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Case Series

Anamoly scan: A mandatory screening tool

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ABSTRACT

Introduction: Congenital anomalies can be due to various reasons. Some of these can significantly affect the viability of the fetus, some may impair quality of life subsequent to delivery and some may not have any significant effect. It is necessary to determine presence of congenital anomalies in-utero for better maternal and neonatal outcomes.
Objectives: To determine presence of congenital anomalies in mothers visiting antenatal clinics by conducting a second trimester anomaly scan.
Materials and Methods: The study evaluated 2000 mothers visiting antenatal care in D. Y Patil Medical College and Hospital in Pune. A second trimester anomaly scan was conducted between 18-20 weeks of gestation in all the patients.
Results: The rate of fetal anomalies noted were 0.8% (16/2000 cases). 13 congenital anomalies were incompatible for viability of the fetus and hence were terminated. 3 congenital anomalies were minor in nature and hence continuation of pregnancy was advised. 2 mothers who were unbooked during antenatal period, delivered anomalous babies and were as well included in the evaluation.
Conclusion: A second trimester anomaly scan should be made mandatory to identify any malformation in the growing fetus as we still have an option to terminate the pregnancy and save the family from the future setback. Also, with the advent of fetal medicine we can identify and treat the fetus in utero which might reduce the future complications.

1. Introduction

In 2013 the neonatal mortality rate was 29 per 1000 live births in India amounting to 7,53,000 neonatal deaths.[1] Currently it is 28 per 1000 live births. Amongst the various reasons for neonatal deaths, 9% are due to congenital anomalies.[2] Another study noted that congenital anomalies account for 8–15% of perinatal deaths and 13–16% of neonatal deaths in India.[3] A second trimester ultrasonography provides important information about the number of fetuses, the gestational age, the location of the placenta, fetal anatomy (congenital anomalies) and maternal anatomy.

The current study is conducted to determine the rate of Congenital anomalies in a tertiary care hospital in Pune, Maharashtra in Western India by conducting a second trimester scan between 18 to 20 week of gestation and also determine the utility of second trimester anomaly scan in determining congenital anomalies.

2. Materials and Methods

This was a prospective study conducted after obtaining due ethical clearance from the institutional ethics committee, DY Patil Medical College, Pimpri, Pune, Maharashtra.

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Informed consent was obtained from the patients after explaining to them the objective and need for the study in their native language. 2000 mothers visiting the antenatal care were included in the study.

2.1. Inclusion criteria

1. Pregnant women of any age group.
2. Patients with gestational age between 18-20 weeks of gestation.
3. Subjects providing valid consent for the study.

2.2. Exclusion criteria

1. Subjects unwilling to undergo second trimester anomaly scan.
2. Subjects with congenital anomalies determined prior to the current scan.

3. Results

The rate of fetal anomalies noted was 0.8% (16/2000 cases). 13 congenital anomalies were incompatible for viability of the fetus and hence were terminated. 3 congenital anomalies were minor in nature and hence continuation of pregnancy was advised. 2 mothers who were unbooked during antenatal period, delivered anomalous babies and were as well included in the evaluation.

Following major congenital anomalies were noted in the fetal scans as described below.

Case 1: Congenital cystic adenomatoid malformation with hydrops: This was noted in primigravida where left lung was replaced by multiple cysts.

Case 2: Anencephaly with craniorachischisis (Figure 1): This was noted in a subject who was second gravida. The earlier delivery was uneventful with no adverse neonatal outcomes.

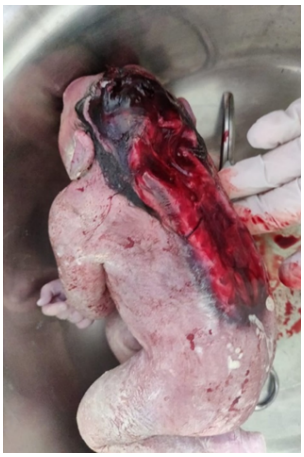

Fig. 1: Anencephaly with craniorachischisis

Case 3: Phocomelia: This was noted in a primigravida where unformed limbs were noted. Both upper and lower limbs were not formed.

Case 4: Proximal urethral obstruction (Figure 2): This was noted in a primigravida. Urinary bladder was noted to be grossly distended and the mother was noted to have gross oligohydramnios. (Figure 2 a) shows the antental USG picture of distended bladder. (Figure 2 b) shows the fetus.

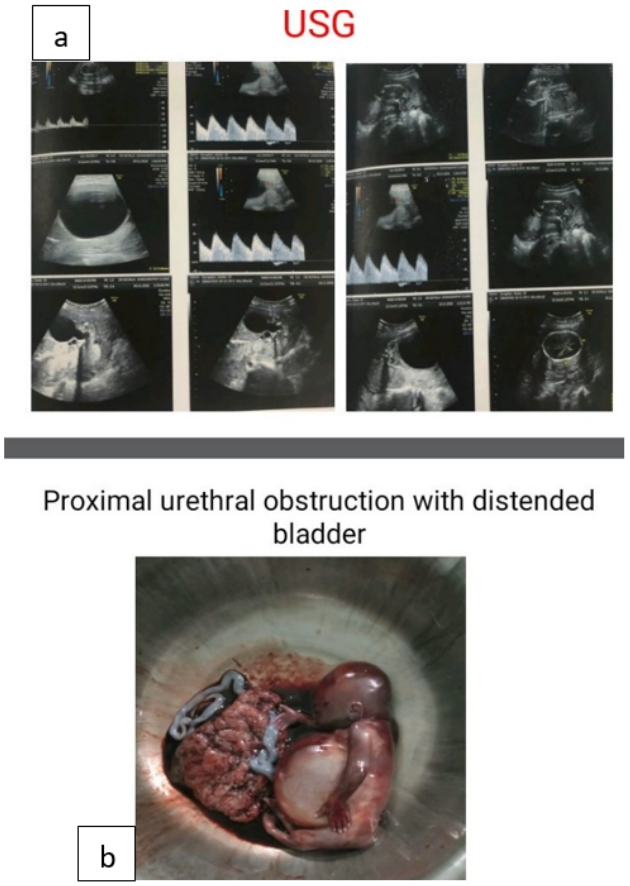

Fig. 2: Proximal urethral obstruction with distended bladder

Case 5: Empty Sella turcica: This was noted in a primigravida patient where there was absence of pituitary gland.

Case 6: Meningomyelocele: This was noted in a primigravida where non closure of spinal canal and the backbone was noted. Non-compliant use of folic acid was observed in mother.

Case 7: Absent stomach bubble: This case was noted in a primigravida mother. The AFI was noted to be 22 cm.

Case 8: Gastroschisis: This was noted in a second gravida patient where abdominal contents were noted to be present outside in the amniotic fluid.

Case 9: Anterior abdominal wall defects: This was noted in a primigravida patient with anterior abdominal wall defects.

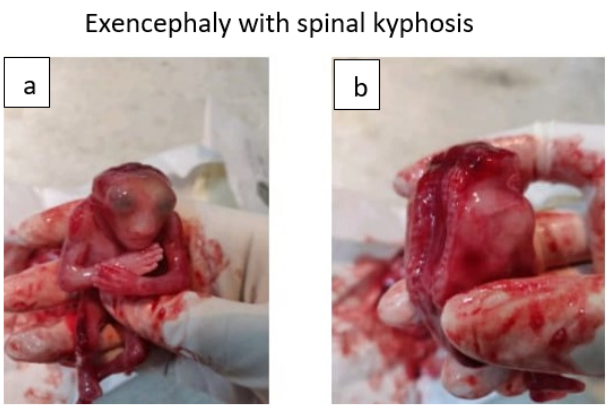

Fig. 3: Exencephaly with spinal kyphosis. Calvarium was absent & brain tissue seen protruding outside. (**a**) Anterior view (**b**) Posterior view

Case 11: Congenital CTEV (Figure 4): There was bilateral CTEV noted in the fetus of a primigravida subject. This was considered as a minor congenital anomaly and the pregnancy was subsequently continued.

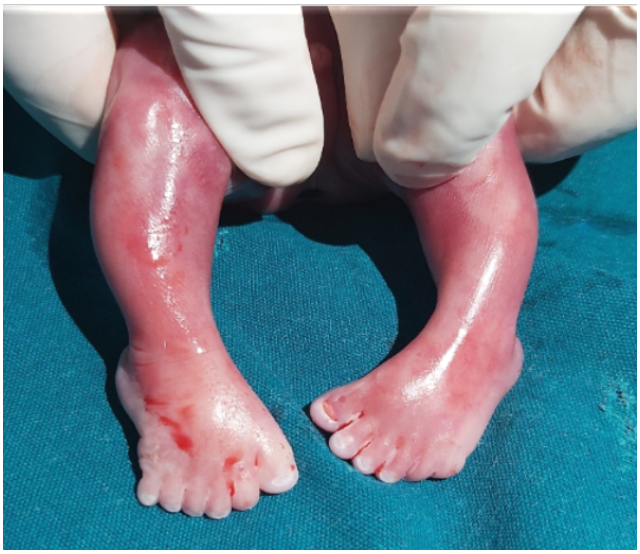

Fig. 4: Congenital CTEV

Case 12: Polydactyly (Figure 5): This was noted in fetus of a primigravida subject with bilateral polydactyly. No other anomalies were noted in the fetus. This was considered as a minor congenital anomaly and the pregnancy was subsequently continued.

Case 13: Cleft lip and palate (Figure 6): This was noted in second gravida patient. The earlier child as well had cleft lip and palate which was surgically corrected. The pregnancy was suggested to be continued till term.

Case 14: Bilateral renal agenesis: Both the kidneys were not formed. This was noted in a primigravida patient.

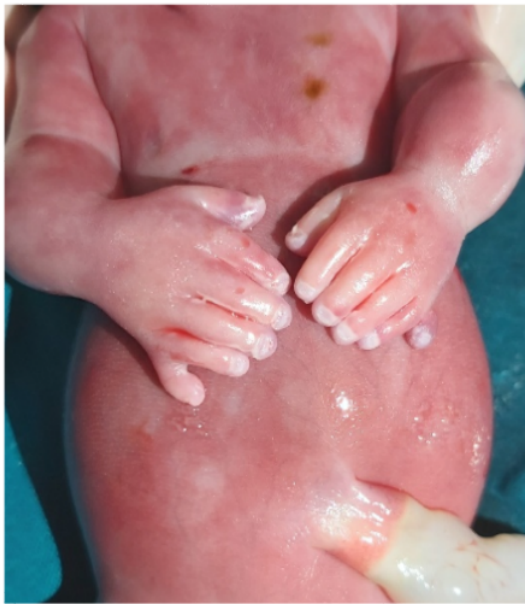

Fig. 5: Polydactyly

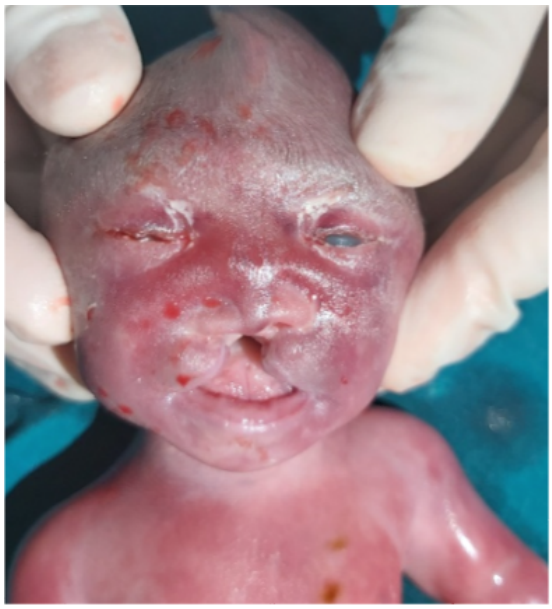

Fig. 6: Cleft lip & palate

Additionally, oligohydramnios was noted.

Case 15: Anencephaly with ventricular septal defect: This was noted in a primigravida patient. The patient had history of exposure to antibiotics for upper respiratory tract infection during early phase of pregnancy.

Case 16: Renal agenesis with incompletely formed urinary bladder: This was noted in a primigravida patient. Patient had history of exposure to pain medication during first trimester. Additionally, severe oligohydramnios was noted.

Table 1: Summary of cases with congenital anomalies

S.No	Congenital anomaly	Pregnancy	Comments/findings
1	Congenital cystic adenomatoid malformation with hydrops	Primigravida	Left lung was replaced by multiple cysts
2	Anencephaly with craniorachischisis	Gravida-2	No adverse outcomes in first delivery
3	Phocomelia	Primigravida	Both upper & lower limbs not formed
4	Proximal urethral obstruction	Primigravida	Bladder was grossly distended +oligohydramnios
5	Empty Sella turcica	Primigravida	Absence of pituitary gland
6	Meningomyelocele	Primigravida	Non-compliant use of folic acid
7	Absent stomach bubble	Primigravida	AFI was 22 cm
8	Gastroschisis	Primigravida	Abdominal contents were noted in amniotic fluid
9	Anterior abdominal wall defects	Primigravida	No other anomalies noted
10	Exencephaly with spinal kyphosis	Primigravida	Brain was present outside the cranium with a kyphosis of spine
11	Congenital CTEV	Primigravida	No other anomalies noted
12	Polydactyly	Primigravida	No other anomalies noted
13	Cleft lip and palate	Gravida-2	Similar history in sibling
14	Bilateral renal agenesis	Primigravida	Oligohydramnios
15	Anencephaly with ventricular septal defect	Primigravida	Exposure to antibiotics for URTI during early phase of pregnancy
16	Renal agenesis with incompletely formed urinary bladder	Primigravida	History of exposure to unknown pain medications in first trimester + oligohydramnios

4. Discussion

The rate of congenital anomalies was noted to be 0.8% in our study. This corresponds with different studies which noted the congenital anomaly rate to be 1.90% in a study by Taksande A et al.[4] and 1.24% by Datta V et al.[5]

Majority of the mothers whose children had congenital anomalies in our study were primigravida 76.92% (10/13) rest 23.08% (3/13) were second gravidas. In a study by Chaturvedi P et al.[6] an increase in frequency was seen in advanced maternal age and in primigravida and fourth gravida mothers.

Only one patient of these 13 had given a positive family history of congenital anomalies where cleft lip and palate was noted in child born in previous pregnancy. Figueiredo JC et al.[7] noted that a family history of clefts as well as having other biological children with a cleft were highly associated with increased risk of developing cleft lip and cleft palate in the current pregnancy.

The major professional societies throughout the world recommend that all pregnant women be offered a mid-trimester ultrasound scan for the detection of structural fetal anomalies.[8–10] This is generally performed between 18 and 22 weeks, though 'at risk' women may receive additional scans. The majority of identifiable anomalies will be detected with 2D ultrasound, but in certain cases (such as facial clefts or talipes equinovarus) three-dimensional (3D) ultrasound may provide additional details.[11]

5. Limitations of the Study

1. The study did not follow up all the patients until delivery of the fetus.
2. The data set is inadequate to extrapolate the data to regional and national trends.
3. Risk factors contributing to anomalies were not evaluated.
4. Chromosomal testing/ DNA analysis was not done.
5. Maternal characteristics contributing to development of congenital anomalies were not studied.

6. Conclusions

Second trimester anomaly scan is an important modality for diagnosis of congenital anomalies. This should be considered as an essential noninvasive procedure in providing ideal antenatal care. Detecting anomalies at an appropriate time before birth can prevent a huge amount of mental, social and physical turmoil to the couple and save the family from the future setback. Also, with the advent of fetal medicine we can identify, treat the fetus in utero which might reduce the future complications & further decrease the rate of neonatal as well as infant mortality rate. Though many congenital anomalies can be detected in first trimester scan, second trimester scan forms the corner stone in diagnosis of anatomical abnormalities in the fetuses. We suggest to conduct similar studies in correlation with first trimester scans and chromosomal/DNA analysis in a greater data set for further conclusive analysis.

7. Source of Funding

None.

8. Conflict of Interest

None.

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