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CONGENITAL ANOMALIES IN NEWBORN;
ASSOCIATION OF MATERNAL RISK FACTORS

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ABSTRACT: Objective: To determine congenital anomalies in newborn and associated maternal risk factors. **Design:** Cross sectional observational study. **Setting:** Department of Gynecology & Obstetrics unit 1 Lahore General Hospital Lahore. **Period**: From Jan 2007—Dec 2007. **Patients & methods:** All the women who have given birth to babies with congenital abnormalities were recorded. Diagnosis of neonatal congenital anomalies was based on clinical evaluation of newborn by experienced neonatologist. **Results**: During the study period, 2872 patients delivered, of which 48 had congenitally malformed babies, makes the prevalence of 16 per 1000 births. Congenital anomalies were most commonly 52.26% seen in the 21-30 years. The most frequent associated risk factor was history of consanguineous marriage in 45.83%. Neural tube defects were found to be commonest type of anomaly. Among the most frequent neural tube defect was hydrocephalus and anencephaly. **Conclusion:** The commonest associated risk factor was consanguineous marriage the frequency of which may be reduced by creating awareness regarding the avoidance of consanguineous marriages. NTD were the most prevalent anomaly detected and early prenatal diagnosis is helpful in decreasing the indirect prevalence of perinatal mortality by offering early termination.

Key words: Consanguineous marriages, NTD Neural tube defects.

INTRODUCTION

Congenital anomalies remain an important cause of the perinatal mortality. Congenital anomalies refer to structural defects, chromosomal abnormalities, metabolic errors and hereditary disease present at birth. These are gross or microscopic structural defects detected in utero or diagnosed at birth or later on. In Pakistan, about 6-9% perinatal deaths are attributed to congenital malformations[1]. These malformations represents the end result of dysmorphogenesis and can occur as single or multiple entities[2]. Isolated congenital abnormality is the structural defect, which can be traced down to one isolated error in the morphogenesis, while multiple congenital malformations result from two or more different morphogenic errors that occur during the development of the same individual[3].

Common factors responsible for congenital anomalies are chromosomal aberrations, hereditary predisposition and diabetes mellitus[4]. Preconception screening and counseling offer an opportunity to identify maternal risk factor and to recommend proper protective measures even before the pregnancy begins as pre-conception daily intake of folic acid offer a protective effect[5]. About 65-75% cases of congenital anomalies result from multifactorial etiology[6].

Different problems and factors can increase this problem such as; consanguineous marriages can increase the frequency of autosomal recessive disorders[7]. Large family size, which may increase the number of affected children in families with autosomal recessive conditions[8].

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The relatively high birth rate of infants with chromosomal disorders related to advanced maternal age such as Down's Syndrome[9]. Family history of diabetes and previous history of gestational diabetes increase the risk of congenital abnormalities. High grade fever and infections during first trimester increase the risk of CNS anomalies.

In the developing countries early detection of congenital abnormalities by serum markers and ultrasound has reduced the incidence of congenital anomalies. It has reduced the mortality and morbidity by early detection and selective termination of pregnancy[10]. Ultrasound scan can diagnose 70-80% of congenital abnormalities if performed at appropriate time by an expert sonologist[11].

This study was conducted to determine the association of congenital malformations in newborn and associated maternal risk factors.

PATIENTS AND METHODS

This study was conducted at Gynae Unit 2 Lahore General Hospital Lahore during the period of 1st January 2007 to 31st December 2007.Booked as well as unbooked patients were included in the study. Patients once identified ante or postnatally were admitted for further work up, which included the detailed history and examination.

Important points which were asked during history taking were age, parity, infection or fever with rash in the early pregnancy, recurrent abortions, consanguineous marriage, previous large size babies, family history of abnormal babies and diabetes, drug intake during 1st trimester of pregnancy. Detailed physical examination was performed. A detailed physical examination of the baby born with congenital abnormalities was also done. Baseline investigation such as Blood group, Rh factor, complete examination of Blood and Urine and Blood sugar were carried out in all patients, as well as ultrasound of pelvis and abdomen was done. Maternal serum markers were done in few patients. The record of patients was maintained on a proforma with all the investigations and findings.

RESULTS

During the study period, 2872 deliveries occurred and 48 neonates had congenital malformations giving the prevalence of 16/1000 births. Among these, 96% were unbooked patients (less than 2 visits in out patients department). Maternal age ranged from 20 to 39 years and the mean age was 24.51± 5 years. 52.2% were between 21 to 30 years of age and none of them were above 40 years. Out of 48 cases 10 were PG, 23 were G3, 14 were G5 and one case of G11.Out of 48 babies 33 were male (68.75%).

Table I show relevant risk factors associated with congenital anomalies of fetus. Major associated risk factor found in this study was consanguineous marriage (45.83%) which was markedly high. Other frequent factor was with history of fever and non specific infections (20.8%) and none of them had any documented proof of any specific infection. About 14.5% patients presented with previous history of abortion and 10.4% presents with past history of congenitally abnormal babies. Family history of diabetes was seen in 8.3% cases.

Table-I. Associated risk factors for congenital anomalies

Risk factors	No	%age
Past H/O consanguineous marriage	22	45.83%
H/O fever/infection in the current pregnancy	10	20.8%
Past H/O abortion	7	14.15%
Past H/O still birth/NND	5	10.4%
Past H/O congenitally abnormal babies	5	10.4%
H/O diabetes mellitus	5	10.4%
H/O drug intake in pregnancy	4	8.3%
Family H/O diabetes	4	8.3%
Family H/O congenitally abnormal babies	3	6.25%
H/O cardiac diseases in mother	2	4.16%
H/O smoking by mother	1	2.08%
H/O irradiation in pregnancy	0	0

In table II detail congenital anomalies of central nervous system are shown which was found in 38 patients. Among these were 14 cases of hydrocephalus and 9 of anencephaly. These were seen with meningomyelocele and meningocele. Encephalocele and microcephaly were found in lesser number of babies.

Table-II. Congenital Anomalies of Central Nervous System.		
Anomalies	**No**	**%age**
Hydrocephalus	14	29.16%
Anencephaly	9	18.75%
Meningomyelocele	7	14.58%
Meningocele	5	10.41%
Encephalocele	1	2.08%
Spinal bifida	1	2.08%
Microcephaly	1	2.08%

In table III congenital anomalies of musculoskeletal system are shown Talipes equinovarus was seen in cases. Other anomalies were less commonly seen.

Table-III. Congenital Anomalies of Musculo-skeletal System		
Anomalies	**No**	**%age**
Talipes Equinovarnus	4	8.33%
Syndactyl	1	2.08%
Polydactyl	1	2.08%
Club foot	1	2.08%
Achondroplasia	1	2.08%

Other defects included facial malformations, gastrointestinal defects and genitourinary system anomalies as shown in table IV, V, VI, VII.

Table-IV. Facial Anomalies		
Anomalies	**No**	**%age**
Cleft lip	2	4.16%
Cleft palate	1	2.08%

Table-V. Anomalies of gastrointestinal system		
Anomalies	**No**	**%age**
Fetal Ascites	3	6.25
Omphalocele	1	2.08%

Table-VI. Anomalies of Genitourinary		
Anomalies	**No**	**%age**
Ambiguous genitalia	3	6.25%
Undescended testes	2	4.16%
Polycystic kidney	1	2.08%

Table-VII. Miscellaneous anomalies		
Anomalies	**No**	**%age**
Hydrops fetalis	3	6.25%
Vascular navi	1	2.08%

Presence of any chromosomal or genetic disorder could not be investigated and documented in these cases due to non-availability of technical facilities.

DISCUSSION

In our study the prevalence was 16/1000 births. It is note worthy that only 48 babies out of 2872 births with congenital anomalies were diagnosed at birth, systemic malformations like cardiac anomalies, duodenal atresia and diaphragmatic hernia, which are only diagnosed on imaging method, remain unidentified.

The important predisposing factor associated with congenital anomalies include consanguineous marriages which was found to be around 44%.

In first cousin marriages the risk of malformations increases to 5-8 % as compared to 2-3 % incidence in general population. The incidence of congenital anomalies in UAE is 10.5 % where cousin marriages account for 54 %[12]. The percentage of first cousin marriage has been reported as 11.4 % in Egypt and 37.1 % in Pakistan[13]

A study from Rawalpindi depicted 40 % prevalence of congenital anomalies in related parents as compared to 26 % in non-related parents[14].

About 10.4 % mothers were diabetic as compared to 25 % as seen in other study[15]. Incidence of congenital anomalies among diabetic mothers has been reported to be 6-13 % as compared to 1-3 % in general population which can be reduced by strict metabolic control around the time of conception and during period of organogenesis[16,17].

It is also reported that pre-gestational diabetes mellitus is a significant risk factor for developing fetus and associated 3-5 fold increase in major malformation rate.

In this study neural tube defects were the most common congenital anomalies (72 %) NTD was also reported as most common birth defect from India as 4-15/10000 live births and in United States as 1/2000 births[18,19]. Hydrocephalus and anencephaly were the commonest in our study, which is also seen in other studies[20]. This abnormality can simply prevented by folic acid supplementation during pregnancy[21] for which public awareness should be increased. If an early prenatal diagnosis is made and selective termination of pregnancy can be offered. Diagnostic modalities can help in early diagnosis such as ultrasonographic, biochemical tests, chorionic villous sampling, and amniocentesis.

Chromosomal causes are another cause of congenital defects. But in our study it was seen that the congenital anomalies seen in younger age group and mostly seen in 12-30 years of age and only one case of 40 years of mother with congenital anomalous baby seen which is in contrast to other studies where 32 % of mothers were aged 35 years and above[20]. Younger age group may be the reason for not detecting any case of Downs's syndrome in this study.

Rate of cardiac malformations in this study is very low, may be due to under diagnosis because of lack of sophisticated diagnostic techniques and neonatal follow up loss[22]. Increased rate of detection of cardiac anomalies is possible by incorporating four chamber views of heart[23].

Thus by creating awareness among the population regarding the avoidance of consanguineous marriage, periconceptional use of folic acid supplementation, screening of high risk cases and early detection of by routine use of ultrasonography around 16-20 weeks of gestation as well as offering early termination of pregnancy in cases of lethal anomalies, the Perinatal morbidity and mortality can indirectly be reduced.
Counseling of couples after the baby is born with congenital anomalies is very important to prevent recurrence in the subsequent pregnancy.

CONCLUSION

The incidence of congenital abnormalities was 16% or 28/1000 deliveries. Frequency of consanguineous marriages was found as the significant associated risk factor for congenital anomalies. The commonest anomaly detected was neural tube defects. Early screening preferably by ultrasonography should be performed at around 18-20 weeks rather than 1st trimester and offering early termination of pregnancy in cases of lethal anomalies, the perinatal mortality and morbidity can be reduced.

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