Diagnosis of congenital anomalies, its demographic patterns and associated maternity risk factors among children hospitalized at Lal Ded Hospital, Srinagar

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***Abstract*-** Congenital anomalies (CAs) are a major cause of stillbirths and neonatal mortality in India. The extent and patterns vary over time and across different geographical locations. The objective of the study was diagnosis of CAs using ultrasound (USG), different demographic patterns, types of CA, mode of delivery, gestational age and associated maternal risk factors in both booked and unbooked urban and rural pregnant women at Department of Obstetrics and Gynecology, Lal Ded Hospital (tertiary health care setting), Government Medical College (GMC), Srinagar. Pregnancy outcome of both booked and unbooked pregnant women were followed till delivery for the ascertainment of CAs. The congenital abnormalities were detected during antenatal period using ultrasonography (USG), after delivery and abortion. A total of 5539 mothers were followed till their pregnancy outcome was recorded. Among them 5500 live births were recorded. Rate of incidence of CAs were 0.9/1000 live births. The unbooked women showed indices of 64%, while as, the booked women carried 36% of CAs. Further, rate of CA was comparatively higher among rural women (74%) as compared to the urban women (24%). The pattern in the incidence of CAs observed with regards to maternity parity indicated that multiparity and primiparity women had 64- and 36% CAs, respectively. Results also indicated that incidence of CAs were higher among preterm (64%) than full term births (16%). About 80% of all CAs were diagnosed on USG while as only 20% of CAs were found among non diagnosed participants post delivery. The major maternal risk factors for CAs included polyhydramnios (32%), anemia (24%) and oligohydramnios (20%). Other risk factors included history of previous C-section (16%), breech (10%), Rh negative condition (10%), and events of having twins (4%). Association of CAs with gender was primarily more linked to males (60%) in contrast to females (40%). High percentage of CAs was associated with live (40%) and still births (40%), and a comparatively low level was associated with aborted cases (20%). The predominant defects were observed in CNS (52%) followed by the musculo-skeletal defects (32%), renal (16%), abdominal wall (16%), and cardiac defects (8%). The results of the study indicate that CAs continue to occur majorly among rural women with multiple associated risk factors when compared to the urban women.

I. Introduction

Congenital anomalies (CAs) or birth defects usually include structural or functional irregularities of prenatal origin, resulting from an abnormality occurring during the development process (**Ameen et al., 2018**). Medical science attaches a lot of value to CAs for various reasons, for example, CAs can be a significant parameter to tackle or reduce neonatal mortality (**Ara et al., 2018**). An Indian cohort study established that CA rates are affecting one in forty four births and the prevalence was identical to the stillbirth prevalence, highlighting its public health importance (**Bhide et al., 2016**). Since all congenital anomalies are not lethal, thus, the children born with several non-fatal anomalies survive with disability or need lifelong care, leading to huge out-of-pocket expenditure for families (**Liu et al., 2016**). While CAs are the leading cause of death in children in children less than five years of age in the high-income countries, they are not considered to be significant public health problems in low- and middle-income countries (LMICs) due to public health under-prioritization in such countries (**Bhide et al., 2016**). The leading causes of infant morbidity and mortality in LMICs countries include malnutrition and infections, whereas cancer, accidents and congenital malformations are considered as leading cause in rich countries (**Doddabasappa et al., 2018**).

According to a study conducted by World Health Organization (WHO), 17 - 42% of infant mortality was attributed to congenital anomalies (**Boyle et al., 2018**). This study also documented that ignoring termination of pregnancy with congenital anomaly and stillbirths underestimates the global burden of disease and obscures lack of progress in primary, secondary and tertiary prevention. CAs account for about 8-15% of perinatal deaths and 13-16% of neonatal deaths across the India (**Doddabasappa et al., 2018**). Birth related anomalies such as congenital heart disease caused 261 247 deaths globally in 2017 among infants aged less than one years (**Zimmerman et al., 2020**). Among other documented CAs include kidney and urinary

tract anomalies (**Viswanathan et al, 2020**), preterm births, intrapartum complications, neonatal sepsis, pneumonia (**Bhide et al., 2016**), orofacial defects such as cleft palate and cleft lip (**Neogi et al., 2017**), etc. Anomalies in central nervous system, birth weight, gestational age, male sex, consanguineous marriage, maternal obesity and previous family history of CAs were significantly associated with increased risk of CAs (**Pandala et al., 2019**). Analysis of nationwide data showed that two most frequently reported anomalies in India are anencephaly which is preventable via preconception folate supplementation, and talipes which can be prevented using low cost interventions (**Bhide & Kar, 2018**).

Prevalence rates of CAs recorded in developing countries are likely to be underestimated due to limited of diagnostic facilities or accurate medical records (**Ameen et al., 2018**). A community based study indicated that mothers with risk factors like extreme of ages, illiteracy, worse obstetric history, and history of CAs in previous babies are at increased risk of fetal congenital malformation (**Malik et al., 2019**). In a large study from South India, high prevalence of CAs was found in high risk pregnant (HRP) women when compared to general population, while as low parental age was contributed toward CAs in primiparity women and the consanguinity was also found to be a predisposing factor for CAs in HRP with previous bad obstetric history (**Sunitha et al., 2017**). Usually the major risk factors for CAs include maternal age, drug intake, teratogens, radiation exposure, maternal illnesses, smoking, and alcohol consumption (**Wagner et al., 2019**). Maternal diseases such as rubella infections, diabetes mellitus, hypothyroidism, folic acid deficiency, exposure to alcohol and environmental chemicals are all other factors that cause birth defects (**Doddabasappa et al., 2018**). For accurate and robust testing of CAs, the physicians mostly rely on imaging such as ultrasound and magnetic resonance imaging (MRI). The other antenatal screening methods include maternal serum markers, chorionic villus sampling, amniocentesis, and cordocentesis (**Wagner et al., 2019).**

This study aimed at presenting the spectrum of various CA, demographic patterns, associated maternal risk factors and their outcome on CAs, among both booked and unbooked urban and rural pregnant women, for period of 18 months (March 01, 2018 to August 01, 2019)

II. Materials and Methods

Hospital settings and ethics approval

The current study was conducted at Lal Ded Hospital (tertiary health care setting), Government Medical College, Srinagar, under supervision of Prof (Dr.) Farhat Jabeen, Head of Department, Department of Obstetrics and Gynaecology, with proper approval from Institutional Ethical Committee GMC, Srinagar (Ethical committee report no.).

Participants in study

Pregnant women, both booked and unbooked between time period March 01, 2018 to August 01, 2019, at Department of Obstetrics and Gynecology, Lal Ded Hospital (tertiary health care setting), GMC, Srinagar, were scrutinized for congenital abnormalities (CAs). All CAs were detected either during the antenatal period by ultrasonography (USG), or after delivery and abortion. The record potential participants (pregnant women) were maintained by health staff of Lal Ded Hospital, GMC, Srinagar.

Period of the study

Total period of study was 18 months (March 01, 2018 to August 01, 2019) keeping feasibility, manpower and resources in consideration.

Other formalities

Oral consent was taken from every participant before collecting any information. All unbooked participants were enquired whether or not they have undergone USG or any other radiological investigation. They were also requested to enquire whether any pregnant woman has undergone USG or any other radiological investigation to ascertain whether she was diagnosed with any CA on making a home visit. The workers were advised to keep the detailed address and phone numbers of such women with them so that such women were traced and followed up later by the investigator.

Other details obtained from participants include exposure to any risk factor, history of previous child births, and any other illness among participants. History, examination recordings and USG findings were documented in detail. The booked participants were kept under constant supervision till completion of the study to record any CAs manifestations. All CAs recorded were classified by organ system according to the 10th version of the WHO International Classification of Diseases (ICD-10) (WHO, 1992).

Analysis of data

Data were analyzed using computer software MS Excel. The patient's characteristics were reported as percentages (%) as considered appropriate.

III. Results and Discussion

Participants

A total of 5539 antenatal women including both booked and unbooked at Department of Obstetrics and Gynaecology (OB/GYN), Lal Ded Hospital, GMC, Srinagar during March 01, 2018-August 01, 2019 (18 months). Total number of 5500 live births was recorded by medical staff of hospital. A total of 50 children (30 males and 20 females) had CAs and were recorded and classified as per ICD-10 of WHO (1992). Out of total 50 babies had CAs resulting in the incidence rate of 0.9/1000 live births (approximately). Most of the neonates had single CA, while few of them had multiple CAs.

Demographic pattern of CAs

The current study covered various demographics of CAs among rural/urban, booked/unbooked and primi-/multiparity among pregnant women at Lal Ded Hospital, GMC, Srinagar. Results showed that CAs were seen more commonly seen in rural women (76%) as compared to the urban women (24%). The possible reason for higher rates of CAs found among rural women due to limited resources and advanced diagnostic techniques available in such areas (**Ara et al., 2018**), lack of appropriate genetic counseling (**Doddabasappa et al., 2018**) among other factors. Both booked and unbooked pregnant women were accessed for the rate CAs and the study outcomes showed that

booked exhibited lower rates of CAs (34%) compared to unbooked women who showed 64% CA rate, possibly due to early antenatal scans that helps in prior detection of CAs and possible medical interventions. Another study showed that malformations among newborns born to unregistered mothers were significantly high (**Baruah et al., 2015**). Current study also established a close association between maternal parity and birth defects. Results showed that multiparity is associated with an increased rate of birth defects (64%) compared to primiparity (36%). Some studies show that primiparity was more likely to be related to adverse maternal outcomes (**Schimmel et al., 2015; Wang et al., 2011**) while some studies have shown significant association between multiparity and the occurrence of CAs (**Jawad et al., 2017; Pandala et al., 2019**).

Diagnosis of CAs using USG

The ultrasound (USG) is the most widely used diagnostic tool in obstetrics for detection of developmental disorders among new children prenatally with great certainty (**Erős & Beke, 2017**). In the current study, USG was employed as main diagnostic tool and the sensitivity in the detection of CAs was significantly helpful among pregnant women. Maximum percentage (80%) of CAs were detected using USG, while as a comparatively small percentage (20%) of CAs among pregnant women were diagnosed with diagnostics other than USG. **Kashyap et al. (2015)** also documented 50% early detection of malformation using first trimester USG at tertiary care centre in India, thus, highlighing the need of early diagnosis and timely intervention of prenatal CAs detection using USG.

Delivery mode and age of gestation in case of children born with CAs

In the current study, the mode of delivery and gestational age was recorded for both booked and unbooked pregnant women. In this study, it was seen that 68% of children with CAs were born via vaginal delivery, 20% were aborted fetuses, and 12% of children were born by Lower (uterine) Segment Caesarean Section (LSCS). The gestational age of children with CAs were mostly born through preterm deliveries (64%) with respect to full term deliveries (16%).

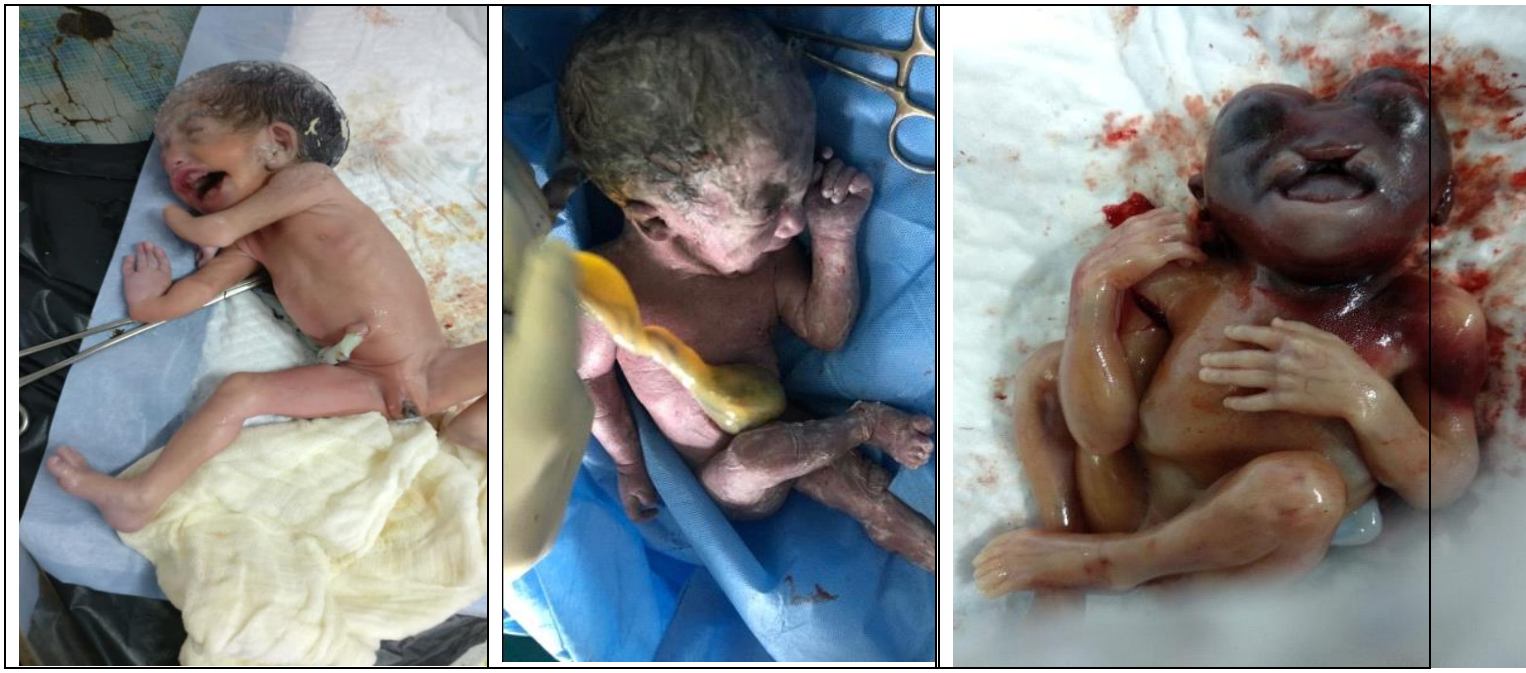
Association of CAs with gender and type of birth in children

In most of the studies, there has been no gender difference in indices of CAs, however, present study found a higher association of CAs with males (60%) as compared to females (40%). Similarly, among CAs positive cases, 296 fetuses were males, while as 274 cases had female gender (**Rehan et al., 2019**).

Furthermore, prevalence of CAs was similar in live and still birth (each 40%), however, higher than rate of CAs in abortive mode deliveries which was only 20%. Similarly, prevalence of congenital malformations was found identical in live and stillbirth in the cohort study carried out by **Bhide et al. (2016).** However, in another study, the incidence of congenital malformations was higher among abortions and preterm deliveries when compared to stillbirths (**Kanhere et al., 2015**).

Different types of CAs in children

In this study, central nervous system (CNS) was the most common system (52%) involved followed by musculoskeletal malformations (32%) (**Fig. 1**). Percentage of renal and abdominal wall defects occurred at same rate (16% each), while as minimum percentage of cardiac malformations (8%) was documented in the study. **Kanhere et al. (2015)** also observed CNS as most common CA followed by musculo-skeletal defects. In another study, CNS defects were the most recognisable malformations at birth as gastrointestinal- and cardiovascular system (**Pandala et al., 2019**). Similarly, 74.4% of patient had CNS congenital anomaly, and the most common anomaly was anencephaly followed by meningocele/myelomeningoceles (Jain et al., 2018). Unlike our results, **Dewangan et al. (2016)** found that majority of the CAs were associated with musculoskeletal system (58%) followed by cardiovascular (28%) and genitourinary (18%). Furthermore, our study found that CNS malformations were better detected in the antenatal USG than any other procedures.



• (b) (c)

Figure 1: The most common CAs included the musculoskeletal (a & b) and anencephaly (c) anomalies in the study

Association of gestational age of babies with CAs

Regarding the gestational age of the malformed neonates, present study found a significantly increased incidence of CAs among preterm neonates (64%) than full term (16%). Similar observation were made by were found by **Doddabasappa et al.** (2018).

Various maternal risk factors and their associated with CAs

Multiple maternal risk factors associated with CAs noted in this study include polyhydramnios (excess amniotic fluid in the amniotic sac), anemia, oligohydramnios (less amniotic fluid in the amniotic sac), previous C - section, breech, Rh negative and twins (**Table 1**). All such maternal risk factors lead to CAs such as malformations in CNS, musculo-skeletal defects, renal, abdominal wall and cardiac defects. Our findings were similar to congenital anomalies (CNS, gastrointestinal and musculoskeletal) reported by **Rathod & Samal (2020).** Invasive antiretroviral therapy, primiparity, pregestational overweight and obesity, folic acid deficiency and smoking were identified as risk factors for vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities (**Van de Putte et al., 2020)**. Nitrate exposure was associated with a significantly increased risk of limb deficiencies, and weakly associated with an increased risk of congenital heart- and neural tube defects (**Blaisdell et al., 2019**).

Table 1— Risk factors and their association with CAs

Risk factors	**Percentage (%)**
Polyhydramnios	32
Anemia	24
Oligohydramnios	20
Previous C section	16
Breech	10
Rh negative	10
Twins	4

IV. Conclusion

Current study tried to understand the prevalence of congenital anomalies (CAs) among women treated (booked/unbooked) at Lal Ded Hospital, GMC, Srinagar. We found a huge rural/urban divide in terms of percentage of CAs in both booked and unbooked pregnant women. Besides, a huge variation was found in terms of percentage of CAs in primi- and multiparity cases. Our study showed that quality USG screening can increase the detection rates of fetal malformations during prenatal period. Furthermore, rate of CAs showed huge variations with respect to mode of deliver and gestational age of the neonate. Higher association of CAs with was found with males as compared to females, and prevalence of CAs was similar in live and still birth, but higher than rate of CAs in abortive mode of deliveries. The most common CAs included defects in CNS musculoskeletal system. Polyhydramnios, anemia, oligohydramnios, previous C-section, breech, Rh negative and twins were the common maternal risk factors that led to different types of CAs reported in the study. Thus, study concludes that public awareness about preventable risk factors is to be created and early prenatal diagnosis and management of common anomalies is strongly recommended. An early diagnosis and management can result in better outcome of these newborns with CAs

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