygous expression of recessive gene inherited from their common ancestors[21]. Although age of the parents especially maternal age >35 years is a well documented risk factor for chromosomal abnormalities[16] but this was not the case in our study because we have not screened the women for chromosomal abnormalities. In the study conducted by Fatema et al only 3.33% mothers were beyond 35 years[12]. There was no mother of age >35years in the study conducted by Padma et al[13].

Family history was positive in 3(4.8%) cases. Raza et al have shown a positive family history in 19.4% cases[10].

Different studies have shown that congenital anomalies have a significant correlation with smoking[16, 17]. None of the mother who had congenitally abnormal fetus gave history of exposure to smoking in our study. In a local study conducted in Karachi on infants having congenital anomalies only 18.1% of the mother had smoked at least once during their pregnancy[10].

CONCLUSION

Among the risk factors for the occurrence of congenital anomalies, lack of periconceptional use of folic acid and consanguineous marriages were the two most important risk factors in our study. It is therefore recommended that general awareness should be created regarding these risk factors and the periconceptional use of folic acid should be emphasized. Since most of the women presented during late pregnancy, it is important that antenatal care should be emphasized and it should be improved to detect and manage congenital anomalies in time.

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CONTRIBUTORS

RK planned the study, did data analaysis and wrote manuscript. SW, RA and FJ helped in manuscript writing. SJ supervised the study. All authors contributed significantly to the final manuscript.