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HOSPITAL-BASED STUDY OF CONGENITAL FETAL ANOMALIES IN GRAVID PATIENTS REFERRED FOR ANTENATAL ULTRASOUND STUDY.

Radiodiagnosis

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ABSTRACT

Antenatal ultrasound screening is used to assess fetal congenital anomalies and syndromes, especially during first trimester and early mid-trimester; later triaged for dedicated anomaly scan based on screening test results including soft and major chromosomal markers or any structural defects found on screening. The aim of our present record based study is to assess burden of congenital fetal anomalies in gravid females referred for antenatal ultrasound examination in our hospital which is a tertiary care centre in hilly region of Garhwal of North Indian state of Uttarakhand.

Materials and methodology: This descriptive retrospective study conducted between March 2016 to December 2016 was assessed from records of antenatal ultrasound studies, with descriptive analysis using tests of significance and proportions wherever necessary; using statistical significance of p-values < 0.05.

Results: Total 767 antenatal ultrasound patients referred from Obstetrics and Gynecology department were assessed this period in which 16 positive cases of congenital fetal anomalies including syndromes were found, suggesting a prevalence of 2.1% of fetal anomalies in hilly Garhwal population. Most common system involved was Urinary system (6 cases [37.5 %]), central nervous system (2 cases[12.5 %]), GI Tract (3 cases[18.75 %]), feto-fetal transfusion syndrome(2 cases[12.5 %]) and rest cases with miscellaneous conditions.

Conclusion: Antenatal ultrasound study of gravid patients was used in our study to assess burden of fetal anomalies and predict the antenatal, intra-natal and postnatal outcome of such pregnancies.

KEYWORDS

Antenatal Ultrasound, Fetal Anomalies, Gravid Patients.

Introduction:

Antenatal ultrasound screening and dedicated anomaly scan is used to assess fetal congenital anomalies and syndromes, especially during first trimester and early mid-trimester; based on screening test results including soft and major chromosomal markers or any structural defects found on ultrasound study[1,2,3,4,7,8]. The aim of our present record based study is to assess burden of congenital fetal anomalies in gravid females referred for antenatal ultrasound examination in our hospital which is a tertiary care centre in hilly region of Garhwal of North Indian state of Uttarakhand. The fetal anomalies are increasingly detected with increasing use of advanced ultrasound studies aided by color Doppler, fetal MRI, maternal serum biochemistry, amniocentesis, chorionic villus sampling, karyotyping and other tests[1, 2, 3, 4, 7]. A dedicated anomaly scan[3] is usually performed in 18- 22 weeks gestational weeks of a gravid patient, after a screening ultrasound performed in all gravid patients who undergo antenatal checkup in hospital at least four times on an average during gestational period of nine months. Based on findings of screening ultrasound like soft and major sonographic markers and evidence of structural defects in fetus or embryo, further assessment is done which predicts the future course and management of the gravid condition depending upon the severity of the anomaly found, results of maternal serum biochemistry and other test results[1,2,3,4,7].

Materials and methodology:

This descriptive retrospective study conducted from March 2016 to December 2016; was assessed from medical records of antenatal ultrasound studies, patient data at the time of diagnosis were retrieved on a predesigned proforma, which concerned variables at time of registration: age, sex, clinical features, and indications for ultrasound study and referral department. Total 767 antenatal ultrasound patients of age group between 19 and 40 years referred from Obstetrics and Gynecology department were assessed on Toshiba Nemio Ultrasound color Doppler machine using 3.5 Megahertz convex transducer/ probe. No patient preparation is required except for mothers in early first trimester who need to drink water sufficient to have a full bladder. Coupling gel was applied on maternal abdomen and systematic B-scan assessment of fetus, uterus, placenta, cervix, liquor and biophysical profile was assessed and fetal heart rate was assessed by M-mode. Fetal evaluation of head, spine, four chamber heart view, pulmonary system, outflow tracts of heart, anterior abdominal wall and umbilical cord, fundic bubble, situs assessment, urinary tract, extremities with measurements of BPD, FL, AC, HC with EFW (fetal weight estimation) is done. Data was entered on a Microsoft excel spreadsheet and descriptive statistical analysis was done using proportions wherever necessary.Tests of significance were used statistical significance of p-values < 0.05.

Results:

Total 16 positive cases of congenital fetal anomalies including syndromes were found, suggesting a prevalence of 2.1% of fetal anomalies in hilly Garhwal population. Overall prevalence of congenital fetal anomalies as detected on prenatal sonography in the world as per various studies ranges from 2 to 4%. Since the incidence of anomalies that too structurally visible i.e. on sonography is quite low, so the case study figures low in our study done over a period of 10 months almost; to about 16 cases. Few (upto 10) embryonic demises could not be included in our study as structurally the dead embryo was normal though cardiac activity could not be seen and no investigations were performed in those cases, and they did not turn up for follow-up serum biochemistry investigations. Mean age of presentation in our study was 29.5 years and mean gestational age in weeks of patients coming for ultrasound in our study was 29.43 weeks. Ideally the mean should have been less than 24 weeks for early prenatal detection of anomalies.

Most common system involved was Urinary system (6 cases [37.5 %]), central nervous system (2 cases [12.5 %]), GI Tract (3 cases [18.75 %]), feto-fetal transfusion syndrome (2 cases [12.5 %]) and rest cases with miscellaneous conditions.[**Table 1 and 4]** Most serious anomalies (3 cases) incompatible with life like Anencephaly (Figure 1), spinal anomaly, and dead stuck twin in feto-fetal transfusion syndrome (Figure 3 and 4) were advised medical termination of pregnancy. Six cases (37.5%) of anomalies were detected in second trimester, whereas 10 cases (62.5%) were detected in third trimester. Only one case of esophageal atresia turned up for follow-up ultrasound as she had severe polyhydramnios and related discomfort in which the esophageal blind pouch in neck of fetus (anterior to cervical spine) was well visualized. Co-relating findings with liquor status; we detected polyhydramnios in three cases (Anencephaly, esophageal atresia and small bowel atresia), oligohydramnios was detected in about five cases (1 case of congenital posterior valves (Figure 2) , 2 cases of stuck donor twin in twin-twin transfusion syndrome, one case of bilateral renal pelvic dilatation (RPD) with lateral ventricular dilatation and one case of spinal anomaly with PV leaking of liquor). Rest of the eight (50%) cases had normal liquor as studied by measuring Amniotic fluid indices (AFI). **[Table 2**]. District-wise distribution [**Table 3]** showed maximum cases coming from Rudraprayag district (6 cases [37.5%]), and 5 cases (31.25%) from Tehri Garhwal district, rest were from Pauri (3 cases) and Chamoli district (2 cases). It may be noted here that these two districts (Rudraprayag and Tehri) are remotely located and maybe due to non-availability of tertiary care facilities there as well as poor socioeconomic status, lack of medical facilities, poor road connectivity, frequent antenatal checkups might not have been possible and folic acid supplementation was neglected by patient as Anencephalic fetus was referred from Rudraprayag district.

a) District-wise table showing the percentages of cases having anomalies:

District	No of cases(n=16)
Rudraprayag	6(37.5%)
Tehri Garhwal	5(31.25%)
Pauri Garhwal	3(18.75%)
Chamoli	2(12.5%)

b) Percentage prevalence of system-wise anomalies:

*Overall prevalence of fetal anomalies in our study is 2.1% (16/767 cases):

Sr No	System of fetus involved	No of cases	Overall prevalence(n/767)
1	Urinary Tract	6	0.78 %
2	GI tract	3	0.39 %
3	CNS anomalies	3	0.39 %
4	Feto-fetal transfusion syndrome	2	0.26 %
5	Miscellaneous conditions	2	0.26%

Discussion:

Basic aim of antenatal ultrasound is to assess 1) intra- or extra-uterine gestation or implantation of embryo 2) cardiac activity 3) status of embryo or fetus 4) amniotic fluid 5) any structural fetal defect visible 6) placental assessment 7) fetal lie and presentation 8) cervical os status[1, 2, 3, 4, 7, 8]. Congenital malformations contribute to increasing perinatal morbidity and morbidity as well as disease burden in new born children globally with a varying prevalence of 2- 4 % in general population[1,2,3,4]. Early detection of these congenital anomalies through antenatal ultrasound evaluation, fetal echocardiography, maternal serum biochemistry (triple serum tests or quad screening tests [hCG, estriol, inhibin, alpha-fetoprotein]) is vital to reduce this impact of disease burden with financial, emotional and social repercussions on general population[1, 2, 3, 4, 7]. It aids in counseling parents of affected fetuses and helps in planning early intervention like cautious monitoring in fetus having soft chromosomal markers, or terminate in cases of fetuses incompatible with life due to incurable anomalous condition like anencephaly or even wait till delivery and achieve postnatal surgical correction in a case like posterior urethral valves (PU valves), esophageal atresia, trachea-esophageal fistula or hydrocephalus (aqueductal stenosis)[3, 7]. Such decision depends on the severity assessment of anomaly by sonographer and needs multidisciplinary approach as the case may be and whenever such an emergent situation to intervene by inducing labor or perform Caesarean section arises[1,4]. Needless to say that major birth defects are a major cause of perinatal morbidity and mortality[2].

It is to be noted at the outset that there is no history of previous fetal anomalies or any major risk factor in mother (like alcohol, teratogen exposure, TORCH infection, drug abuse, diabetes mellitus, etc) in the majority of cases detected to have anomaly on ultrasound scan for first time in that pregnancy[1, 2, 3]. Also the trend in third world countries like India, the patients come late for antenatal checkup in late third trimesters due to obvious educational factors, low socio-economic status, lack of tertiary care facilities in remote areas, poor connectivity of roads[2].

Antenatal ultrasound is done during:

1) First trimester, which is period of organogenesis, from fourth through 14 weeks of gestation[2, 3, 7]: embryonic period being 4th to 10th week of gestation, wherein early pregnancy failure, increased nuchal translucency (3 mm or greater) can be detected. Added use of fetal echocardiography in 11-14 weeks gestational period for assessment of ductus venosus flow abnormality like absent or reversed a –wave in waveform, tricuspid regurgitation lead to diagnosis of fetal aneuploidy and cardiac defects[3, 4]. Increased nuchal translucency is seen in aneuplodies, cystic hygromas, congenital rubella syndrome, arthrogryposis multiplex congenita, lethal multiple pterygium syndrome, and many such fetal anomalies[3,4,5,6,7,8].

2) Early mid-trimester ultrasound (18 to 24 weeks) [1, 3, 7]: It is recommended by various health organizations like American college of Obstetricians and Gynecologists, Royal college of Obstetrics and Gynecologists, Canadian Society, as well as National Institute of Child Health and Human development that at least one ultrasound should be offered to all pregnant women between 18 and 20 weeks irrespective of any risk factors, so as to allow for optimal early fetal evaluation for anatomy, chorionicity, gestational age and dating, abnormal placentation and mainly to assess the soft anatomic sonographic markers for aneuploidies like increased nuchal fold (6 mm or greater), choroid plexus cysts, echogenic cardiac focus, echogenic bowel and absent or hypoplastic nasal bone (most sensitive marker for Down's syndrome) [2,3,4]. Mild renal pelvic dilatation (RPD) or pyelectasia, short or long bones, are also soft markers which need assessment by maternal serum biochemistry and later by chorionic villus sampling, amniocentesis, umbilical cord blood sampling for cell free DNA markers/ karyotyping[1, 3, 7]. Echogenic bowel is usually seen in TORCH group of infections, IUGR, intra-amniotic bleeding, cystic fibrosis, GI obstruction[3, 7]. Maternal pelvic assessment including status of cervix and internal os can be assessed[3].

Chorionicity should be assessed as structural anomalies, deformities due to fetal crowding, are quite common in multi-fetal gestations with chances of twin-twin transfusion syndromes, wherein every two-weekly follow-up ultrasounds are needed to detect these abnormalities at the earliest by 16 weeks[3,7]. Identifying such cases in twins can help to plan therapeutic fetal interventions or preventive interventions in utero like release of amniotic bands, laser fetoscopy to produce amniotic septostomy in fetal transfusion syndromes to save donor fetus which has oligohydramnios and can then grow adequately[3, 7]. Ultrasound assessment of anomalies like in meningomyelocele, encephalocele, gastroschisis, help in determining path for birth of such anomalous babies wherein Caesarean section may be planned for to prevent fatal complications in such cases.

Additional assessment by :- a) Fetal MRI at 20-22 weeks and later third trimester fetal MRI helps to resolve conflicting inconclusive cases and plan delivery, intervention if needed like intrauterine procedures [3, 7]b) Fetal echocardiography in early mid-trimester [3, 4]3) Transvaginal ultrasound (TVS) especially in first trimester aid in detection of fetal anomalies; these studies are performed when maternal transabdominal ultrasound is in-conclusive for many maternal and fetal causes like oligohydramnios, unfavorable fetal lie and position, maternal obesity, fetal osseous shadowing artifacts, which preclude optimal fetal evaluation even in hands of a skilled operator[3,7].

All anomalies detected on ultrasound in our study were classified as major or minor based on severity of anomaly, like malformations which are lethal, incompatible with life, incurable, and those associated with severe handicap or need surgical correction are classified as major. Rest can be termed as minor. In our study, almost four cases (25 %) were classified as minor (pelvi-ureteric junction [PUJ] obstruction [1 case], bilateral renal pelvic dilatation (RPD) [1 case] and prominent fundic bubble [1 case]). Rest major subset was 12(75%) cases with major anomalies which needed either termination of pregnancy (anencephaly and spinal anomaly with hyperflexion) or postnatal surgical correction (like esophageal atresia, PU valves, small bowel atresia, trachea-esophageal fistula [TOF]). Cases like with anencephaly, spinal anomaly with hyperflexion, were advised for medical termination of pregnancy which they underwent. Cases with increased nuchal fold and echogenic cardiac focus with echogenic fetal bowel suspected to be having aneuploidy especially Down's syndrome were asked to undergo triple serum tests but they did not turn up for follow-up. Rest of surgically correctable defects like PU valves, PUJ obstruction, esophageal atresia and small bowel atresia, TOF, hydrocephalus were counseled for post-natal surgical correction. Minor abnormalities like renal pyelectasia and unilateral multicystic dyspladstic kidney were also counseled for postnatal ultrasound, out of which pyelectasia turned up for postnatal checkup had mild hydronephrosis due to partial PUJ obstruction.

Conclusion:

Identification of fetal anomalies antenatally helps in preventing perinatal morbidity, mortality. It gives us an opportunity to explain mother's legal right to voluntary abortion of an anomalous fetus, allows to carry out termination in incurable cases, allows intervention in utero, plan delivery and birth path of anomalous baby preventing complications and improving prognosis of fetus, and most important it allows for planning a multidisciplinary approach during delivery or LSCS by pediatrician, anesthetists, pediatric surgeon, neonatologists as and when deemed necessary. Understanding burden of surgical congenital anomalies helps in planning health programs and public outreach programmers like prevention of nervous system anomalies by folate supplementation, educating public for at least four antenatal health checkups with ultrasound during each visit with proper nutritional supplementation.

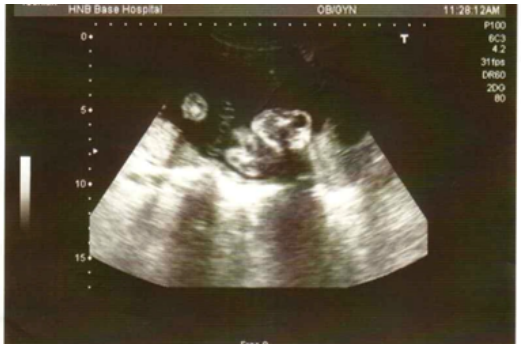

Figure 1- Sonography imxage showing anencephalic fetal head with polyhydramnios

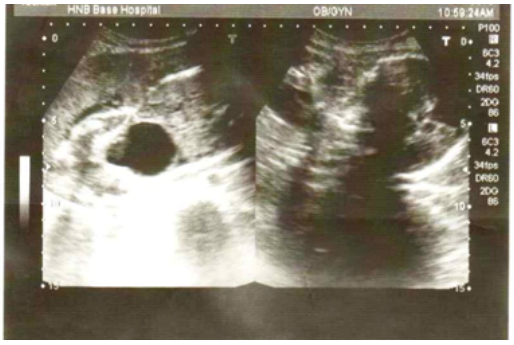

Figure 2- Sonography image showing PU valves with key-hole sign of an obstructed fetal urinary bladder.

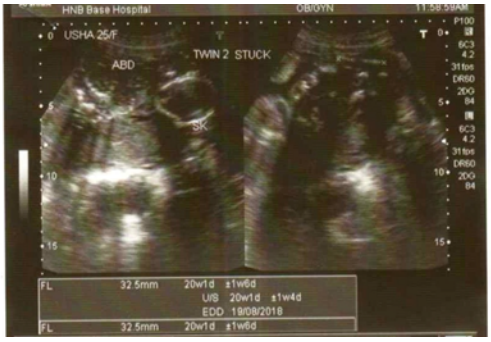

Figure 3- Sonography image showing stuck donor twin with oligo-hydramniotioc sac.

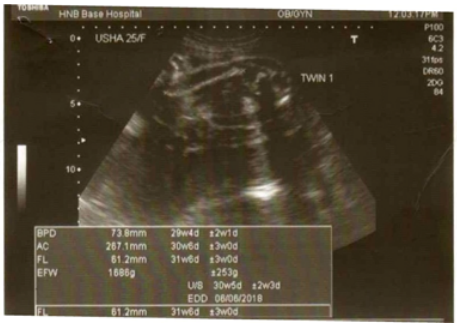

Figure 4- Recipient twin (large sized) in feto-fetal transfusion syndrome- the other fetus twin of donor small sized fetus in figure 3.

Table 1: Composite table with diagnosis (variables) showing GA in weeks and age of presentation

Sr No	Age	GA wks	Diagnosis	AFI
1	29	32	Lt PUJ obstruction	15
2	19	36	PU Valves	9
3	25	19	Echogenic fetal bowel and echogenic cardiac focus	Normal
4	32	15	Increased Nuchal fold thickness	Normal
5	30	33	Esophageal atresia	24
6	27	30	Anencephaly	28
7	23	34	B/L RPD with prominent fundic bubble	14
8	27	28	PU Valves with oligohydramnios	2
9	23	29	Rt Multicystic dysplastic kidney	14
10	27	20	Spinal anomaly with hyperflexion	2
11	23	35	Prominent fundic bubble	9
12	40	36	Small bowel atresia	20
13	22	37	Esophageal atresia with T-O Fistula	20
14	25	28	Hydrocephalus with B/L RPD	4
15	25	29	Twin pregnancy with stuck live donor twin	2
16	25	30	Twin pregnancy with stuck dead twin	2

Table 2: Diagnosis with liquor status as ascertained by AFI shown in Table 1.

Sr no	GA in wks	Diagnosis	Liquor
1	32	Lt PUJ obstruction	Normal
2	36	PU Valves	Normal
3	19	Echogenic fetal bowel and echogenic cardiac focus	Normal
4	15	Increased Nuchal fold thickness	Normal
5	33	Esophageal atresia	Increased
6	30	Anencephaly	Increased
7	34	B/L RPD with prominent fundic bubble	Normal
8	28	PU Valves with oligohydramnios	Reduced
9	29	Rt Multicystic dysplastic kidney	Normal
10	20	Spinal anomaly with hyperflexion	Reduced
11	35	Prominent fundic bubble	Normal
12	36	Small bowel atresia	Increased
13	37	Esophageal atresia with T-O Fistula	Increased
14	28	Hydrocephalus with B/L RPD	Reduced
15	29	Twin pregnancy with stuck live donor twin	Reduced
16	30	Twin pregnancy with stuck dead twin	Reduced

Table 3: District wise distribution of variables (diagnosis)

Sr no	Age	District	Diagnosis
1	29	Pauri	Lt PUJ obstruction
2	19	Tehri Garhwal	PU Valves
3	25	Rudraprayag	Echogenic fetal bowel and echogenic cardiac focus
4	32	Pauri	Increased Nuchal fold thickness
5	30	Pauri	Esophageal atresia
6	27	Rudraprayag	Anencephaly
7	23	Rudraprayag	B/L RPD with prominent fundic bubble
8	27	Tehri Garhwal	PU Valves with oligohydramnios
9	23	Chamoli	Rt Multicystic dysplastic kidney
10	27	Rudraprayag	Spinal anomaly with hyperflexion
11	23	Rudraprayag	Prominent fundic bubble
12	40	Tehri Garhwal	Small bowel atresia
13	22	Tehri Garhwal	Esophageal atresia with T-O Fistula
14	25	Chamoli	Hydrocephalus with B/L RPD
15	25	Tehri Garhwal	Twin pregnancy with stuck live donor twin
16	25	Rudraprayag	Twin pregnancy with stuck dead twin

Table 4: System-wise distribution of cases

Sr No	System of fetus involved	(n=16)No of cases	%
1	Urinary Tract	6	37.5
2	Gastro-intestinal Tract	3	18.75
3	Central nervous system	3	18.75
4	Feto-fetal transfusion syndrome	2	12.5
5	Miscellaneous conditions needing karyotyping	2	12.5

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