Research Article

Study of congenital anomalies during pregnancy

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Abstract

Introduction: congenital anomalies during pregnancy poses major psychological trauma to the parents and family. Neural tube defects are the most common congenital malformations. Various genetic and environmental factors increase the risk of congenital anomalies. **Results**: Total numbers of women with congenital malformations of fetus were 24. Majority of women had Neural tube defects. Most common neural tube defect was anencephaly. Majority of women were between 20 -25 years age group. Around 37% women were second gravida. Majority fetuses were female. H/o recurrence was seen in one woman. **Conclusion**: Early detection and termination of pregnancy will reduce the birth of babies with congenital anomalies and also economical burden, psychological trauma to the parents and family.

Keywords: congenital anomalies, neural tube defects**.**

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INTRODUCTION

Congenital anomalies are one of the most common causes of disability in developed and developing countries. Congenital anomalies during pregnancy poses major psychological trauma to the parents and the family because of the need of termination of pregnancy or if pregnancy continues because of the operative procedure and consequent sequelae that may be present after birth. But these congenital anomalies are of greater interest to Anatomists, Obstetricians, Pediatricians and radiologists. Risk assessment is a part of prenatal care and should begin pre-conceptionally and continue during antenatal period[1]. NTD's are the most common congenital malformations affecting the brain and spinal cord. The incidence of NTD's ranges from 0.5 – 11 /1000 births in different regions of India[2]. Various risk factors, including genetic and environmental factors have been implicated in the causation of congenital anomalies specially NTD's. These risk factors are,

1. Advanced maternal age
2. Previous history of NTD's
3. Nutritional deficiency
4. Inadequate antenatal care also contributes to the increased risk of congenital anomalies in the present pregnancy.

Various studies have reported the teratogenic effect of radiation exposure, maternal hyperthermia, hypervitaminosis A, maternal viral infection and drugs like anticonvulsants which act as folic acid antagonists[3]. NTD's are malformations secondary to abnormal neural tube closure that occur between 3rd and 4th week of gestation.

NTD's are divided into two main groups:

a. Defects affecting cranial structures such as Anencephaly and Encephalocele.
b. Defects involving spinal structures.

Amongst these two groups, cranial malformations are clinically obvious, lethal and incompatible with life while

Spina bifida can range from severe, obvious, open defects to the one which is less easily recognizable[4]. Purpose of our study is to determine the incidence of different congenital anomalies with more focus on NTD's and to study the socio demographic factors related to it.

MATERIAL AND METHODS

This is a prospective, observational study carried out at MNR MEDICAL COLLEGE AND HOSPITAL. All pregnant women who presented with USG report of congenital anomalies in outpatient department as well as in the labour room were included. Unbooked pregnant women who were diagnosed to have congenital anomaly after birth were also included. This study was carried out over period of 2 years, from June 2012 to June 2014. Total numbers of patients with congenital anomalies were 24.

RESULTS

Total number of patients with congenital anomalies including both NTD's and non NTD's were 24. Among these 16 (66.66%) women presented with neural tube defects, most common NTD seen in the study was Anencephaly (n=8) followed by Hydrocephalus (n=4). Less commonly seen NTD's in the study were Meningocele (n=1), spina bifida (n=1), spinal Dysrrhaphysim (n=1) and Dandy Walker Syndrome (n=1). Of 24 women with congenitally anomalous fetuses, 8 had defects other than NTD. Among those 8 women Polydactyly, Syndactyly, Omphaolocele, Prune belly syndrome, Diaphragmatic hernia and Dolicocephaly were found in 1 women each, while Kyphoscoliosis was seen in 2 women. When age wise distribution was considered, it was found that majority of (58.3%) the women were between 20 to 25 years followed by 26 to 30 years of age group. Majority of the (37.5%) women with congenital anomalies were 2nd gravida, while 29.2% of women were primigravida and 3rd gravida, only one women with congenital anomaly was 4th gravida. In the present study, we found that 58.3% of the fetuses with congenital anomalies were female fetuses and remaining were male fetuses. H/o consanguineous marriage was seen in 21.1% of couples. H/o recurrence of congenital anomaly was found in 1 woman. This woman delivered her first baby with Hydrocephalus and unfortunately, in the second pregnancy also she was diagnosed to have a fetus with Hydrocephalus. 33.3% women with diagnosis of fetal congenital anomaly had associated oligohydramnios and 3 women had polyhydramnios. 41.8% of the women at the time of diagnosis were between 21 to 25 weeks gestation. In 3 out of 24 women fetal congenital anomaly was diagnosed at gestational age of less than 20 weeks. While in 4 women (16.6%), the anomaly was not detected antenatally and the anomaly was diagnosed after delivery. All of these women were either unbooked or registered in late 3rd trimester when detection of anomaly is difficult.

Table 1: Age wise Distribution

Age	Congenital anomalies	Percentage
<19 yrs	-	-
20 to 25 yrs	14	58.3%
26 to 30 yrs	8	33.3%
31 to 35 yrs	2	8.3%
>35 yrs	0	0
Total	**24**	**100%**

Table 2: Gravida Distribution

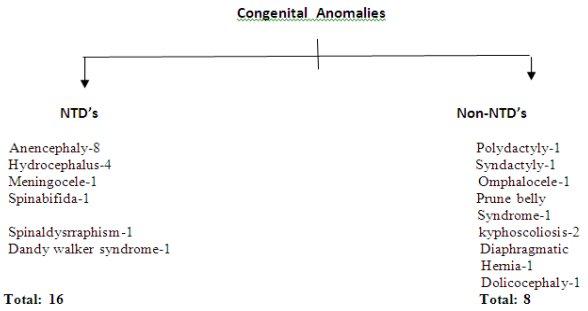
Gravida	Congenital anomalies	Percentage
G1	7	29.2%
G2	9	37.5%
G3	7	29.2%
G4	1	4.1%
Total	**24**	**100%**

Table 3: Gestational age

Gestational age	Congenital anomalies	Percentage
<20 weeks	3	12.5%
21 -25 weeks	10	41.8%
26-30 weeks	4	16.6%
31-35 weeks	3	12.5%
36-40 weeks	4	16.6%
>40 weeks	0	0
Total	**24**	**100**

Table 3: Sex Distribution

Sex	Congenital anomalies	Percentage
Female	14	58.3%
Male	10	41.7%



DISCUSSION

Congenital anomalies can be defined as structural or functional abnormalities including metabolic disorders present at birth. These defects result from defective embryogenesis or intrinsic abnormalities in the development process[5]. Congenital malformations are emerging as important perinatal problem contributing to the psychological trauma and financial burden on the mothers and the families affected. Major congenital anomalies occur in approximately 2-3% of births with a variable frequency in different populations, ranging from 1.07% in Japan to 4.3% in Taiwan[6]. In U.S. among the new born population 3-5% are likely to have a major

congenital malformation and account for more than a quarter of a million affected children each year[7]. The birth prevalence of congenital anomalies in England is 2% and in South Africa it is 1.49% while, its incidence in India is 0.5-11/1000 births in different regions of India. These variations may be explained by racial, social, ecological and economical influences[9]. Incidence of congenital anomalies in our study is 3%. In the study by Delport *et al*, incidence of congenital anomaly was 1.18%[10.] While A.G Jomasir *et al* reported it as 0.29%[11]. Swain *et al* also reported the incidence of malformation as 1.2%[7]. In the present study, NTD's were the most common congenital anomalies accounting for 66.66% of the affected fetuses. Most common NTD's in the study was Anencephaly (33.33%) and Hydrocephalus (16.6%). Swain *et al*[7] also found CNS anomalies to be the most common in their study (39.5%). Anencephaly and Hydrocephalus being the commonest NTD's. Similarly, in the study by Sharada *et al*, the incidence of CNS anomalies was 49.19% [1]. Delport *et al* also found CNS anomalies as the most frequent anomaly accounting for 40 cases, giving an incidence of 2.30 per 1000 live births[10]. While in the study by Rizk Francine *et al*, most of the congenital anomalies were cardio vascular anomalies (16.6%) and limb anomalies (16.6%)[5]. In our study, less commonly seen NTD's were Meningocele (n=1), Spinabifida (n=1), Spinaldysrraphism (n=1) and Dandy walker syndrome (n=1) remaining defects other than NTDs were seen including, Omphalocele, Syndactyly, Prune belly syndrome, Diaphragmatic hernia and Dolicocephaly 1 in each patient. Kyphoscoliosis was found in 2 cases. Majority (58.3%) of the women were between 20-25 years followed by 26-30 years age group. Similarly, in the study by Swain *et al*[7] and A.G. Tomarin *et al*[11] majority of the women diagnosed with congenital anomalies were between 20-34 years. While in the study by Laura *et al*, majority of women were between 30-34 years age group[12]. In the study Sharda *et al* also more number of cases were seen among age greater than 29 years[1]. When parity was considered, 37.5% women were 2nd gravida, 29.2% were primigravida and 29.2% were 3rd gravida and only 4.1% patients were 4th gravida. This indicates that there is no significant difference found with parity in our study. Firouzch Nili *et al*, described that risk of NTD's is higher with increasing parity[3]. In our study, we found that 60.1% of fetuses were female and remaining were male. Swain *et al* did not find any sex prediliction of malformation[7.] While, in the study by Laura *et al* more cases of congenital anomalies were seen in males (1.4%)[12]. Naeimeh Tayebi *et al*, in their study found no effect of gender of fetus associated with prevalence of congenital anomalies[13]. Consanguineous marriages have been described as important risk factor which increases the risk of congenital anomaly[14]. Genetic effect of consanguinity can be traced to the fact that the inbred individual may carry two copies of a gene that was present in a single copy in the common ancestor of his or her consanguineous parents. A recessive gene may thus become prominent first time as inbred descendants after having remained hidden for generations. For this reason consanguinity influences the incidence of some inherited diseases[13,15]. In our study, history of consanguineous marriage was seen in 21.1% of women. Parent's consanguinity was found to be positively associated with congenital malformation in the study by Rizk Francine *et al*[5]. Similarly, in the study by Omatnir *et al*, 14.3% of the major anomalies were seen in women having consanguineous marriage[11]. Mehrabi *et al*[16] showed that, although the consanguinity for malformed fetus was high, there was no significant relationship between malformation and degree of consanguinity. In our study history of recurrence of congenital anomaly was found in one case. This woman delivered her first baby with hydrocephalus and unfortunately in the second pregnancy also she was diagnosed to have a fetus with hydrocephalus. Risk of recurrence of NTD's is approximately 3-4% with the risk being slightly higher if the prior fetus or infant had Anencephaly[17]. In the study by Firouzch Nili *et al*, positive family history of malformation was found in two cases (1.04%)[3]. Jayesh sheth *et al* reported a case of non-consanguineous married couple with recurrent Meningomyelocele in successive pregnancies[18]. Neelam banergee *et al* also reported a case of recurrent neural tube defect in successive pregnancies[19]. Hall TG *et al* found that recurrence rate after index cases with upper NTD's (above T12) was significantly higher than that found in families with index cases with lower NTD's (T11 and below) (3.3% v/s 0.7%). The recurrence rate after an index case with a lower NTD was not significantly different from the base line population risk[20]. In our study, 33.3% women with diagnosis of fetal congenital anomaly had associated oligohydramnios and three women had polyhydramnios. When gestational age was considered, 41.8% of the women were between 21-25wks of gestation. In three out of 24 women, anomaly was diagnosed at less than 20wks gestation. While in 4 (16.6%) women the congenital anomaly was not detected antenatally and was diagnosed after delivery. These 4 women were either unbooked or registered for the 1st time in late 3rd trimester when most of the anomalies were undetected. In the study by Sharada *et al*, all cases were diagnosed after 20wks as antenatal USG was done during 2nd trimester in 10 cases and during last trimester in 22 cases[1]. Firouzch Nili *et al* would not find any correlation between gestational age and the incidence of NTD's[3].

Figure 1: Hydrocephalous

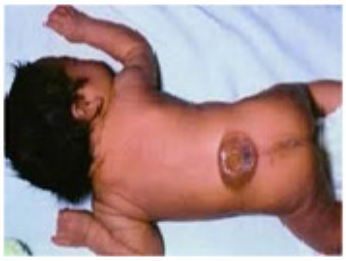


Figure 2: Meningocoele

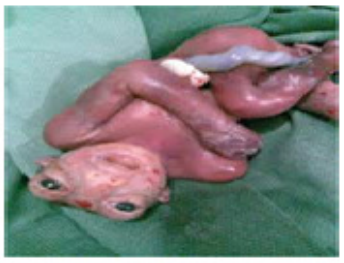


Figure 3: Anencephaly

CONCLUSION

Common congenital anomalies in the present study were NTD's. Anencephaly being the commonest one. NTD's are an enigmatic problem that occurs as a result of the interplay between a number of genetic and environmental factors. Because of this, it is important to avoid exposure to the probable mutagens including radiation, maternal hyperthermia, hypo/hypervitaminosis-A, maternal viral infections and drugs like anticonvulsants. Genetic counselling, prenatal diagnosis and periconceptional folic acid supplementation may reduce the risk of further congenital anomalies especially in parents with previous affected baby. Early detection and termination of congenital anomaly will reduce the birth of babies with congenital anomalies and also economical burden, psychological trauma to the parents and family. Extensive collaboration between Obstetrician, Peadiatrician, Geneticist and Sonologist is required for management of non neural congenital anomaly.

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