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

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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.
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 prevalence 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%). Consanguinity 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

| 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 |                |            |
| Drugs                         | 9                              | 6.43%      |
| 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 cases | 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, tracheoesophageal fistula, fetal heterotaxy, anorectal malformation, congenital diaphragmatic hernia, umbilical hernia and situs inversus totalis. Among the musculoskeletal anomalies seen were femoral hypoplasia, CTEV, genu recurvatum, syndactyly & polydactyly. In 7 cases single umbilical artery was noted. Genitourinary anomalies include penile hypospadisis, renal agenesis, congenital hydrocele, undescended testis, cleft palate. 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%|

In our study majority of congenital anomalies were seen between 20-30 years which is in contrast to the study by Kokate et al 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 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 contrast to the study by Vinodh L et al 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%. These congenital anomalies account for 13-16% of neonatal deaths and 8-15% of perinatal deaths. 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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