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Original Research Article

Prevalance and patterns of congenital anomalies in a tertiary care centre in Pondicherry

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ABSTRACT

Introduction: Advances in ultrasound have helped in better detection of congential anomalies and termination of lethal anomalies at earlier gestational ages These anomalies contribute to major maternal and neonatal morbidity. The prevalence of conge nital anomalies varies with geographical location & ethnicity. In many cases environmental and other maternal risk factors can be identified.

Materials and Methods: The present study is a retrospective cohort study which was conducted in Department of Obstetrics and Gynaecology, Mahatma Gandhi Medical College and Research Institute, Pondicherry over a 3 year period from September 2016 to September 2019. Antenatal women diagnosed with congenital anomalies by imaging who delivered in our hospital were included in this study. Different types of anomalies were classified and risk factors leading to them were assessed.

Results: During the study period out of 6134 deliveries, 140 babies had congenital anomalies leading to a prevalence of 2.28%. 80 of these babies did not have lethal anomalies and survived but medical termination of pregnancy was required in 60 cases. 55% of anomalous babies were males. 60.71% cases were seen in multigravida and 44.3% did not take folic acid in the antenatal period. 25% of cases had history of Gestational Diabetes Mellitus and were on treatment with insulin. Consanguinity was a cause in 27.8% of cases. 72.14% had normal vaginal delivery whereas 27.86% of cases required Caesarean section. Majority of congenital anomalies affected the Central Nervous system accounting for 28.5% of cases followed by gastrointestinal system (20.71%) & musculoskeletal system (20%).

Conclusion: Inspite of good health care facilities in and around Pondicherry, the prevalence of congenital anomalies remains high. Increased awareness and need of proper counselling may help in reducing these anomalies.

1. Introduction

Congenital anomalies are also known as birth defects or congenital malformations. They are important causes of infant and childhood deaths, chronic illness and disability. Congenital anomalies can be defined as structural or functional anomalies (for example, metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth, or sometimes may only be detected later in infancy. Due to these anomalies, an estimated 303 000 newborns die within 4 weeks of birth every year, worldwide.[1] Congenital anomalies can contribute to long-term disability, which may have significant impacts on individuals, families, health-care systems, and societies. The most common, severe congenital anomalies are heart defects, neural tube defects and Down syndrome.

Although congenital anomalies may be the result of one or more genetic, infectious, nutritional or environmental factors, it is often difficult to identify the exact causes. Some congenital anomalies can be prevented. Few methods include vaccination, adequate intake of folic acid or iodine through fortification of staple foods or supplementation, and adequate antenatal care.

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2. Materials and Methods

A Retrospective cohort study which was conducted in Department of Obstetrics and Gynaecology, Mahatma Gandhi Medical College and Research Institute, Pondicherry over a 3 year period from September 2016 to September 2019. Antenatal women diagnosed with congenital anomalies by imaging who delivered in our hospital were included in this study. Different types of anomalies were classified and risk factors leading to them were assessed.

Variables like maternal age, parity, consanguinity, abortions or intrauterine deaths, sibling with malformation, nutrition, addictions, family history of congenital anomalies, conceived after infertility treatment, maternal diabetes, infections, fever and drugs were evaluated. Gestational age at which delivery had occurred, sex, weight of the baby and NICU admission were also noted. Data was collected and analysed by SPSS software.

3. Results

During the study period out of 6134 deliveries, 140 babies had congenital anomalies leading to a prevalence of 2.28%. 80 of these babies did not have lethal anomalies and survived but medical termination of pregnancy was required in 60 cases. Of the 140 babies with congenital anomalies 77 were males whereas 63 were females. In this study the prevalance of congenital anomalies was found to be higher in males at 55%. 55 babies were born to primigravida mothers with a prevalence of 39.3%.

As shown in Table 1, majority of congenital anomalies was seen in the younger age group between 21-25 years (42.1%). Consanuguinity was present only in 27.86% cases. Only 7.86% had history of recurrent abortions or history of IUFD. 5.71% cases had received treatment for primary infertility. Only 55.71% cases had history of folic acid intake. 74.3% had normal BMI. Only 5% had family history of congenital anomalies. 51.43% of congenital anomalous babies crossed 28 weeks of gestation. 25% of cases had history of Gestational Diabetes Mellitus and were on treatment with insulin.72.14% had normal vaginal delivery whereas 27.86% of cases required Caesarean section. 60.71% had a birth weight less than 2.5 kg. (Table 2)

Majority of congenital anomalies affected the Central Nervous system accounting for 28.5% of cases followed by gastrointestinal system (20.71%) & musculskeletal system (20%).(Table 3) Anomalies involving genitourinary system were also common accounting for 11.43% cases. 3.57% cases involved cardiovascular system of which the most common anomaly was Tetrology of Fallot. Syndromic babies accounted for 5% cases. Colloidon baby was the commonest congenital anomaly involving the skin. Sirenomelia (Mermaid baby), Poland syndrome, Pierre

Table 1: Maternal risk factors and congenital anomalies

Maternal Risk Factors	Number of Congenital Anomalies	Percentage
1.Maternal Age		
<21 years	13	9.29%
21-25	59	42.1%
26-30	58	41.4%
31-35	7	5%
36-40	3	2.14%
2. Parity		
Primipara	55	39.29%
Multipara	85	60.71%
3. Consanguinity		
Present	39	27.86%
Absent	101	72.14%
4. History of recurrent abortions or IUD		
Present	11	7.86%
Absent	129	92.14%
5. History of Infertility treatment Present	8	5.71%
Absent	132	94.29%
6. History of		
Maternal Infection	9	6.43%
Drugs	15	10.71%
Folic acid intake	78	55.71%
7. Nutrition Status		
Undernourished	27	19.29%
Normal BMI	104	74.29%
Obese	9	6.43%
8. History of any previous anomaly or Family History		
Present	7	5%
Absent	133	95%
9. Gestational Age		
<12 weeks		
12-20 weeks	40	28.57%
20-28 weeks	28	20%
28-40 weeks	72	51.43%
10. GDM		
Present	34	24.29%
Absent	106	75.71%
11. Mode of delivery		
Vaginal Delivery	101	72.14%
Caesarean section	39	27.86%

Table 2: Fetal factors & congenital anomalies

Fetal Factors	Number of case	Percentage
1. Sex		
Male	77	55%
Female	63	45%
2. Birth weight		
<2.5 kg	85	60.71%
>2.5 kg	55	39.29%

Robinson syndrome, VACTERL group anomalies were among the few syndromic babies. Hydops fetalis was also seen in 5 babies which had a non-immune etiology. The most common CNS anomaly was anencephaly followed by meningocoele, hydrocephalus, spina bifida, Arnold Chiari malformation, holoprosencephaly, corpus callosum agenesis, cystic hygroma and microcephaly. Gastrointestinal anomalies included cleft lip, cleft palate, tracheaesophageal fistula, fetal heterotaxy, anorectal malformation, congenital diaphragmatic hernia, umbilical hernia and situs inversus totalis. Among the musculoskeletal anomalies seen were femoral hypoplasia, CTEV, genu recurrvatum, syndactyly & polydactyly. In 7 cases single umbilical artery was noted. Genitourinary anomalies include penile hypospadisis, renal agenesis, congenital hydrocoele, undescended testis, clitoromegaly. Bilateral congenital cataract was seen in 3 cases with a history of maternal fever in first trimester in 1 case. Malformed ears were seen in 5 cases.

Table 3: Distribution of congenital anomalies according to major system involved.

System Involved	Number of cases	%
Central Nervous System	40	28.5%
Gastrointestinal System	29	20.71%
Musculoskeletal System	28	20%
Cardiovascular System	5	3.57%
Genitourinary System	16	11.43%
Chromosomal anomalies/Syndromes	7	5%
Eyes	3	2.14%
Others	8	5.71%
Skin	4	2.86%

4. Discussion

The percentage of congenital anomalies in this study was 2.28%. This is similar to other studies by Shamma M et al[2] and Shatanik Sarkar et al[3] where the incidence was 2-3%, 2.2% respectively. But global estimates suggest that congenital anomalies affect 2 – 3% of births.[4] Assuming 2% birth prevalence, and 25,595,000 births in 2013, an estimated 511,900 births may have been affected with a congenital anomaly in India.[5]

In our study majority of congenital anomalies were seen between 20-30 years which is in contrast to the study by Kokate et al[6] where maternal age >30 was the most important risk factor. 42.9% of anomalies in our study were lethal anomalies whereas in the study by Kokate et al 80% of the babies were compatible with life and 20% were non compatible. In our study incidence of congenital anomalies was more in multipara with a prevalence of 60.71%. This is in accordance with the study by Pandala P et al[7] where higher percentage of congenital anomalies was seen in birth order more than 4. In our study 51.43% of anomalous babies crossed 28 weeks of gestation which is similar to the study by Kokate et al where the incidence was 72%.

The most common congenital anomaly in our study was central nervous system anomaly followed by gastrointestinal and musculoskeletal anomaly. This is in contast to the study by Vinodh L et al[8] where the most common anomaly detected was musculoskeletal anomaly (24%) followed by CNS and genitourinary system anomalies. In the study by Kokate et al craniospinal anomalies was commonest (44%) followed by musculoskeletal (30%) and syndromic anomalies (12%).

5. Conclusion

The incidence of congenital anomalies in India is around 2.5%.[9] These congenital anomalies account for 13-16% of neonatal deaths and 8-15% of perinatal deaths.[10,11] Preconceptional counselling, folic acid intake and avoiding consanguineous marriages can help in reducing the incidence of congenital anomalies. Proper detection of congenital anomalies by 18 weeks will help patients in planning termination before 20 weeks according to MTP Act. Genetic counselling also plays a role in patients with repeated anomalous babies or syndromic babies.

In utero fetal surgeries have advanced to such an extent reducing neonatal mortality and improving outcome. In places where the patient population are educated and necessary precautions are already taken, the best we can do is to reduce the morbidity and mortality associated with such congenital anomalies.

6. Source of funding

None.

7. Conflict of interest

None.

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