

# Pattern of distribution of congenital anomalies in tertiary hospital a prospective study

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## Abstract

**Aims:** To study the incidence of congenital anomalies among the patients in department of obstetrics of Vydehi institute of obstetrics and gynaecology and the contributing factors. **Settings and Design:** This was a prospective study was conducted between 1 January 2012 to 31 December 2012 at department of obstetrics and gynaecology of Vydehi institute of medical sciences and research centre. **Methods and Material:** All the pregnant patients attending the outpatient department of obstetrics and gynaecology were included in the study. Routine ante natal investigations were done. Additional investigations were done in patients with systemic diseases and obstetric complications. Detailed examination of the abortus and the fetus was done in patients who underwent abortion or delivered a still born fetus. Mothers who had live births were examined as a unit within first 24 hours and the baby was followed up till 72 hours of life. **Results:** The incidence of congenital anomalies in our study is 2.1% with central nervous system anomalies being most common among them accounting for 40% of the total anomalies. 70% of the patients with anomalies were in the age group of 20 to 30 years. 21% of the patients gave history of consanguinous marriage. **Conclusions:** This study helps to know the pattern of congenital anomalies in the teaching hospital and the various gestational and familial factors contributing to it. However this study was done in teaching hospital and the sample may not be representative of the population at large. Community based prospective studies are required to identify the pattern of anomalies and contributing factors.

**Keywords:** congenital anomalies

**Key Messages:** Congenital anomalies are emerging as the important cause of neonatal mortality and morbidity of late. It is important to know the incidence of anomalies and various contributing factors.

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## INTRODUCTION

Congenital anomalies are defined as abnormality of physical structure or form seen at birth or few weeks after birth<sup>1</sup>. Infection which was most common cause of infant mortality is dealt effectively by the advent of antibiotics making congenital anomalies as an important cause of

infant mortality. The etiology of congenital abnormality may be genetic in 30–40%, environmental in 5–10%<sup>2</sup> and in about 50% of cases; the cause is not known<sup>3</sup>. The birth of an infant with major malformations, whether diagnosed antenatally or not, evokes an emotional parental response<sup>4</sup>. Parents feel guilty on learning of the existence of a congenital anomaly and anxious about having next pregnancy with abnormal child and require counselling. Early recognition of congenital anomalies are important to plan immediate treatment as in choanal atresia, tracheo esophageal fistula etc. The worldwide incidence of congenital disorder is estimated at 3-7%<sup>5</sup>, but actual numbers vary widely between countries<sup>5</sup>. Prevalent studies of congenital anomalies are useful to establish baseline rates, to document changes over time and to identify clues to etiology. They are also important for health services planning and evaluating antenatal screening in populations with high risk.

## MATERIAL AND METHODS

This study was done in department of obstetrics and gynecology of Vydehi institute of medical sciences and research Centre between Jan 2012 to Dec 2012. All pregnant patients presenting to department of obstetrics were included in the study. A detailed history was taken with respect to the maternal age, history of exposure to drugs and radiation during antenatal period, occupation, family history of consanguinity and anomalous babies, history of systemic diseases, obstetric complications and history of repeated abortions. Routine antenatal investigations, nuchal translucency scan (to mothers presenting before 13 weeks), anomaly scan (to patients presenting before 22 weeks). All patients were screened for gestational diabetes mellitus according to WHO protocol. Additional investigations were done in patients with systemic diseases and obstetric complications. Detailed examination of the abortus and the fetus was done in patients who underwent abortion or delivered a still born fetus. Mothers who had live births were examined as a unit with in first 24 hours and the baby was followed up till 72 hours of life.

## RESULTS

**Table 1:** System wise distribution of anomalies

System	Number	percentage
Cardio vascular	7	25.9
Central nervous	11	40.7
Gastrointestinal	3	11.1
Skeletal system	4	14.8
Facial	2	07.4
<b>Genito urinary</b>	<b>1</b>	<b>03.7</b>

**Table 2:** Maternal profile

Factors	Number	Percentage
<b>Maternal age</b>		
<20	1	5.5
20-30	16	7.8
>30	1	5.5
<b>parity</b>		
primi	7	38
Gravida 2	11	61.1

Gravida4	1	5.5
<b>Family history</b>		
yes	1	0.8
<b>consanguinity</b>		
2 <sup>nd</sup> degree	3	16.6
3 <sup>rd</sup> degree	1	5.5
<b>Mode of delivery</b>		
abortion	8	44.4
Ceasarean section	5	27.7
Full term vaginal delivery	2	11.1
Pre term vaginal delivery	3	16.6
<b>sex</b>		
male	11	61.1
<b>female</b>	<b>6</b>	<b>33.3</b>

**Table 3**

<b>Central nervous system</b>	
Anencephaly	2
Hydrocephalus	3
Meningocele (figure 2,3)	2
Agnesis of cerebellar vermis	1
Spina bifida	1
Encephalocele	1
Craniorachischisis (figure4)	1
<b>Cardiovascular system</b>	
Atrial septal defect	1
Aortic atresia	1
Patent ductus arteriosus	1
Single umbilical artery	1
Single ventricle	1
Hypoplastic left heart	1
Pulmonary artery hypertension	1
<b>Skeletal system</b>	
Skeletal dysplasia	2
Sacral agnesis	1
Poly dactyly	1
<b>Cranio facial anomalies</b>	
Cleft lip (figure1)	2
Cleft palate	2
<b>Gastrointestinal system</b>	
Esophageal atresia	1
Imperforate anus (figure 2)	1
Intraperitoneal cyst	1
<b>Genitourinary system</b>	
<b>Bilateral absent kidneys</b>	<b>1</b>



**Figure 1:** Cleft lip and palate with polydactyly



**Figure 2:** Meningomyelocele, CTEV, imperforate anus, rectovaginal fistula



Figure 3: Meningomyelocele



Figure 4: 16 week foetus with craniorachischisis

During the period of 12 months the numbers of births including live births, still births and abortions were 917 among them 19 fetuses were anomalous. Table 1 enumerates the system wise incidence of congenital anomalies. Central nervous system anomalies (figure 2, 3, 4) were most common accounting to 40% of anomalies followed by cardiovascular system. Facial anomalies (figure 1) accounted for just about 7%. 60% of the anomalous babies were male. Table 2 enumerates the maternal profile in the present study. 78% mothers with the anomalous babies were in the age group of 20 to 30 years. About 70% of deliveries in the study period also happened in mothers of aged between 20 to 30 years. 60% of the mothers were second gravidae. Only one patient gave a family history of babies with congenital anomalies. One patient gave history of intake of norethisterone during periconceptional period. one patient in our study had congenital heart disease so did her baby. one patient had gestational hypertension. 21% of patients gave history of consanguinity.

## DISCUSSION

The incidence of congenital anomalies in our study is 2.1% similar to other studies where incidence varied between 1.5% to 2.1%<sup>6,7</sup>. 78% of the anomalies were found in central nervous system in our study similar to Aobu *et al*<sup>7</sup>. In a study by Singh *et al* musculoskeletal system was the most affected. 78% of the anomalous babies were seen in the woman aged between 20 to 30 years. However 80% of the deliveries in the study period were also in the same age group. *et al* found that the anomalies more in the woman aged more than 35 years<sup>6</sup>. Study by Hollier *et al* showed that advanced maternal age beyond 25 years was associated with congenital anomalies not caused by aneuploidy.<sup>8</sup> Consanguinity increases the prevalence of rare genetic disorders. The incidence of consanguinous marriages in Islamic countries is about 10-45% and the incidence of congenital anomalies is about 7.9%<sup>9</sup>. In western countries where the consanguinous marriages are rare the incidence of congenital anomalies is 3%. Our study had 4 patients

with history of consanguinity. one patient with consanguinous marriage also had family history of anomalous babies. However they refused parental as well as fetal karyotyping. Maternal folate deficiency, diabetes mellitus, iodine deficiency are linked to congenital anomalies. Periconceptional supplementation of folic acid reduces the incidence neural tube defects by 73%. In our study 45% of the patients had iron deficiency anemia. No other micronutrient deficiency was documented. Low socioeconomic status is associated with the anomalies of respiratory system and circulatory system. This finding can be because of nutritional deficiencies in mothers of low socio economic status and also environmental exposures such as indoor pollution<sup>10</sup>. 63% of the anomalous babies belonged to low socio economic status in our study. Maternal Use of recreational drugs, alcohol, antiepileptic drugs have been for long associated with the congenital anomalies in the foetus. Maternal uses of NSAIDs are associated with the cardiac septal defects. Only one patient had used norethisterone in our sample. 60% of the anomalous foetus were male in our study similar to the findings of Singh *et al*. 60% of the woman with anomalous fetuses were second gravidae. Grover N *et al* found an increased incidence of congenital anomalies in among fourth gravidae. Our hospital caters predominantly to urban middle class. Grand multi parae form less than 5% of our patients which explains differing results of our study. Maternal folic acid deficiency, low socio economic status, consanguinity are some of the modifiable risk factors for congenital anomalies in our study. During preconceptional counselling emphasis on folic acid supplementation, avoidance of the use of recreational drugs preconceptionally, avoidance of consanguinity if there is family history of anomalies and referring the patient for genetic counselling are some of the steps to be incorporated in preconceptional counselling to reduce the incidence of anomalies. Incorporation of NT scan and double marker in the routine antenatal care will avoid the late diagnosis of anomalies and emotional turmoil in parents.

## CONCLUSION

This study helps to know the pattern of congenital anomalies in the teaching hospital and the various gestational and familial factors contributing to it. However this study was done in teaching hospital and the sample may not be representative of the population at large. Community based prospective studies are required to identify the pattern of anomalies and contributing factors.

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