ORIGINAL RESEARCH PAPER | Medical Science | Volume : 6 | Issue : 8 | August 2016 | ISSN - 2249-555X | IF : 3.919 | IC Value : 74.50

Role of Ultrasonography in Detection of Major Anatomical Foetal Anomalies in Northern Bihar

KEYWORDS: foetal anomalies, prenatal diagnosis, ultrasonography, antenatal screening

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ABSTRACT *Role of Ultrasonography in detection of Major Anatomical Foetal Anomalies in Northern Bihar.* ***OBJECTIVES:*** *To define the incidence and systemic distribution of major foetal anomalies in northern Bihar.* ***METHODS:*** *Live 10000 singletons of 9 to 41 wks of gestation were scanned and statistically analysed during a period of 05 years.* ***RESULTS:*** *The incidence of major foetal anomalies was found to be 3.04%.The central nervous system was the commonest involved(40.4%),followed by the genitourinary tract (19.4%)and the gastrointestinal tract (10.2%).* ***CONCLUSION:*** *This study recommends the use of a minimum one antenatal ultrasound screening preferably in the late first trimester. Where facilities are available, an early first trimester scan followed by mid – trimester and late third trimester scans can surely avoid many maternal and foetal complications.*

INTRODUCTION:-
The objective of this study was to sonographically detect foetal malformations and to evaluate associated anomalies and outcome of pregnancy.

MATERIAL AND METHODS:-
This study was done in the Department of Radiology JLNMCH, Bhagalpur over a period of 5 years from 01st June 2011 to May 2016. A total of 10,000 cases were studied. The pregnant females referred for routine as well as targeted imaging contributed the population for this study. Only live singleton pregnancies of 09 to 41 weeks gestations were analysed. The amniotic fluid volume was assessed subjectively. Those with vesicular mole and intrauterine demise were excluded. Metabolic diseases and other abnormalities without evident structural variations were also excluded. Those with major anomalies were serially assessed and followed up for confirmation of the diagnosis.

RESULTS: -
The majority of the cases referred for scanning were for confirmation of the gestational age and for the exclusion of associated pathologies and anomalies. A more specific indication was the disproportionate uterine size compared to the period of amenorrhoea. The other common indications were vaginal bleeding and discrepancies of growth. Those mothers with history of previous anomalous foetuses, peritoneal infections or exposure to teratogens formed only a small group.

Polyhydramnios of moderate to severe degree was noted in 200 pregnancies. The Central Nervous System, Genitourinary and Gastrointestinal system were commonly involved in these cases. Pregnancies with severe degree oligohyramnios often ended up with early intrauterine demise. There were 304 anomalous features in the population studied.

There were 123 foetus with CNS involvement (40.4%), 59 with genitourinary involvement (19.4%), 26 with skeletal system involvement, 31 with gastrointestinal involvement (10.2%), 15 with cardiovascular involvement (4.9%) and 50 (16.5%) with less frequent anomalies and overlapping multiple anomalies.

Table: 1 The distribution of CNS anomalies in 123 cases.

Anomalies	Number	% Age
Anencephaly	31	10.2%
Meningomyelocele	14	4.6%
Spina bifida	03	0.98%
Acrania	03	0.98%
Hydrocephalus	36	11.84%
Dandy Walker malformation	03	0.98%
Arnold Chiari malformation	02	0.6%
Corpus callosal agenesis	01	0.3%
Microcephaly	11	3.6%
Holoproscencephaly	12	3.9%
Porencephaly	02	0.6%
Hydranencephaly	02	0.6%
Schizencephaly	01	0.3%
Kyphosis	01	0.3%
Teratoma	01	0.3%
Total	123	40.4%

The Genitourinary tract (GUT) anomalies were diagnosed in 59 foetuses. Among them hydronephrosis were seen in (26) foetuses. The cystic renal diseases were documented in 10 foetuses. The commonest presentation was the presence of multiple cysts of varying sizes. Bladder outlet obstruction was detected in 06 foetuses.

Table: 2, Distribution of genitourinary anomalies.

Anomalies	No	% age
Hydronephrosis	26	8.5%
Cystic kidneys	12	3.9%
Bladder Outlet Obstruction	08	2.6%
Renal Agenesis	11	3.6%
Megacystics	02	0.6%
Total	59	19.4%

The skeletal system was involved in 26 cases. Limb reduction abnormalities were seen in 19 cases followed by 05 cases of achondroplasia.

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Table: 3, Distribution of skeletal anomalies.

Anomalies	No	% Age
Limb bone shortening	19	6.2%
Achondroplasia.	05	1.6%
Osteogenesis imperfecta	02	0.6%
Total	26	8.5%

The gastrointestinal (GIT) anomalies were seen in 31 foetuses. Atresias-esophageal, duodenal and intestinal predominated and were followed by diaphragmatic hernias in 09. All of them presented with considerable degree of polyhydramnios.

Table: 4. The distribution of gastrointestinal anomalies.

Anomalies	No.	Percentage
Esophageal atresia	08	2.6%
Duodenal atresia	06	1.9%
Intestinal atresia	02	0.6%
Diphragmatic hernia	09	2.9%
Omphalocele	01	0.3%
Gastrochisis	05	1.6%
Total	31	10.2%

The cardiovascular system CVS was affected in 16 foetuses in the form of major structural abnormalities.

Table: 5. The distribution of CVS anomalies.

Anomalies	No.	% age
Cardiomegaly	03	0.98%
Dextrocardia	02	0.6%
Ectopia Cordis	01	0.3%
Atrial septal defect	02	0.6%
Ventricular septal defect	03	0.98%
Fallot's tetralogy	01	0.3%
Right atrial dilatation	03	0.98%
Total	15	4.9%

The other or miscellaneous group included 50 foetuses. Among them 10 foetuses had classical features of hydrops (non- immune) and 7 had jugular lymphatic obstruction sequence. Multisystems were involved in 13 cases.

Table: 6. Distribution of miscellaneous anomalies.

Anomalies	No.	% age.
Hydrops	10	3.3%
Jugular lymphatic obstruction sequence.	07	2.3%
Multisystemic	13	4.27%
Macrosomia	02	0.6%
Ovarian cyst	01	0.3%
Pleural effusion	03	0.98%
Pericardial effusion	05	1.6%
Amniotic band disruption.	05	1.6%
Cystic adenomatoid formation of lung	04	1.31%
Total	50	16.5%

DISCUSSION

Many reports have highlighted the advantage of routine foetal surveillance by antenatal ultrasonography for various indications. Some studies have argued against the usefulness of the same. The interpretation and comparison of the results of these studies are difficult because of varying criteria. The present study was limited to determining the incidence and systemic distribution of major anomalies in the specified population.

The overall incidence of major foetal anomalies in the study was 3.04% which falls within the range reported in the literature (Reyneir et al[2], Whiteman & Reece[3] and Anderson et al[4]). Different authors have reported an incidence ranging from 1.27 to 3% in larger series. The true incidence should have been more if the intrauterine demises, molar pregnancies and multiple gestations were included.

The CNS was the commonest involved in this study (40.4%). Weston et al [6] reported similar findings. Among CNS anomalies the Neural tube defect (NTD) dominated. Among sonographically identifiable NTD anencephaly has the highest incidence (10.2%) and higher sensitivity & specificity of early diagnosis. Spinal defects contributed for 0.98% of the major anomalies among singletons. This is almost equal to that reported from United Staes[7]. The incidence among the newborns is decreasing with the widespread use of antenatal ultrasound scanning in the past three decades. Hydrocephalus was the 2nd commonest (11.84%) CNS anomaly after anencephaly. This usually manifests in 2nd trimester and needs serial assessments for diagnosis and prognostication. Its incidence is reported to be 0.3 to 0.8 per 1000 births[8]. Corpus callosal agenesis and Dandy Walker malformations were relatively infrequent in my study. The commonest subtype of holoprosencephaly noted in 25 foetuses was the alobar variety. The incidence of holoprosencephaly decreased by the time of birth because of early spontaneous abortions. Microcephaly contributed for a significant share (3.6%). A normal cephalometry in early trimester doesn't exclude the possibility of microcephaly.

Next to CNS the genitourinary tract was most commonly involved (19.4%). The commonest presentation was as unilateral (Pelviureteric junction obstruction) or bilateral hydronephrosis (bladder outlet obstruction) in 3.6%. This is consistent with the report that the commonest genitourinary tract anomaly is hydronephrosis. Cystic renal diseases diagnosed among 12 foetuses except in cases of infantile polycystic kidney disease. Bladder outlet obstruction was detected by early second trimester [13]. Severe degree oligohydramnios in the absence of urinary bladder shadow on repeated attempts was associated with bilateral renal agenesis. The incidence of this anomaly was 3.6% in our studies.

The skeletal system anomalies were the 4th commonest contributor (8.5%). The overall incidence of skeletal anomalies is reported to be 2.4 per 10,000 births [15]. Most of them manifested by shortened long bones. Few cases of achondroplasia and lethal achondroplasia could be diagnosed with 100% specificity. However a specific antenatal classification of the skeletal dysplasia is often difficult.

The GIT anomalies (10.2%) were mainly in the form of atresias associated with considerable degree of polyhydramnios. A recent study has estimated the overall incidence of this anomaly as in 2,200 births[17]. The incidence of abdominal wall defect is also low (1.6%) as reported by a European study[18].

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The incidence of CVS anomalies was low (4.9%). This was due to the fact that the population surveyed included random sample and not high risk group alone. It has been established that only significant structural anomalies could be diagnosed antenatally and that also in the later period gestation. It has to be stressed that the four chamber view and the outflow tracts and aortic arch details supplemented by Colour Doppler mapping increase the diagnostic sensitivity of cardiac anomalies. Early trimester Trans-vaginal scanning also enhances the detection rate.

Among the miscellaneous group (16.5%) the majority was diagnosed as non-immune hydrops (3.3%). Jugular Venous lymphatic obstruction was seen in (2.3%). Because of the fact that these foetuses have increased chromosomal abnormality and usually undergo spontaneous abortion in early pregnancy, the number of affected foetuses after birth is much less. Multisystemic anomalies in foetuses hinted at the possibility of chromosomal abnormalities. Macrosomia was significantly less (0.6%) as compared to 1-2 % incidence in developed countries.

CONCLUSISON:

The overall incidence of major anomalies among singleton pregnancies was 3.04% with the commonest system affected was CNS followed by genitourinary tract and skeletal anomalies. The validity of prenatal ultrasonography examination in detecting foetal congenital abnormalities was found to be very high. The study recommended that ultrasonography examination should be become part of the antenatal care provided in primary health care centres.

Acknowledgement:-
The authors thanks to Department of Obstetrics & Gynaecology and Department of Radiology, JLNMCH, Bhagalpur for the assistance, kind cooperation in the preparation of patient's protocol for this study.

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