Congenital anomalies in antenatal ultrasound scan at a tertiary care teaching hospital

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ABSTRACT

Introductions: Early detection of congenital fetal anomalies by ultrasonography (USG) helps to identify the severity and termination of pregnancy in severe cases. Anomalies cause significant perinatal morbidity and mortality. Aim of this study was to analyse real-time USG detection of prenatal anomalies.

Methods: This was a cross-sectional descriptive study of fetal anomalies detected during 2nd and 3rd trimester antenatal USG scan done from 2011 to 2015, in Patan Hospital. Age of mother, parity, gestational age when anomalies detected and, anomalies of previous child, use of folic acid were analysed. Descriptive analysis was done.

Results: There were 35,571 deliveries in four years period with 134 (0.37%) congenital anomalies, 99 (79.2%) detected during antenatal USG during 2nd and 3rd trimester. Among 99 anomalies, 29 (29.29%) were detected before 22 weeks. Central nervous system (CNS) anomalies were 47, gastrointestinal 23, renal 12, skin and fetal compartment 8 and rest were less common. Young mothers of less than 30 years were 109 (81.34%) and primi were 85 (63.4%).

Conclusions: Antenatal ultrasound can detect fetal anomalies in 2nd and 3rd trimester with higher frequencies belonging to the central nervous system followed by gastrointestinal tract anomalies and renal.

Keywords: congenital anomalies, gestational age, prenatal ultrasound scan
INTRODUCTIONS

Early detection of congenital fetal anomalies by ultrasonography (USG) helps to identify the severity of disease, its outcome and mode of delivery. It also helps in need for early termination of pregnancy. Second trimester USG between 18 and 22 weeks is the standard for fetal anatomical assessment. Fetal anomalies that are undetectable in first trimester, are detectable in second trimester. Congenital anomalies occur in 3 to 5 % of all pregnancies and 2 to 3 % of all births. They are important cause of perinatal morbidity and mortality in up to 20 to 30% of perinatal death. Aim of this study was USG detection of prenatal anomalies during second and third trimester to improve antenatal care.

METHODS

This was a cross-sectional descriptive study of fetal anomalies detected during antenatal USG scan from second to third trimester, from April 2011 to March 2015 (4 years) in Patan Hospital, Patan Academy of Health Sciences, Lalitpur, Nepal. Hospital numbers of patients with anomalies detected in ultrasound record were noted. These files were traced from the medical record section. Age of mother, parity, gestational age when anomalies detected, past history of anomalies, use of folic acid and the details of anomalies were analysed. First trimester ultrasounds were excluded.

Ethical approval was taken from the Institutional Review Committee (IRC) of PAHS. Data entry and descriptive analysis was done using SPSS software.

RESULTS

There were 35,571 deliveries including termination of pregnancy in 2nd and 3rd trimester. Among them, 134(0.37%) cases of congenital anomalies occurred: 99 (73.88%) were detected in antenatal USG, table 2. The gestational age when congenital anomalies detected was 28.54 +/- 7.25 weeks, half of them after 30 weeks with two peaks at 32 and 33 weeks.

Central nervous system had the highest common anomalies detected 40 (32%) followed by gastrointestinal anomalies 33 (26.4%). Among 26 cases that were not detected, 13 cases were clubfoot, 11 cleft lips, one knee not flexed, one white hair and nine had incomplete information, Table 1 and 2.

Among 99 cases with anomalies, 29 (29.3%) were detected before 22 weeks and 70 (70.7%) after 22 weeks. Half of central nervous system (CNS) anomalies were detected before 22 weeks whereas gastrointestinal (GIT) renal and musculoskeletal (MSK) after 22 weeks. Cystic hygroma, congenital diaphragmatic hernia and multicystic dysplastic kidneys were detected before 22 weeks.

Among 134 cases, 109 (81.34%) were in mothers of less than 30 years of age and 21(18.66%) more than 30; primi were 85 (63.4%) and multiparous49 (36.5%). Among 49 multiparous, 3 cases had previous abnormal child (one each of CNS, GIT and MKS). Sex determination done in 73anomalies had33 (45.2%) male,39 (53.4%) female and 1(1.3%) ambiguous genitalia.

DISCUSSIONS

In our study, congenital anomalies occurred in 134 cases out of 35,571 (0.35%) total birth, less than 2-7% of other studies. In a study of 10 months duration from our institute reported it at 0.81%.

In our study, CNS were more common, with higher frequencies of anencephaly followed by hydrocephalus and meningomyelocele. Folic acid is given once a couple plan baby and through the first trimester of pregnancy to reduce such central nervous system anomalies. Yet, all the cases in our study had taken folic acid during pregnancy.
In our study 57.5% of anomalies were detected after 22 weeks, similar to the report by N Kashyap with 52.1% detected after 20 weeks.³ We had total 15 anencephalies, 11 detected after 22 weeks; however, they had no USG done earlier. The USG should be done

Table 1. Congenital Anomalies during antenatal (n=134)

| Anomalies                        | Frequency | Detected | Not detected | Percent |
|----------------------------------|-----------|----------|--------------|---------|
| Anencephaly¹                     | 15        | 15       |              | 11.2    |
| Cleft Lip²                       | 13        | 2        | 11           | 9.7     |
| Clubbed Foot⁶                    | 13        | 0        | 13           | 9.7     |
| Hydrocephalus¹                   | 11        | 11       |              | 8.2     |
| Meningomyelocele³                | 10        | 10       |              | 7.5     |
| Hydronephrosis³                  | 8         | 8        |              | 6.0     |
| Hydrops Fetus⁴                   | 8         | 8        |              | 6.0     |
| Esophageal Atresia²              | 7         | 7        |              | 5.2     |
| Cystic Hyroma⁸                   | 6         | 6        |              | 4.5     |
| Omphalocele²                    | 5         | 5        |              | 3.7     |
| Gastrochisis²                   | 4         | 4        |              | 3.0     |
| Multicystic Dysplastic Kidney³   | 3         | 3        |              | 2.2     |
| Congenital Pulmonary Airway Malformation⁵ | 2       | 2        |              | 1.5     |
| Ascitis²                        | 2         | 2        |              | 1.5     |
| Thanatophoric Dysplasia⁶         | 2         | 2        |              | 1.5     |
| Holoprosencephaly¹               | 2         | 2        |              | 1.5     |
| Dilated Bowel²                   | 2         | 2        |              | 1.5     |
| Encephalomenigocele¹             | 1         | 1        |              | 0.7     |
| Skeletal Dysplasia⁶              | 1         | 1        |              | 0.7     |
| Polycystic Kidney¹               | 1         | 1        |              | 0.7     |
| Cardiomegaly⁷                    | 1         | 1        |              | 0.7     |
| Dandy-Walker Syndrome¹           | 1         | 1        |              | 0.7     |
| Congenital Diaphragmatic Hernia³ | 1         | 1        |              | 0.7     |
| Sequestration of Lung³           | 1         | 1        |              | 0.7     |
| Dilated Urinary Bladder³         | 1         | 1        |              | 0.7     |
| Cyst Neck⁴                      | 1         | 1        |              | 0.7     |
| Knee not Flexed⁶                 | 1         | 1        |              | 0.7     |
| White Hair⁴                     | 1         | 1        |              | 0.7     |
| Leg Absent⁴                     | 1         | 1        |              | 0.7     |
| Sub-Total                       | 125       |          |              | 93.3    |
| Missing System (uncategorised)  | 9         |          |              | 6.7     |
| Total                           | 134       | 99       | 26           | 100.0   |

Note: Systems involved: ¹Central Nervous, ²Gastrointestinal, ³Renal, ⁴Skin and Fetal Compartment, ⁵Respiratory, ⁶Musculoskeletal, ⁷Cardiovascular, ⁸Lymphatic

Table 2. System involved in congenital anomalies

| System Involved                  | Total |
|----------------------------------|-------|
| Central Nervous System           | 40    |
| Gastrointestinal Tract           | 33    |
| Musculoskeletal                  | 18    |
| Genitourinary                    | 13    |
| Skin and Fetal Compartment       | 10    |
| Lymphatic                        | 6     |
| Respiratory                      | 4     |
| Cardiovascular                   | 1     |
| Missing System (Uncategorised)   | 9     |
| Total                            | 134   |
as early as 11 weeks to detect the anencephaly so that the pregnancy can be terminated earlier within the legal framework of the country.\(^7\)

Facial defect like cleft lip has low prediction in our study only 2 (15.3 \%) of 13 cases were detected on USG, comparable to the study done by Clementi M et al.\(^8\) However, recent study shows, it can be detected in up to 65\%.\(^9\)

We had only one case of heart anomaly, the cardiomegaly. We could not do detail scan of heart due to lack of fetal echocardiography service. Study done by Shama Munin et al. detected only quarter of cardiac defects by ultrasound scans.\(^10\) The low detection of cardiac anomalies in our study could be because we included four chamber view of USG scan; similar to the study by Nayab Ali, et al where none of cardiac defects were diagnosed on the antenatal scan.\(^11\) On the other hand, neuro scan sensitivity has been reported in 14-45\%,\(^12\) higher than ours.

In our study clubfoot were not detected by ultrasound in antenatal scan as all cases were isolated club foot and not associated with other deformities whereas Roselli P, et al. had simple clubfoot diagnosed postnatally; and complex clubfoot associated with other deformity were diagnosed during prenatal ultrasound.\(^13\)

The incidence of congenital anomalies in our study is more in primi (56\%), and younger <30 years (81.3\%), which is different from the study done in India reporting higher incidence in multiparous and older age.\(^14\)

Limitation of our study was twin and multiple pregnancies not included, as well as chromosomal anomalies. Also, the USG scans were done by different operators on different machines, which may give diverse results. Consanguinity of marriage was not recorded in the file, which could be due to reluctant from patient or they may not have specifically asked for it.

CONCLUSIONS

Antenatal ultrasound can detect fetal anomalies in 2\(^{nd}\) and 3\(^{rd}\) trimester. Central nervous system anomalies were highest followed by gastrointestinal tract. Anomalies like clubfoot, cleft lip or skeletal dysplasia were frequently missed.

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