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ORIGINAL RESEARCH

Distribution of congenital anomalies with respect to associated maternal factors and their detection in antenatal period

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ABSTRACT

Introduction: Congenital anomalies account for 8-15% of perinatal death and 13-16% of neonatal death in India[1]. A study of distribution of congenital anomalies and their early detection allows decisions to be made regarding termination of pregnancy, in-utero treatment, mode of delivery, post-partum management, counselling of the parents regarding prognosis and risk of recurrence.

Methodology: A prospective enrolment of all consenting pregnant women delivering anomalous new-borns within 1 year of study period was done. All study variables were studied in detail. Antenatal ultrasound of all women was noted for weeks of gestation, any fetal anomaly, any liquor or placental abnormality. At the time of delivery, all babies were analysed for gross congenital anomalies.

Results: 33.33% anomalous babies were born to mothers of age group of 30-39 years. 50% women had significant past medical and/or family history. 26.66% of anomalous babies were associated with anaemia in pregnant women. 66.66% cases were detected by ultrasonography, while 26.6% cases were missed on USG. 76.66% anomalies were detected between 28 to 36 weeks of gestational age. Most commonly involved system is central nervous system in 53.33% cases followed by Musculo-skeletal system in 40% cases.

Conclusion: The study emphasizes on preventive measures for congenital anomaly such as, premarital counselling, periconceptional care in the form of improving maternal nutrition, folic acid supplementation, avoidance of teratogenic agents and the need of anomaly scan in second trimester for early diagnosis of congenital anomalies and their termination to prevent perinatal morbidity and mortality.

Key words: Congenital anomaly, antenatal ultrasound.

INTRODUCTION

A congenital anomaly may be defined in terms of physical structure as a malformation – an abnormality usually found at birth or during the first few weeks of life; or defined more widely to include functional disturbances as a defect, any irreversible condition existing in a child before birth in which there is sufficient deviation in the usual number, size, shape, location or inherent character of any part, organ or cell constituent to warrant its designation as abnormal[1,2]. They are an important cause of perinatal morbidity and mortality and account for 8-15% of perinatal death and 13-16% of neonatal death in India[3]. In addition to this, congenital anomalies widely impose heavy direct and indirect costs on health care system. This makes it imperative to detect them early in the pregnancy. Early detection of defects allows decisions to be made regarding termination of pregnancy, in-utero treatment, mode of delivery, post-partum management, counselling of the parents regarding prognosis and risk of recurrence. To facilitate early detection and speedy management, it's crucial to focus on

the distribution of anomalies, involvement of organ or system, associated or causative maternal factors and antenatally detection of anomaly which this study endeavors.

METHODOLOGY

A prospective observational study of all pregnant women delivering anomalous new-borns in a tertiary care hospital of south Gujarat over a period of 1 year from May, 2020 to April, 2021 was conducted. All women delivering anomalous new-borns were explained the purpose of study and those who consented for participation were included. To begin with patient's detailed history was taken, which includes her demographic details, obstetric history (History of previous abortion, previous live/still birth, history of congenital anomalies in her previous children), significant history of any disease and/or drug/radiation exposure, personal history and family history. Antenatal ultrasound of all women was noted for weeks of gestation, any fetal anomaly, any liquor or placental abnormality. Later, all the mothers were examined thoroughly for current pregnancy and managed accordingly. All new-borns were analysed for gross congenital anomalies, functional and/or metabolic defects and were managed accordingly. Mothers were observed during their hospital stay. All collected data was compiled, managed, analysed using SPSS software version 26.

RESULTS

Total 30 patients were found to be delivering anomalous new-borns during study period.

Pregnant women delivering anomalous babies were classified into four groups according to their age. The study result suggests, majority of women delivering anomalous babies belonged to age group of 20-29 years, which is 63.33%. However, 33.33% anomalous babies were born to mothers of age group of 30-39 years (TABLE- 1)

As this study aims to find out associated and/or causative factors for congenital anomaly, significant medical and family history was recorded. 15 out of 30 study subjects had no significant medical history. 10% of the women had history of febrile illness in the antenatal period**,** which includes tuberculosis in two women and history of dengue fever in one woman. 6 out of 30 women (20%) were exposed to drugs in the antenatal period which includes AKT, ART, ovulation agents – clomiphene citrate and aspirin. 20% of study subject had chronic diseases, such as hypothyroidism, diabetes mellitus, HIV infection and poliomyelitis. History of consanguineous marriage was present in 2 study subjects, which is 6.66%. 2 women had history of congenital heart disease in previous babies.(TABLE- 1)

Maximum cases, 8 out of 30 pregnancies (26.66%) were associated with anemia in pregnant women. 4 cases (13.33%) had gestational hypertension and 2 cases (6.66%) had diabetes mellitus. Out of which, 1 had gestational diabetes mellitus, while another had overt diabetes mellitus. 1 woman (3.33%) developed pre-eclampsia in her antenatal period and delivered an anomalous baby. 80% anomalous fetuses had normal volume of amniotic fluid. 16.6% fetuses were associated with polyhydramnios. Only 1 fetus had oligohydramnios, due to neurogenic bladder. (TABLE- 1)

TABLE- 1

Variable	Parameter	Congentinal anomalies present(%)
Age	**<20**	**3.33**
	20-29	**63.33**
	30-39	**33.33**
	>40	**0**
Significant medical history	**Any febrile illness**	**10**
	Exposure to drugs/radiation	**20**
	Consanguinity	**6.66**
	Chronic disease	**20**
	Family history of congenital anomalies	**6.66**
	No significant medical history	**50**

Maternal high risk factors	**Anemia**	**26.66**
	Gestational hypertension	**13.33**
	Gestational diabetes mellitus	**6.66**
	Preecclampsia/ecclampsia	**3.33**

Out of 30 anomalies, 20 cases (66.66%) were detected by ultrasonography. 2 women (6.66%) did not have any antenatal scan. 8 anomalies (26.6%) were missed on USG.(TABLE-2)
Maximum number of anomalies that is 76.66% were detected via USG between 28 to 36 weeks of gestational age. Only 10% cases were detected before 20 weeks of gestation. 6.66% cases were detected between 20-28 weeks. 6.66% cases got detected after 36 weeks of gestational age.(TABLE-2)
66.6% anomalous babies had live birth. 33.3% babies were still born. Out of all still born, 20% were delivered at full term, 30% were delivered at preterm and 50% were delivered before age of viability.(TABLE-2)
Most commonly involved system is central nervous system in 53.33% cases followed by Musculo-skeletal system in 40% cases. Majority of the CNS anomaly had neural tube defect. In anomalies of Musculo-skeletal system, 33.3% had CTEV and 16.6% had defect in cranio-facial skeletal in form of cleft lip alone and along with cleft palate. 6.66% newborns had involvement of gastro-intestinal system in form of imperforate anus and anorectal malformation. 6.66% newborns had involvement of genitourinary system in form of multicystic dysplastic kidney and renal agenesis. Involvement of cardiovascular system was found in 3.33% in form of Atrial Septal Defect with Tricuspid Regurgitation. Single case of respiratory system involvement as Pulmonary Atresia contributed to 3.33% out of 30 anomalous babies. 13.3% had involvement of multiple system. All 4 cases had involvement of CNS and Musculo-skeletal system together.(TABLE-2)

TABLE-2

Variable	Parameter	Percentage congenital anomalies
Gestational age of detection(weeks)	**<20**	**10**
	20-28	**6**
	28-36	**77**
	>36	**7**
Fetal outcome	**Live birth**	**67**
	Still birth	**33**
System involved	**Genitourinary tract**	**6.66**
	Gastrointestinal system	**6.66**
	Cardiovascular system	**3.33**
	Respiratory system	**3.33**
	Muskulosceletal	**40**
	Central nervous system	**53.33**

DISCUSSION

The data gathered as part of study helped to figure out associated factors for the congenital anomaly. Many of these factors are interrelated with each other and play as confounding factors. In this study, three study subjects had more than one positive history. One woman having tuberculosis and taking AKT contributed in two different categories of medical history. The woman with hypothyroidism was found to be exposed with clomiphene citrate and thyroid replacement therapy. One study subject, with history of HIV infection and Diabetes Mellites was taking ART, oral hypoglycemic agent and aspirin falls into three different categories of history for this study.(TABLE- 1)
All these results draw our attention to create more preventive strategy for congenital anomaly. It can be done by rational use of ovulation and induction agents, proper genetic counselling of before and

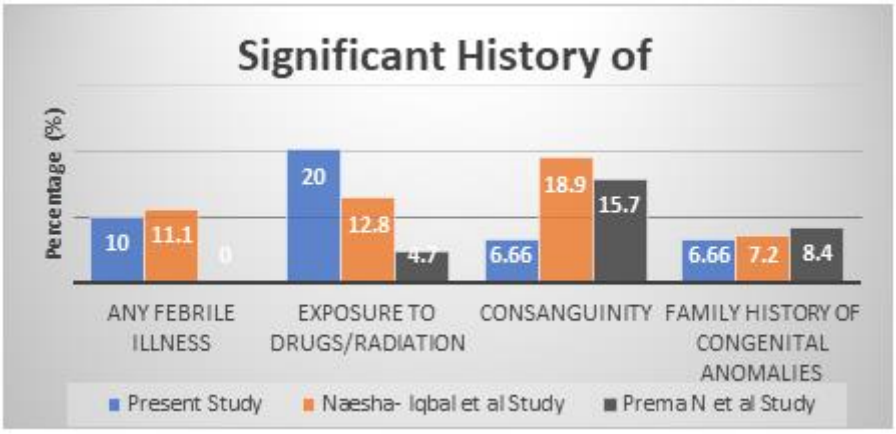
after marriage and advanced research regarding teratogenicity of all the drugs used to control chronic disease.

Association of consanguinity with congenital anomalous varies according to social customs of study population. However, percentage of association of exposure to drugs and consanguinity in study subjects seems to appear higher than general population.

26.66% pregnant women were associated with anemia. 13.33% women had gestational hypertension. 6.66% had diabetes mellites. Higher association of congenital anomalies with anemia can be related to deficiency of Iron and/or Folic Acid, which can be prevented by supplementation with Folic acid in 1st trimester and Iron + Folic acid 2nd trimester onwards.(TABLE- 1)

The study results were compared with Naesha-Iqbal et al study, conducted from 2009 to 2011 and Prema N et al study, conducted from 2015 to 2016. The graphical presentation of result of all the studies is done.(CHART-1)

CHART-1



6 out of 8 missed anomalies were of Musculo-skeletal system (75%) followed by gastro-intestinal system, which includes CTEV, cleft lip, cleft palate, imperforate anus. This result draws our attention to the need of more detailed anomaly scan by using advanced methods in ultrasound examination. All 8 women did not have any early trimester anomaly scan and they had their first ultrasound after 28 weeks of gestational age.(TABLE-2)

Time of detection of an anomaly was noted. Maximum number of anomalies that is 23 (76.66%) were detected via USG between 28 to 36 weeks of gestational age. The anomalies got detected after 36 weeks, include anencephaly and gross hydrocephalus and these women did not have any early trimester scan. Lethal anomalies detected after 28 weeks of gestational age, will be unable to change pregnancy outcome. In most of the cases, reason for late detection is delay in taking antenatal care and visit. Early detection of anomaly should be enhanced through early registration in 1st trimester and timely anomaly scan at least by 18-20 weeks by creating awareness amongst pregnant women and their families. (TABLE-2)

Outcome of anomalous new-borns were studied. 20 out of 30 (66.6%) anomalous babies had live birth. 10 (33.3%) babies were still born. Out of 10 still born anomalous baby 9 had neural tube defect involving central nervous system, while only 1 had defect in Musculo-skeletal dysplasia.(TABLE-2)

Out of 30 anomalous babies, most commonly involved system is central nervous system in (53.33%) 16 cases followed by Musculo-skeletal system in (40%) 12 cases. 2 cases of Musculo-skeletal system had guarded prognosis. One was lethal skeletal dysplasia and another was congenital diaphragmatic hernia. In present study, 4 babies had involvement of multiple system. All 4 cases had involvement of CNS and Musculo-skeletal system together. Out of which, antenatal USG diagnosis of all involved system was done in only 1 case. 2 cases had only diagnosed involvement of CNS. 1 study subject did not undergo any USG scan in her antenatal period. (TABLE-2)

CONCLUSION

This observational study helped to know the pattern of congenital anomalies and the relationship of various gestational and familial factors and the importance of ultrasound in diagnosing anomalies.

This study provided baseline information for future prevention and better management of patients likely to have babies with congenital anomalies.

Most of the congenital anomalies are preventable by proper premarital counselling, periconceptional care in the form of improving maternal nutrition, folic acid supplementation, avoidance of teratogenic agents. Maternal anemia and malnutrition pose a serious threat to both mother and fetus, hence fortification of food with iron, folic acid, iodine should be carried out. Anemia should be tackled early in reproductive age group. High risk patients should be counselled and prepared in pre and periconceptional period. Genetic counselling at different time period helps in reduction of congenital anomalies, morbidity and mortality resulting from these anomalies.

Most of the anomalies can be picked up by fetal anomaly scan in early mid trimester by 16-18 weeks. The study emphasizes the need of anomaly scan in second trimester for early diagnosis of congenital anomalies and their termination to prevent perinatal morbidity and mortality. We would recommend nuchal translucency scan in all high-risk pregnant women for early detection of chromosomal anomalies.

Facilities for management of anomalous new-borns should be made available at tertiary care hospitals. However, parents of any surviving anomalous child should receive emotional support and reassurance.

REFERENCES

1. Hudgins L, Cassidy SB. Congenital anomalies. In: Martin RJ, Fanroff AA, Walsh MC, eds. Neonatal–perinatal medicine. 8th ed. Philadelphia: Mosby-Elsevier 2006:561-81.
2. Ali A, Zahad S, Masoumeh A, et al. Congenital malformations among live births of Arvnd Hospital, Ahwaz, Iran–a prospective study. Pak J Med Sci 2008;24(1):33-7.
3. Park K. Congenital malformations. In: Park K, ed. Park's textbook of preventive and social medicine. 15th ed. 2005:379-80.
4. Desai NA, Desai A. Congenital anomalies: a prospective study, Bombay hospital. Indian Journal of Pediatrics 2005; 70:213-23.
5. Naseha A, Iqbal Y. Incidence of congenital anomalies in tertiary health care centre. J. Evolution Med. Dent. Sci. 2016;5(67):4826-4833, DOI: 10.14260/jemds/2016/1099
6. Prema N, Sudhakaran R, Divya. B.V, Meerabai V, Maharani. Prevalence and associated factors of congenital anomalies in a tertiary care centre in Tamilnadu. Obs Rev:J obstet Gynecol 2016;2(4):55-63.DOI: 10.17511/joog.2016.i04.03.
7. Daliri S, Sayehmiri K, Asadollahi Kh, Rezaei N, Saroukhani D, Karimi A. Prevalence of Congenital Anomalies in Iran: A Systematic Review and Meta-analysis. Iranian Journal of Neonatology. 2018 Jun: 9(2).