

Original Research

Congenital Anomalies in Second and Third Trimester- An Enigma

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ABSTRACT:

Objectives: To evaluate the antenatal incidence of major congenital abnormalities and its pattern of distribution. **Materials and Methods:** Retrospective, analytical hospital based study of 1162 delivered patients who aborted or delivered congenital anomalous babies for a period 6 months. Relevant information regarding maternal age, parity, gestational age, birth weight, antenatal ultrasound was documented. **Results:** Out of total 1162 live /still birth, 20 feti were found to have congenital malformations. The overall incidence being 1.72% (17.2 per 1000 births). Commonest anomalies were of Central Nervous System followed by other systems. **Conclusion:** Regular Antenatal visits, prenatal diagnosis (Level 11 Scan) are recommended for early intervention and even planned termination and appropriate treatment after birth when needed. Folic acid intake is recommended for prevention of Neural Tube Defects in future pregnancy.

Key words: Second and Third Trimester, Congenital Anomalies.

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INTRODUCTION:

Congenital anomalies (birth defects, congenital disorders or congenital malformations) are defined as structural or functional anomalies that occur during intrauterine life and can be identified prenatally, at birth, or sometimes may be later in infancy.¹ The incidence is 3-5% of live births in the United States, but in India the reported incidence is 2.5%. Congenital anomalies accounts for 8% to 15 % of perinatal deaths and 13% to 16% of neonatal deaths in India.²

Prevalence is as low as 1.07% in Japan and as high as 4.3% in Taiwan. These variations of prevalence may be explained by social, racial, ecological and economical influences.^{3,4}

In India, Urban Infant Mortality Rate (IMR) is 27 as compared to Rural IMR is 44 per 1000 live births with sepsis, pneumonia and diarrhoea being major causes of mortality in rural areas. The transition in causes of mortality in urban areas is accelerated by availability of standard maternal and pediatric services⁵ resulting in

increasing contribution of congenital anomalies to neonatal deaths.

Screening and diagnostic techniques have been developed which makes it possible to diagnose congenital anomalies early. This enables obstetrician and perineonatologist to offer counseling regarding continuation or termination of pregnancy and decide on time and place of delivery. It also allows for identifying the need of postnatal intervention including surgery.

Advances technology have provided the ability to offer fetal therapy instead of resorting to termination of pregnancy, if identified early. The prenatal ultrasound at 18 to 20 weeks is optimum time where a fetus can be labeled with a good degree of accuracy. Approximately 70% of major anomalies and 45% of minor anomalies can be detected at this time. Moreover, this is the time when dating of pregnancy can be done accurately with 10 weeks + 7 days.

Medical and Rehabilitative services for affected children through govt health services in India are currently limited,

resulting in out of pocket expenditure for affected families. Some of these conditions can be prevented by primary care intervention targeted towards women in pre conception, intra conception and antenatal period.⁶⁻⁸

Congenital Anomalies are important causes of infant and childhood deaths, chronic illness and disability. The resolution of sixty third World Health Assembly (2010), Member States agreed to promote primary prevention by:

1. Developing and strengthening registration and surveillance system.
2. Developing expertise and building capacity.
3. Strengthening research and studies on etiology, diagnosis and prevention promoting international cooperation.¹

Around 40%-60% of congenital anomalies are of unknown etiology. 20-25% of anomalies the cause is multifactorial. 10-13% are attributed to environment and 12-25% to genetic causes.^{9,10} The most prevalent congenital anomalies are congenital heart defects, neural tube defects and Down syndrome.¹¹⁻¹³ All congenital anomalies are not lethal. Babies born with several non fatal anomalies survive with disability or need life long

care, leading to catastrophic expenditure for affected families.¹⁴⁻¹⁶ The true magnitude of the number of births affected by congenital anomalies in India is unknown due to lack of National Birth Defects Surveillance.¹⁷

MATERIAL AND METHODS:

A Retrospective, analytical hospital based study of patients was undertaken who aborted or delivered congenital anomalous fetus or baby for a period of 6 months in our Obstetrics and Gynaecology unit of Bebe Nanaki Mother and Child Care Centre of Government Medical College, Amritsar There were a total of 1162 deliveries noted in this period. Relevant information regarding maternal age, parity, gestational age, mode of delivery, fetal outcome and folic acid intake was documented. Significant antenatal history like maternal illness, ingestion of drugs, exposure to radiation and complications of labor was recorded. Whether the patient had an antenatal ultrasonography (USG) scan and findings were noted. All the aborted fetus and newborns were examined for congenital malformations soon after delivery.

RESULTS:

During the study period congenital anomalies was seen in 20 delivered aborted foetus and delivered babies.

TABLE 1: DEMOGRAPHIC PROFILE

	Number of patients (n=20)	Percentage (%)
Maternal Age		
<19	1	5.00%
20-29	17	85.00%
>30	2	10.00%
History of Folic acid intake		
Present	7	35.00%
Absent	13	65.00%
Literacy		
Illiterate	3	15.00%
Primary School	17	85.00%
Parity		
Primi	5	25.00%
Multi	15	75.00%

Table no.1 shows 1(5%) patient was in age group <19, 17(85%) patients were in age group (20-29), >30 years were 2 (10%). 3 (15%) patients were illiterate and 17(85%) patients had done primary schooling. All the patients were unbooked and were from rural area. 5 (25%) patients were primigravida and only 7 (35%) of patients gave definitive history of folic acid intake.

TABLE 2: ULTRASONOGRAPHY AT DIFFERENT GESTATIONAL AGE

	Number of Patients (n=20)	Percentage
Ultrasound		
Live fetus at scanning	11	55.00
IUD fetus at scanning	9	45.00
Gestational Age		
2 nd trimester	5	25
3 rd trimester	15	75

The incidence of congenital malformation was 1.72% among 1162 (total births) during the 6 months period. 11 (55%) fetu were live at the time of scanning, 9 (45%) fetu were IUD. 5(25%) patients were diagnosed with congenital anomalies in 2nd trimester and 15(75%) in 3rd trimester ultrasonography respectively. Three (15%) of the fetus had multiple congenital anomaly.

TABLE 3: ANOMALIES INVOLVING DIFFERENT SYSTEM (N=20)

Category	Number (N=20)	Gestational age of detection
CNS		
Hydrocephalous	2	29 weeks, 34.4 weeks
Anencephaly	4	14 weeks, 17.4 weeks, 28 weeks, 28 weeks
Encephalocoele	1	19 weeks
Arnold Chiari	1	20.4 weeks
Spina Bifida	1	29 weeks
Holoprosencephaly	1	25 weeks
Caudal regression	1	29 weeks
Dorsal Spine Splaying	1	28 weeks
Dandy Walker vermian hypoplasia	1	34.2 weeks
GIT		
Gastro Chisis	1	After Delivery
Abdomen not formed	1	28 weeks
CVS		
Tricuspid regurgitation	1	36.3 weeks
All Heart chambers dilated	2	34.3 weeks, 32.4 weeks
Pericardial effusion	2	34.3 weeks, 32.4 weeks
Musculoskeletal		
Skeletal dysphasia	1	17.6 weeks
Others		
Congenital cataract	1	After delivery
Cleft lip and palate	2	After delivery
Down syndrome	1	After delivery
Cystic hygroma	1	25 weeks
Cystic thoraco abdominal mass	1	25 weeks

CNS was the most common system affected followed by CVS. Anencephaly was the commonest malformation seen 4 (20%). Some of the congenital anomalies, which were not diagnosed on USG, were detected after delivery (congenital cataract and cleft lip and palate). A case of Down syndrome was detected after delivery because the patient had not got level II scan done and triple/quadruple test for ruling out Trisomy 21 and Trisomy 18 could not be done.

TABLE 4: DISTRIBUTION OF CASES ACCORDING TO FETAL OUTCOME

Mode of Delivery	No. of Patients	Percentage
Preterm vaginal delivery	6	30
LSCS	9	45
Expulsion	5	25

Fetal Outcome	Number of fetus
Female	
Alive	8
Dead	8
Male	4

Out of 20 cases, 9(45%) Patients were operated for caesarean section due to obstetrical causes and 6(30%) had preterm vaginal delivery and 5(25%) expelled congenitally malformed fetu. Out of which 8 females were alive and 8 were still born and 4 live male were born.

TABLE 5: HIGH RISK FACTORS ASSOCIATED WITH PREGNANCY

Risk Factors	Number of Patients (n=20)	Percentage
Hypertensive disorder in pregnancy	3	15.00
Epilepsy	1	5.00
Thyroid Disorders	1	5.00
Previous 1 caesarean	5	25.00
Previous 2 caesarean	4	20.00

Various associated risk factors were Hypertensive disorder in pregnancy 3(15%), Epilepsy 1 (5%), Hypothyroidism 1(5%) and Previous Caesarean 9(45%) in our study.



Figure 1: Photograph showing congenital cataract



Figure 2: Photograph showing skeletal dysplasia



Figure 3: Photograph showing cleft lip



Figure 4: Photograph showing Anencephaly

DISCUSSION:

Advanced diagnostic technology, especially Ultrasonography (Level 11), has made it possible to detect increased number of birth defects in infants antenatally and during the neonatal period.

In present study, attempts have been made to find out the antenatal incidence of anomalies in our hospital who had ultrasonography done in 2nd and 3rd trimester of pregnancy.

The antenatal incidence of congenital malformation in the present study was 1.72% (17.2 per 1000 births) which is comparable with the observations of Mahela et al¹⁹ 1.73%, Souka et al²⁰ 1.21%, Nakling et al²¹ 1.47%, but was less than Alia et al²² 2-3 %, this may be due to different geographical area, social factor, racial difference, observer variation and equipment quality.

Although McIntosh et al²³ reported in 1954 that only 43% of congenital abnormalities are diagnosed at birth, the present rate of detection may be higher because of current improvements in health care and advances in diagnostic techniques.

Though elderly age group is considered as risk factor for congenital anomaly, in our study younger age group (20-29) showed higher frequency 17(85%), this may be due to earlier age of marriage in our scanning population.

In our study, majority of malformed babies are among multipara 15(75%). Our result is consistent with this finding from other study Jawad S et al²⁴ where the prevalence of malformations among multipara was 76% .

In present study, congenital malformations of the central nervous system were the highest followed by the malformations of gastrointestinal system. Similar findings were observed by Mahela et al¹⁹ Perveen et al²⁵, Agarwal et al²⁶ whereas Sawardekar et al²⁷ have observed gastrointestinal anomalies to be the most common one. The high occurrence of central nervous system anomalies in present study could be explained by a lack of foods fortified with folic acid, very low conventional intake of folic acid, and inadequate dietary intake of foods rich in folic acid, such as vegetables. These folic acid intake problems were probably due to poor appetite and nausea during pregnancy, and poor antenatal care. Better antenatal care might have allowed diagnosis of these anomalies at an earlier stage. Studies have shown that daily maternal intake of folic acid alone or in the form of multivitamin supplements before conception until the first trimester of pregnancy can help in preventing the occurrence and recurrence of neural tube defects.²⁸

Different rates might be attributed to the fact that the present study was a hospital-based case series while the other study was conducted using a population-based surveillance program. Hospital-based system have particular advantages over a population-based system for collection of good quality information, more complete and accurate data and a more readily available set of birth statistics.

Various associated risk factors noted in the present study were having similar spectra of risk factors as seen in other studies conducted in recent time.

The prognosis for anencephalic feti is poor. Most feti are still born and most born alive die within 24 hours. Ensuring folic acid supplement during preconceptional phase can lower the frequency of Neural Tube Defects. Folate is a water-soluble B vitamin that must be obtained in the diet or through supplementation. For >50 yr, it has been known that folate plays an integral role in embryonic development. Patient with no history of intake of folic acid periconceptionally and in the first trimester of pregnancy have higher incidence of congenital information.¹⁸

CONCLUSION:

Antenatal ultrasound is a non invasive highly sensitive, accurate and cost effective imaging technique which gives good results in experienced hands. Meticulous screening for pregnant ladies by ultrasound, especially in 2nd trimester and follow up of anomaly scan is advisable. This detects the structural and the functional abnormalities in the developing fetus including genetic disorders and congenital malformations. Many biochemical markers can be used if any abnormality on ultrasound is detected. Genetic counseling of the patient can be done if Level 11 Scan is done at the proper time and termination of pregnancy can be offered to those who fall under the preview of MTP Act, that is, before 20 weeks of gestation.

In our study, Most of the patients reported late with anomaly scan and could not be offered MTP because of legal restraints, though many of these patients were carrying fetu which were incompatible with life.

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