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STUDY OF GROSS CONGENITAL MALFORMATION IN NEWBORN

Obstetrics & Gynaecology

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ABSTRACT

BACKGROUND : According to WHO congenital anomalies are defined as structural or functional anomalies including metabolic disorders which are present at the time of birth .Congenital Anomalies are a major health problem accounting for 8 to 15% of perinatal deaths and 13 to 16% of neonatal deaths in india .This study was done to know the frequency ,pattern of congenital anomalies and various risk factors and predisposing factor ,which may help to develop strategies for patient counselling and management . The objective of study was to present the spectrum of various congenital anomalies , epidemiological features of pregnant women with anomalous fetus.

METHODS : Retrospective ,analytical hospital based study of 252 patient who delivered or aborted congenital anomalous babies from a period of june 2016 to june 2017. Relevant information regarding maternal age , parity , gestational age, sex and the outcome was documented

RESULTS: During the study period 252 congenital anomalies were seen in delivered babies and aborted fetus. The incidence of congenital anomaly in our study was 2.42%. It was seen more in women with increased parity. Incidence of congenital malformations was higher among those who aborted or were delivered preterm .Central nervous system was the most common system involved followed by musculoskeletal system . Anencephaly was the most common malformation seen in patients

CONCULSION: Our study concluded, that the number of congenital anomaly was recorded more in patient with higher parity and age .Central nervous system was commonly involved. Early diagnosis, antenatal Ultrasonography , proper counselling for this pregnancy and subsequent pregnancy is needed for proper management of the problem

KEYWORDS

Congenital Anomalies(CA) , newborn, central nervous system, prematurity, still birth.

INTRODUCTION

Birth defects, congenital abnormalities and congenital anomalies (CAs) are interchangeable terms used to describe developmental defects that are present at birth. [1] CAs are a diverse group of disorders of prenatal origin that can be caused by single gene defects, chromosomal disorders, multifactorial inheritance, environmental teratogens and micronutrient deficiencies. CAs may be inherited or sporadic, isolated or multiple, gross or microscopic [2, 3]. Early intrauterine period during the first trimester, especially the 3rd to 8th week of gestation, is the time period for organogenesis of the fetus. Maternal infections such as rubella, maternal illnesses like diabetes mellitus (DM), iodine and folic acid deficiency, exposure to medicinal and recreational drugs including alcohol and tobacco, certain environmental chemicals, and doses of radiation are some of the major factors implicated in the causation of Cas in developing fetus[3].

The present study was carried out to determine the overall incidence, epidemiology and distribution of various CAs in fetus and newborn delivered at our teaching hospital, as well as the relation of CAs to various fetomaternal risk factors.

AIMS AND OBJECTIVES

- To study the incidence and epidemiology of CAs in fetus and newborn delivered at this hospital.
- To assess the association of occurrence of CAs with demographic profile of the parents including age and parity.
- To identify various associated maternal high-risk factors.

METHODS

The study conducted within time period of 12 month from june 2016 to June 2017.Antenatal history included Maternal age, parity and gestational age at the time of diagnosis, history of drug intake , associated Maternal condition, previous history of affected children. All live birth and still born babies were examined for the presence of congenital malformation at the time of birth. Detail general and systemic examination of the baby was carried out. Baby's gestational age and birth weight were noted. For antenatal diagnosis ,2-D USG was used as diagnostic modality in most cases because of ready availability in our hospital. X-Ray was used to diagnose many skeletal disorders only in postnatal babies to avoid intrauterine radiation exposure.

OBSERVATIONS AND DISCUSSION

- From a total 10,391 deliveries, which included both livebirths as well as IUFD/stillbirths at the institute over the study period, there were 252 babies born with congenital anomalies. The incidence of CAs is 2.42%. This rate co-relates with the rates of incidence of anomalies in studies carried out in areas having similar socio-demographic profile, from Indian cities of Kolkata, Wardha& Ahmedabad , and other countries like Egypt, Iran, Nigeria and Pakistan [12,14,15,22].

TABLE 1: INCIDENCE OF CONGENITAL ANOMALY

No. of Deliveries	No. of babies with anomaly	Incidence (in %)
10,391	252	2.42%

- In this study, the incidence of CAs per 100 deliveries increases with increasing maternal and paternal age. Conception and delivery at later age has more probability of occurrence of genetic defect as well as chromosomal abnormality, which can lead to more number of CAs. Down's syndrome is a classic example of age-associated congenital anomaly. This trend corresponds to studies by Parmar[18], Anjum[19] and SugnaBai[20].

TABLE 2: DEMOGRAPHIC DETAILS AND CHARACTERISTICS

Charact eristic	Total number of deliveries (n=10,391)	No. of anomalous baby (n=252)	Incidence (in %)

	1.Maternal Age (M)	2.Paternal Age (P)	1.Maternal Age (M)	2.Paternal Age (P)	1.Maternal Age (M)	2.Paternal Age (P)
Up to 20 years	988	926	19	17	1.92	1.83
21-25 years	4224	2986	106	61	2.51	2.04
26-30 years	3599	3884	94	98	2.63	2.52
31-35 years	1518	1198	31	54	2.05	2.70
More than 35 years	62	597	2	22	3.22	3.68

- With relation to parity, higher incidence was found in higher order pregnancies. The probable explanation of this phenomenon can be the fact that higher order pregnancies are usually seen at higher maternal age. Moreover, this data correlates to the data from studies by Sarkar[13] and Mohanty[21].

Characteristic	Total number of deliveries (n=10,391)	No. of anomalous baby (n=252)	Incidence (in %)
3.Order of Baby (Parity)			
Primipara	4580	100	2.18
2nd	2899	69	2.38
More than 2nd	2912	110	3.77

- Out of the total 252 cases of CAs in this study, more than 2/3rd of the foetuses delivered pre-term. This can be explained by the phenomenon of natural selection. The results also correspond with Sarkar[13] and El Koumi[17].

Characteristic	Total number of deliveries (n=10,391)	No. of anomalous baby (n=252)	Incidence (in %)
4.Maturity			
Pre term (<37 weeks)	1710	149	8.74
Full term (≥37 weeks)	8681	103	1.18

- A significantly higher incidence of congenital anomalies was observed among the still birth(14.92%) in the present study as compare to live births(1.84%). It is consistent with earlier reports of Parmer[18] ,Aiyar[23] and Agrawal[23].

Characteristic	Total number of deliveries (n=10,391)	No. of anomalous baby (n=252)	Incidence (in %)
5. Live born / Stillborn			
Live births	9922	182	1.84
Still births / IUFD	469	70	14.92

- The incidence of CAs in male babies was 1.56 times that of female babies in our study, with 1% of anomalous foetuses having ambiguous genitalia. Other studies of Taksande[12], Chaturvedi[24], A Khan[15] and Mohanty[21] corroborated the findings. While in El Koumi[17] and K Shah[14] studies, no significant difference was observed. In Parmar study, the incidence was more in female babies.

Characteristic	Total number of deliveries (n=10,391)	No. of anomalous baby (n=252)	Incidence (in %)
6.Sex of Baby			
Male	6452	180	2.78
Female	3937	70	1.77
Ambiguous	2	2	100

- The maximum cases of CAs in this study affected the nervous system, followed by the musculoskeletal system and gastrointestinal system. The possible explanation of higher percentage of nervous system defects can be that they are more evident at birth and can be recorded more carefully than other defects. Among the nervous system anomalies , the maximum number of cases were seen of anencephaly , followed by spina bifida and hydrocephalus.

TABLE 4: SYSTEM-WISE INCIDENCE OF CONGENITAL ANOMALY

System	Present Study (n=200)	El Koumi, Egypt17 (n=252)	Ekwere, Nigeria16 (n=200)	Sarkar, Kolkata13 (n=286)	Taksande, Wardha12 (n=179)
1.Nervous (CNS + Neural Tube defects)	25.5%	20%	24.5%	11.2%	15%
2.Gastrointestinal	20.5%	16%	30.5%	15%	17%
3.Genitourinary	12.5%	13%	5%	10.5%	13%
4.Musculoskeletal	22%	19%	15.5%	33.2%	24%
5.Cardiopulmonary	8.5%	9%	7.5%	10.9%	11%
6.Others	11%	23%	17%	19.2%	20%

- Diabetes mellitus is a known risk factor for CAs, which was corroborated in the present study - 15%. Maternal hyperthermia during organogenesis is also a leading risk factor associated in 13% of cases in this study. A 1988 study by Anand[25] et al. suggested maternal hyperthermia during organogenesis to be a significant risk factor for the occurrence of CAs.

TABLE 5: RISK FACTORS ASSOCIATED WITH ANOMALIES*(More than one risk factors were present in many cases)*

Risk Factors	No. of Cases %
Fever during first trimester (>100® C for 48 hours)	13
Anaemia (Uncorrected moderate anaemia <9 g/dl)	8
Pre-eclampsia (BP >140/90 mmHg with albuminuria)	7
Diabetes Mellitus	15
Polyhydramnios (AFI > 25 cm)	32
Oligohydramnios (AFI < 5 cm)	25
History of Recurrent Pregnancy Loss (>3 Spontaneous Abortions)	5
History of anomalous child in previous pregnancy	12
Epilepsy	4
Malformation of Uterus	7
Conceived after treatment for Infertility	2

SUMMARY

The incidence of CAs in this study is 2.42%. The incidence of CAs per 100 deliveries increases with increasing maternal & paternal age and higher order of parity. Out of the total 252 cases of CAs in this study, more than 2/3rd of the foetuses delivered pre-term. The incidence of CAs in male babies was 1.57 times that of female babies in our study, with 1% of anomalous foetuses having ambiguous genitalia. The maximum cases of CAs in this study affected the nervous system, followed by the musculoskeletal system and gastrointestinal symptoms. Among the nervous system anomalies, the maximum no. of cases were seen of anencephaly, followed by spina bifida and hydrocephalus. Most common gastrointestinal anomaly seen is Tracheooesophageal fistula ,followed by duodenal atresia and omphalocoele. Among the musculoskeltalsystem,cleft lip and cleft palate were more common followed by talipesequinovarus while in genitourinary system, unilateral or bilateral renal agenesis and ectopic kidney were commom. Maximum cases of Ventricular septal defect were seen from cardiopulmonary system.

DISCUSSION

Congenital anomalies are the major cause of the new born deaths within first few weeks of birth and can result in long term disability with a significant impact on individual , families, societies and health care system. In 50 % cases exact cause is not identified but the risk factors can be linked with the causation of malformation.

The structural defects are obvious but there are certain functional defects like haemophilia(bleeding disorders) are not diagnosed until infancy. Thus, we should investigate thoroughly for the mothers with risk factors an try to prevent as well as early diagnose the congenital anomaly.

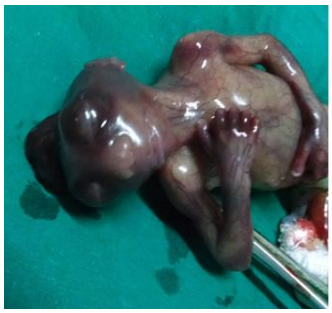
CONCLUSION

The present study concluded that incidence of congenital anomaly is 2.42%.Though with increasing awareness among people and better

healthcare facilities congenital anomalies are diagnosed earlier and interventions planned accordingly. With increasing age and increasing parity there is more incidence of CAs.

Central nervous system anomaly was commonly involved ,anencephaly being the commonest. Cause of neural tube defect is folate deficiency. Therefore folate supplementation in target population can help in reducing the incidence of neural tube defect. Increasing awareness of maternal care ,early diagnosis, antenatal USG ,proper counselling for this pregnancy and subsequent pregnancy can take care of the couple to face this dreaded complication of pregnancy
A early detection and termination of fetus and congenital anomalies will reduce birth of babies with congenital anomalies. It will also ease the economic burden, psychological trauma to patient and family. Collaboration between obstetrician , physician, genetist and sonologist is required for management of viable congenital anomalies.

COLOUR PLATE - A



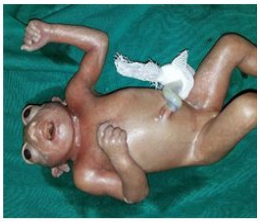


Figure 1 and Figure 2 – Anencephaly

COLOUR PLATE - B

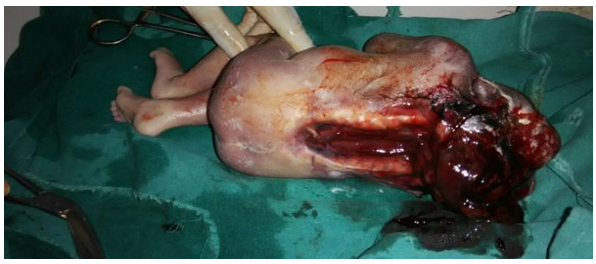


Figure 3 - Open Neural Tube Defect with Anencephaly

Figure 4 – Open Spina Bifida

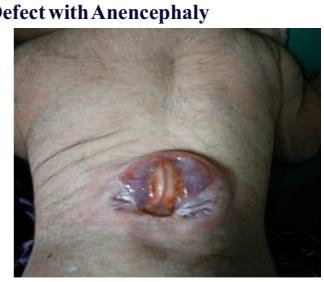


Figure 5 – Meningomyelocele

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