Section: Obstetrics and Gynaecology

Original Research Article

EXPLORING PRENATAL DIAGNOSIS AND THE INCIDENCE OF CONGENITAL FETAL ANOMALIES IN A TERTIARY CARE HOSPITAL IN NORTHERN INDIA

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Abstract

Background: Congenital anomalies, present at birth, can cause physical or mental disabilities and even be fatal. Globally, eight million children are born with such anomalies yearly, and 3.3 million do not survive past five. Among the survivors, 3.2 million may experience mental and/or physical disabilities. This prospective hospital study, conducted in 2021-2022 at a Northern Indian tertiary care hospital, centered on pregnant women beyond 10 weeks of gestation visiting the Antenatal OPD who underwent NT/NB scans. Patients with abnormal NT/NB scans or congenital malformations were included in the study, which aimed to determine the incidence of fetal congenital anomalies through pregnancy ultrasounds and investigate associated risk factors and fetal outcomes. The study found a 1.43% incidence of fetal congenital malformations. Among the patients, 22.8% underwent first-trimester medical termination of pregnancy, while 76% had abnormal Level 2 scans at 18-22 weeks. At birth, 1.14% of babies were found to have congenital malformations. Early detection of major anomalies offers parents the choice of a safer and less emotionally distressing pregnancy termination, facilitating improved intervention and management planning. Additionally, it helps reduce unnecessary costs and eases the emotional, physical, and psychological burden on families associated with carrying a child with disabilities.

INTRODUCTION

Congenital anomalies, or birth defects, are major contributors to disability and infant mortality worldwide, with approximately 303,000 newborn deaths within four weeks of birth annually, affecting 3-6% of infants globally each year.[1] The occurrence and types of congenital anomalies can change over time and across different regions, reflecting complex interactions between genetic, environmental, socio-cultural, socio-economic, racial, ethnic, and epidemiological factors, both known and unknown.[2] Advanced maternal age is linked to abnormal fetal development and chromosomal issues such as Down's syndrome. When congenital anomalies are identified in older mothers, specific tests like microarray, FISH, or karyotyping are necessary to pinpoint the exact cause.[3] Amniocentesis is the most common invasive prenatal diagnostic procedure, with an accuracy rate of approximately 99.4% and minimal false results. However, these invasive methods are primarily used for high-risk cases due to their time-consuming nature and cost-effectiveness. They can also carry a risk of procedure-related abortion in some instances. For low-risk patients, prenatal diagnosis typically involves non-invasive screening using ultrasound and maternal serum biochemistry.[4] With the availability of advanced diagnostic tools like NT/NB Scan and Level 2 Ultrasound, there has been an increase in the detection of congenital anomalies during pregnancy. Early diagnosis allows for necessary interventions and measures to improve the perinatal and long-term outcomes of affected babies. Anomalies compatible

with life require follow-up, while lethal anomalies should be managed in accordance with gestational age and established rules.[5] The actual extent of the impact of congenital anomalies on births in India remains unknown because there is no national surveillance system for birth defects.[6] The study's goal was to examine the incidence, obstetric factors, and prenatal anomaly detection related to severe fetal congenital anomalies. Data is essential because there is currently no information regarding the impact of pregnancies or births affected by congenital anomalies on healthcare utilization, particularly in cases of pregnancy termination due to fetal anomaly detection. It's important to note that while several studies have assessed the prevalence of congenital anomalies in India, most of them focused on recording congenital malformations at birth, rather than studying those diagnosed in utero and subsequently terminated medically.

MATERIALS AND METHODS

This hospital-based prospective study, conducted with institutional approval and informed consent, took place from 2021 to 2022 at a Northern Indian tertiary care hospital. It focused on pregnant women beyond 10 weeks gestation attending the Antenatal OPD who underwent NT/NB scans. Those with abnormal scans or congenital malformations were included, following them to term. Inclusion criteria involved willing singleton pregnant women beyond 10 weeks gestation, while exclusion criteria encompassed those unwilling to undergo sonographic evaluation or with multiple pregnancies. In this study, we enrolled patients in the ANC clinic and conducted detailed medical assessments, including high-risk factors. All patients received NT/NB scans at 11-13 weeks of gestation. Abnormal results prompted double marker testing, with further confirmatory tests like NIPT/CVS if needed. Termination was advised for lethal or non-compatible-with-life anomalies. Patients with minor anomalies continued pregnancy and underwent a Level 2 ultrasound at 18-22 weeks. Those with normal results needed no further tests, but those with minor anomalies received additional marker tests. If required, chromosomal tests like karyotyping, FISH, or microarray were done. Termination was advised for anomalies not compatible with life. Patients who declined termination were monitored until term, and fetal outcomes were recorded.

RESULTS

During the study period, we included 4,890 ANC patients. Among them, 4,820 patients (98.57%) showed no signs of gross fetal congenital anomalies, while 70 patients (1.43%) were identified with fetal congenital anomalies. Therefore, the incidence of congenital anomalies in fetuses in our study was 1.43% (Table 1 and Figure 1).

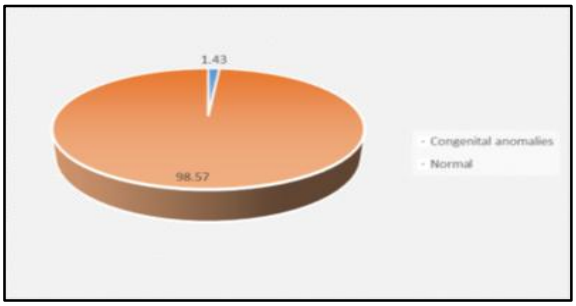

Figure 2: Incidence of fetal congenital anomalies

All patients had NT/NB ultrasounds during the first trimester (11-13 weeks of gestation). Among them, 50% had NT measurements >3 mm with no NB, 31% had NT >3 mm with NB present. Anencephaly was found in 9% of cases, spina bifida in 4%, and other anomalies in 3%. Nine patients with lethal malformations (6 anencephaly, 3 spina bifida) were advised of medical termination of pregnancy (MTP). The remaining 59 out of 70 patients underwent Dual marker tests for further confirmation of fetal congenital anomalies (Table 2 and Figure 2).

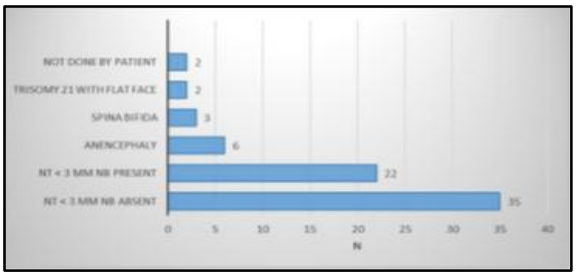

Figure 2: Distribution of patients according to USG (NT/NB SCAN) findings

Out of the 70 patients, the majority 54 (77.14%) chose to continue their pregnancies following the NT/NB scan and first-trimester MTP. They were then recommended to undergo a Level 2 ultrasound at 18-22 weeks of gestation for a comprehensive assessment of structural anomalies. Out of the 54 patients who underwent the Level 2 ultrasound, 76% showed abnormal findings, while 24% had normal results. Those 13 patients with normal Level 2 scans did not require further congenital anomaly testing and proceeded with regular ANC visits and routine investigations until term (Table 3 and Figure 3).

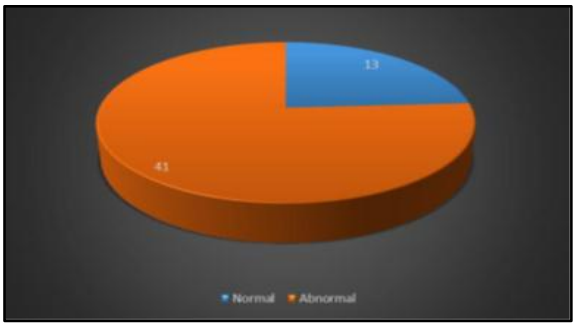

Figure 3: Distribution of the cases according to USG level II

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On USG Level 2, various congenital anomalies were detected, including

- Duodenal atresia and echogenic bowel in 4 patients (10%).
- Polydactyly in 4 cases (10%).
- Cleft lip and cleft palate in 3 patients (7.5%).
- Anencephaly, ASD/VSD with echogenic focus in the left ventricle, cardiac dextrorotation, partial encephalocele, umbilical cord with 2 vessels, ventriculomegaly, and non-immune hydrops fetalis, each occurring in 2 patients (5%).
- Amniotic band, limb atresia, bilateral club foot in the fetus, lung agenesis, bilateral renal agenesis with moderate hydronephrosis, cystic hygroma with ventriculomegaly, Ebstein anomaly of the heart, gastroschisis, hydrocephalus, limb atresia with femoral asymmetry, meningocele with congenital heart disease, omphalocele in the fetus with absent anterior abdominal wall, pulmonary valve atresia and stenosis, skeletal dysplasia, spina bifida, TOF (Tetralogy of Fallot), and transposition of great vessels, each found in 1 patient (2.4%) (Table 4 and Figure 4).

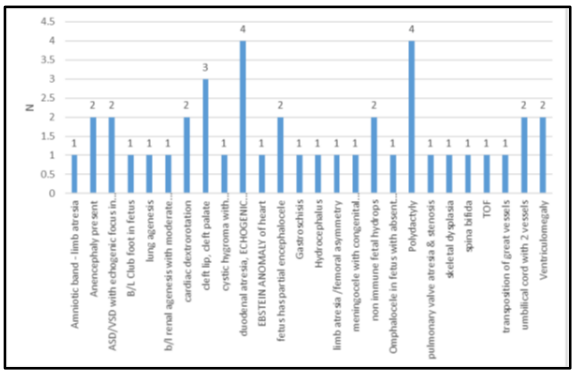

Figure 4: Distribution of cases according to various anomalies on USG (LEVEL 2)

Among the 70 patients with abnormal NT/NB scans, 16 (22.85%) chose to terminate their pregnancies in the first trimester, while 9 patients (12.85%) opted for termination in the second trimester. In total, 25 pregnancies (35.71%) were terminated due to fetal congenital anomalies, while the remaining 45 patients (64.28%) continued their pregnancies to term with regular antenatal care (Table 5 & Figure 5).

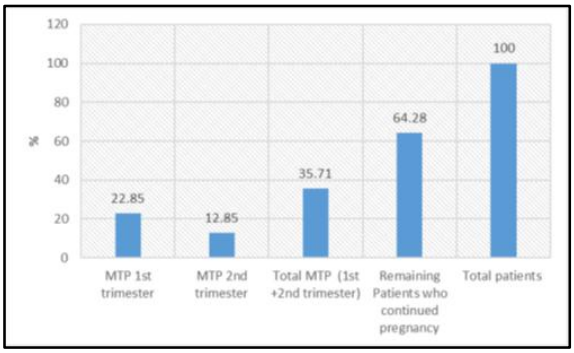

Figure 5: Distribution of the patients according to the outcome of pregnancy.

Out of the total 45 babies born, 41 were live births (91.12%), while 4 were stillborn (8.88%), primarily due to congenital anomalies (Table 6 & Figure 6).

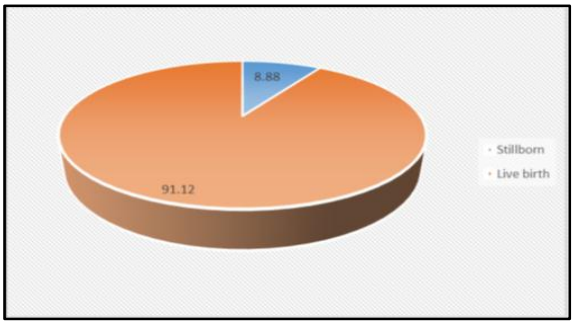

Figure 6: Distribution of the cases according to the fetal outcome

Out of the 41 live births, 33 (80.48%) were healthy babies, while 8 (19.52%) were born with congenital malformations (Table 7 & Figure 7).

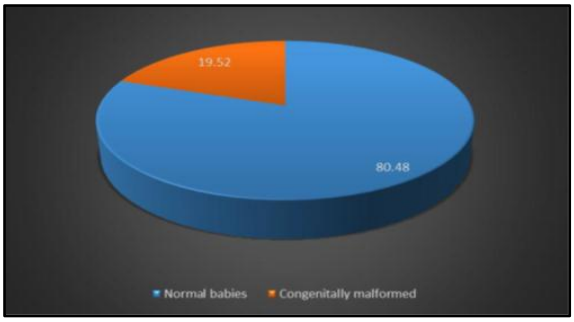

Figure 7: Distribution of cases according to congenital malformation in babies at birth

Table 1: Incidence of fetal congenital anomalies

ANC patients	Number	Percent
Congenital anomalies in the fetus	70	1.43
Normal	4820	98.57
Total	4890	100

Table 2: Distribution of patients according to USG (NT/NB SCAN) findings

USG NT/NB finding	Frequency	Percent
NT >3 mm NB absent	35	50
NT >3 mm NB present	22	31
Anencephaly	6	9
Spina bifida	3	4
Flat face	2	3
Not done by patient	2	3
Total	70	100

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Table 3: Distribution of the cases according to USG level II

USG level 2	Number	Percent
Normal	13	24
Abnormal	41	76
Total	54	100.0

Table 4: Distribution of cases according to various anomalies on USG (LEVEL 2)

USG LEVEL 2 anomalies	Number	Percent
Amniotic band - limb atresia	1	2.4
Anencephaly present	2	5
ASD/VSD with echogenic focus in the left ventricle	2	5
B/L Club foot in fetus	1	2.4
Lung agenesis	1	2.4
B/L renal agenesis with moderate hydronephrosis	1	2.4
Cardiac dextrorotation	2	5
Cleft lip, cleft palate	3	7.5
Cystic hygroma with ventriculomegaly	1	2.4
Duodenal atresia, echogenic bowel	4	10
Ebstein anomaly of heart	1	2.4
Fetus has partial encephalocele	2	5
Gastroschisis	1	2.4
Hydrocephalus	1	2.4
Limb atresia /femoral asymmetry	1	2.4
Meningocele with congenital heart disease	1	2.4
Non-Immune Fetal hydrops	2	5
Omphalocele in fetus with absent anterior abdominal wall	1	2.4
Polydactyly	4	10
Pulmonary valve atresia & stenosis	1	2.4
Skeletal dysplasia	1	2.4
Spina bifida	1	2.4
TOF	1	2.4
Transposition of great vessels	1	2.4
Umbilical cord with 2 vessels	2	5
Ventriculomegaly	2	5
Total	41	100.00

Table 5: Distribution of the patients according to the outcome of pregnancy

Outcome of pregnancy	Number	Percent
1st trimester MTP	16	22.85
2nd trimester MTP	9	12.85
Total MTP (1st +2nd trimester)	25	35.71
Remaining Patients who continued pregnancy	45	64.28
Total patients	70	100

Table 6: Distribution of the cases according to the fetal outcome

Fetal outcome	Number	Percent
Stillborn	4	8.88
Live birth	41	91.12
Total	45	100.0

Table 7: Distribution of cases according to congenital malformation in babies at birth

Condition at birth	Number	Percent
Normal babies	33	80.48
Congenitally malformed	8	19.52
Total	41	100

DISCUSSION

A congenital anomaly is a structural, functional, or metabolic abnormality present at birth, leading to physical or mental disability or even fatality. Globally, eight million children are born with congenital anomalies annually, and sadly, 3.3 million of them do not survive past age five. Among the survivors, 3.2 million may experience mental and/or physical disabilities.[7] While all pregnancies carry a risk of congenital malformations, some are at higher risk. There's a vital need for regular and comprehensive screening for fetal congenital anomalies, with a primary focus on early detection of major anomalies. Early diagnosis empowers parents to make informed decisions regarding the continuation or termination of pregnancy, as well as potential options for fetal or postnatal intervention.[8] In our study, all 4,890 registered antenatal patients underwent NT/NB scans during the first trimester (11-13 weeks of gestation) to screen for fetal congenital anomalies. Among these patients, 70 cases were found to have abnormal USG NT/NB scan reports, resulting in an incidence rate of 1.43%.

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Among the 70 patients in our study, the majority (50%) had NT measurements exceeding 3 mm with absent NB, while 31% had NT measurements exceeding 3 mm with NB present. We observed anencephaly in 9% of patients and spina bifida in 4%. Flat face deformity was noted in 3%, and 2% of patients did not undergo the advised NT/NB scan. Nine patients with lethal congenital anomalies (anencephaly and spina bifida) were recommended first-trimester medical termination of pregnancy (MTP) due to incompatibility with life. The remaining 87.15% (59 out of 70) were advised to undergo the Dual marker Test for further confirmation of fetal congenital anomalies. Castro et al. found that approximately 60-67% of congenital fetal malformations could be identified before the 12th week of pregnancy.[9] Oztekin et al. conducted an analysis of 1,085 pregnancies, where 1.29% were detected in the early (1st trimester screening) and an additional 0.47% in the second trimester screening, resulting in a total incidence of 1.75%. This incidence is similar to our study, which reported an incidence of 1.43%.[10] Out of all antenatal clinic attendees, 18 patients (0.38%) were recommended for first-trimester medical termination of pregnancy (MTP) due to fetal congenital anomalies. However, only 16 patients (0.33%) proceeded with the MTP. The remaining 54 patients opted for a Level 2 ultrasound at 18-22 weeks. Among them, 13 patients (24%) had normal Level 2 scans, while 41 patients (76%) showed abnormalities. The average gestational age for the ultrasound diagnosis via Level 2 was within the 18-22 week range. Although ultrasound is highly sensitive for prenatal congenital anomaly diagnosis, it was not feasible before 20 weeks in 90.6% of cases. The detailed fetal anatomic survey conducted in the Level 2 ultrasound is crucial and should not be overlooked, as it serves as an excellent diagnostic tool for ruling out congenital anomalies. Out of the 41 patients with abnormal Level 2 scans, 10 exhibited major congenital anomalies that were incompatible with life, leading to a recommendation for termination. The remaining 31 were advised to undergo more specific tests, such as the Quadruple marker test or amniocentesis. Based on these test findings, second-trimester medical termination of pregnancy (MTP) was recommended to 15 patients, but only 9 patients chose to proceed with it. Out of the babies born, 33 (80.48%) were healthy, while 8 (19.52%) were born with congenital malformations. In a study by Sitkin et al., they reported a rate of 26% for live births with congenital anomalies.[11] Meanwhile, in a study conducted by Verma et al, out of 431 stillborn babies, 79 were found to have congenital malformations (18.3%), highlighting a higher prevalence compared to live-born babies.[12]

CONCLUSION

Antenatal ultrasound examinations in the first and second trimester are highly valuable for detecting congenital anomalies, including rare ones. Early identification of major anomalies provides parents with the option for safer and less emotionally distressing pregnancy termination, enabling better planning for interventions and management. This approach also minimizes unnecessary expenses and reduces the emotional, physical, and psychological strain on the family associated with carrying a child with disabilities.

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