



## Original Research Article

## Pattern of congenital anomalies and associated maternal risk factors : A study from Mysore, South India

Mamatha Shivanagappa<sup>1</sup>, Swathi Hassan Kumarachar<sup>1,\*</sup>, Mahesh Mahadevaiah<sup>2</sup>, Madhumitha Mahesh<sup>1</sup>

<sup>1</sup>Dept. of Obstetrics and Gynaecology, JSS Hospital, JSS Medical College, JSS Academy of Higher Education and Research, Mysuru, Karnataka, India

<sup>2</sup>Dept. of General Medicine, JSS Hospital, JSS Medical College JSS Academy of Higher Education and Research, Mysuru, Karnataka, India



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## ABSTRACT

**Introduction:** Morphological abnormalities arising due to structural defects or abnormal formation of tissues or organs are known as Congenital malformations. Most of the malformations have mixed genetic and environmental causation. Incidence of congenital anomalies differs from country to country and from region to region within same country. This study aimed to the study maternal risk factors and their association with pattern of congenital anomalies in a tertiary care hospital of South India.

**Materials and Methods:** This was a retrospective study. Data from case records of mothers admitted with congenital anomalies in the fetuses were reviewed and analysed. The case records were from the department of Obstetrics and Gynecology, JSS Hospital over a two year study period. There were 47 mothers who had given birth to anomalous fetus and their details were studied with respect to risk factors involved and the pattern of congenital anomalies.

**Results:** The maternal risk factors such as history of previous abortions(27.7%), consanguinity (10.6%), rh-negative pregnancy (6.4%) and anomalous uterus (6.4%) were the most significant factors identified. Congenital anomalies were mostly found to be involving central nervous system(74.5%), musculoskeletal system (29.8%) and cardiovascular system(12.6%).

**Conclusions:** Major maternal risk factor identified among the study group was history of previous abortions. Incidence of anomalies were most involving central nervous system and musculoskeletal system.

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### 1. Introduction

Morphological abnormalities arising due to structural defects or abnormal formation of tissues or organs are known as Congenital malformations. Fetal development is influenced by various genetic and environmental factors leading to defective embryogenesis and intrinsic abnormalities resulting in birth defects in the newborn. Congenital anomalies are third common cause for perinatal mortality in India. It is found that 2.5% of newborns are found to have birth malformation at birth.

Congenital anomaly is present at birth but that itself does not indicate whether the cause is genetic or non genetic. Most of the malformations have mixed genetic and environmental causation. Incidence of congenital anomalies differs from country to country and from region to region within same country. This could be due to different environmental factors, genetic factors influencing environmental factors and also due to variability in the nature of study. There is a paucity of data in this aspect from the southern region of India. This study aims at identifying various maternal risk factors present in the mother with anomalous fetus and most common organ systems involved in congenital anomalous fetus.

\* Corresponding author.

E-mail address: [hkswat2193@gmail.com](mailto:hkswat2193@gmail.com) (S. Hassan Kumarachar).

## 2. Materials and Methods

This was a retrospective study. Data from case records of 47 mothers admitted with congenital anomalies in the fetuses were reviewed and analysed with reference to various risk factors present the pattern of congenital anomalies. The case records were taken from the Department of Obstetrics and Gynecology, JSS Hospital, Mysore City. JSS Hospital is a Tertiary Care Teaching and Research Hospital attached to JSS Academy of Health Sciences and Research (A deemed University) over a two year study period. (Jan 2017 to Dec 2018).

Details of antenatal history was noted which included age of the mother, parity, consanguinity, history of abortions, treatment for infertility, medical comorbidities, history of risk factors associated with congenital anomalies like maternal infections, medications taken for epilepsy, diabetes, hypertension and renal disease. System wise classification of anomalies were done. All the data collected was statistically analyzed and percentage distribution was calculated.

## 3. Results

Primigravida mothers were (48.9%) and multigravida were (51.1%). There was no significant difference noted with reference to the parity of the mothers. Most of the mothers, 59.6% belonged to younger age group of 20-25yrs. Remaining 25.5% belonged to 26-30yrs and rest 14.9% above 30 yrs. 91.5% of the anomalies were detected during the period of less than 28 weeks of gestation. Rest 6.4% and 2.1% in 28-34 weeks and above 35 weeks respectively. (Table 1)

Among the maternal risk factors history of previous abortions (27.7%), consanguinous marriage (10.6%), Rh negative pregnancy and anomalous uterus (6.4%) each. Other significant history pertained to history of prior antiepileptic drug therapy, history of diabetes mellitus, history of hypothyroidism, history of prior infertility treatment (2.1%) each. Risk factors such as exposure to teratogenic treatment, family history of anomalies, pre eclampsia, anemia, cardiac disorders were not present in the study group (Table 2).

Most commonly affected system among the study group was central nervous system 74.5% (35) and Musculoskeletal system 29.8% (14). Other systems like cardiovascular 12.8% (6), gastrointestinal 10.6% (5), genitourinary 8.5%, miscellaneous 8.5% and respiratory 2.1% were affected accordingly. (Table 3).

## 4. Discussion

Congenital anomalies prevalence vary from country to country and within the country due to racial, cultural, social and ethnic influence. It is the leading cause of death in the children below five years of age in high income countries.

**Table 1:** Maternal details

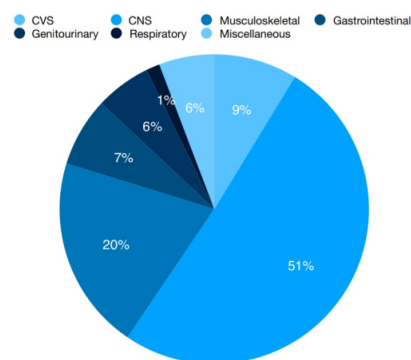
|                     |       | Frequency | Percentage |
|---------------------|-------|-----------|------------|
| Party               | Primi | 23        | 48.9       |
|                     | Multi | 24        | 51.1       |
| Age                 | 20-25 | 28        | 59.6       |
|                     | 26-30 | 12        | 25.5       |
|                     | >30   | 7         | 14.9       |
| Period of Gestation | <28   | 43        | 91.5       |
|                     | 28-34 | 3         | 6.4        |
|                     | >35   | 1         | 2.1        |

**Table 2:** Risk factors

|                               | Frequency | Percentage |
|-------------------------------|-----------|------------|
| Consanguinity                 | 5         | 10.6       |
| Exposure to teratogenic agent | -         | -          |
| Infertility treatment         | 1         | 2.1        |
| Family history of anomalies   | -         | -          |
| History of Abortion           | 13        | 27.7       |
| Anti epileptic drugs          | 1         | 2.1        |
| Diabetes mellitus             | 1         | 2.1        |
| Hypothyroidism                | 1         | 2.1        |
| Pre-eclampsia                 | -         | -          |
| Anaemia                       | -         | -          |
| Cardiac Disorders             | -         | -          |
| Rh negative pregnancy         | 3         | 6.4        |
| Anomalous Uterus              | 3         | 6.4        |

**Table 3:** System wise classification of anomalies

|                  | Frequency | Percent |
|------------------|-----------|---------|
| CVS              | 6         | 12.8    |
| CNS              | 35        | 74.5    |
| Musculoskeletal  | 14        | 29.8    |
| Gastrointestinal | 5         | 10.6    |
| Genitourinary    | 4         | 8.5     |



**Fig. 1:** Diagrammatic representation of system-wise classification of anomalies

WHO reports 3-7% of newborns are born with congenital anomalies every year.<sup>1-4</sup> As per literature higher maternal age is a risk factor for congenital anomalies. In this study, most of them i.e 59.6% (28) belonged to 20-25yrs age and 25.5%(12) belonged to 26-30yrs. Studies by Parmar and Savaskar noted higher incidence of anomalies in mother's age more than 35yrs.<sup>5,6</sup> (Table 1)

Incidence of congenital anomalies were found to decline with increasing birth order in a study by Parmar but study by Swain and Savaskar observed more in multigravida.<sup>6,7</sup> In this study, no significant difference was noted with the parity of the mother and the incidence of anomalies similar to the results by Anand et al.<sup>8</sup> (Table 1)

With respect to period of gestation, abnormal fetuses are likely to be delivered prematurely or aborted as significant number of babies have chromosomal anomalies. The study by Prajapati and Patel had higher incidence of anomalies in preterm babies than term babies.<sup>9,10</sup> Here in the present study, 91.5% of the anomalies detected in less than 28 weeks, either expelled spontaneously or terminated in view of anomaly. Rest 6.4% between 28-34weeks and 2.1% above 35weeks.(Table 1)

The study by Gupta and Verma observed history of previous abortions, anemia, hydramnios, preeclampsia to be associated in mother's with anomalous babies.<sup>11,12</sup> The present study noted that history of previous abortions (27.7%), consanguinity (10.6%), Rh negative pregnancy(6.4%) and anomalous uterus (6.4%). Other risk factors like diabetes (2.1%), hypothyroidism (2.1%) and infertility treatment (2.1%) were also noted.(Table 2)

Among the system most commonly affected 74.5% was Central Nervous System(CNS) followed by 29.8% of Musculoskeletal system. Other systems like Cardiovascular System(CVS) 12.8%, gastrointestinal 10.6%, genitourinary 8.5%, respiratory 2.1% and miscellaneous 8.5% were affected accordingly. (Table 3) There were a case each of sirenomelia, Elli's Van Crevald syndrome, Apert syndrome in the present study.

Studies by Mashuda<sup>4</sup> Gupta<sup>11</sup> and Francine<sup>13</sup> showed similar results to that of our study with CNS malformations being most commonly involved in 41.9%, 16.6% and 29.8% respectively. However the study by Vinitha et al observed genitourinary system to be most commonly involved with 28.5%.<sup>14</sup>

## 5. Conclusion

Major maternal risk factors observed was history of previous abortions followed by consanguinity. Incidence of anomalies were most commonly involving central nervous system and musculoskeletal system.

Pre-conception counselling, antenatal folic acid supplementation and screening of high risk mother's with serum markers, chorionic villous sampling and amniocentesis is the need of the hour for early diagnosis and treatment.

Preventive public health measures which are needed include educating the adolescent girls and mother's regarding risks involved with early and late motherhood, ensuring rubella vaccination, consanguineous marriage, substance abuse and need for adequate intake of iron, folic acid and iodine supplements during pregnancy.

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## 8. Conflict of interest

None declared

## 9. Ethical approval

This was a retrospective study and hence not applicable

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**Mahesh Mahadevaiah** Professor

**Madhumitha Mahesh** House Surgeon

### **Author biography**

**Mamatha Shivanagappa** Associate Professor

**Swathi Hassan Kumarachar** Junior Resident

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