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ORIGINAL ARTICLE

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Major anatomical fetal anomalies in Northern Kerala

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OBJECTIVE(S) : To define the incidence and systemic distribution of major fetal anomalies in Northern Kerala.

METHOD(S) : Live 30,030 singletons of 9 to 41 weeks of gestation were scanned and statistically analysed during a period of 15 years and 7 months.

RESULTS : The incidence of major fetal anomalies was found to be 2.59% (P=0.046). The central nervous system was the commonest involved (39.20%), followed by the genitourinary tract (18.09%) and the skeletal system (11.79%). The incidence of neural tube defects was 17.26%. The sensitivity and the specificity values of the screening results were 72.2% and 98.6% respectively with a P value of 0.046.

CONCLUSION(S) : 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 fetal complications.

Key words: fetal anomalies, prenatal diagnosis, ultrasonography, ultrasound scan, antenatal screening

Introduction

The study was aimed at defining the incidence of major fetal anomalies in northern Kerala that could be detected by routine ultrasound scanning. The quantification of the true incidence and the definition of the systemic preponderance help in stratification of the affected fetuses for prognostication and tailoring of management policies.

Material and Methods

The pregnant females referred for routine as well as targeted imaging contributed the population for this study. Only live singleton pregnancies of 9 to 41 weeks' gestation were analysed. Plural pregnancies were excluded because of statistically lower number. 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. Minor anomalies of minimal surgical or cosmetic significance were not included. Those with major anomalies were serially assessed and followed up for confirmation of the diagnosis. Suspicious findings were verified from peroperative findings, autopsy or follow up records.

For ultrasonographic scanning Philips SDR 1550 and Philips P 600 (transabdominal linear convex probes of 3-3.5 and transvaginal probe of 5.5 - 6.5 MHz along with color Doppler facilities) were used. The total population studied was 34,240 fetuses of which 33,030 were singleton and 1,210 plural pregnancies. The author personally conducted all the scannings from January 1985 to August 2000 (15 years and 7 months).

Results

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

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Polyhydramnios of moderate to severe degree was noted in 450 pregnancies of the total sample studied. The central nervous system and gastrointestinal system were commonly involved in these cases. No ultrasonographically detectable cause could be defined in few. Significant degree of oligohydramnios was seen in 85 pregnancies. Genitourinary tract anomalies were found as the major contributor for this. Pregnancies with severe degree oligohydramnios often ended up with early intrauterine demise, so that the cause was unidentifiable by echoes in many instances.

There were 857 anomalous fetuses in the population studied. The system wise distribution is represented in Figure 1. There were 336 fetuses with central nervous system (CNS) involvement (39.21%), 155 with genitourinary involvement (18.09%), 101 with skeletal system involvement (11.79%), 91 with gastrointestinal involvement (10.62%), 45 with cardiovascular involvement (5.25%) and 129 (15.05%) with less frequent anomalies and overlapping multiple anomalies.

Figure 1. System wise distribution of 857 anomalous fetuses (2.59%) among 33.030 live singletons. The less frequent entities including the lymphatic and respiratory systems are included among the "others" (miscellaneous)

Table 1 gives the details of central nervous system (CNS) anomalies. Anencephaly Hydrocephalus accounted for 25% of them.

Table 1. The distribution of the CNS anomalies

Anomalies	Number	Percentage
Anencephaly	108	12.60
Cephalocele	14	1.63
Meningo/myelocele	11	1.28
Spina bifida	06	0.70
Iniencephaly	02	0.23
Acrania	07	0.82
Hydrocephalus	102	11.90
Dandy Walker Malformation	07	0.82
Arnold Chiari malformation	04	0.47
Corpus callosal agenesis	02	0.23
Microcephaly	25	2.92
Holoprosencephaly	25	2.92
Porencephaly	05	0.74
Hydranencephaly	03	0.35
Schizencephaly	03	0.35
Kyphosis	03	0.35
Teratoma	03	0.35
Arachnoid cyst	03	0.35
Choroid plexus cyst (bilateral)	03	0.35
Total	336	39.21%

The genitourinary tract (GUT) anomalies were diagnosed in 154 fetuses (Table 2). Among them, hydronephrosis was seen in 81 fetuses. Fetuses with dilated renal pelves of less than 8 mm before 32 weeks and less than 10 mm after 32 weeks were considered to be within normal limits. The cystic renal diseases were documented in 25 fetuses. The commonest presentation was the presence of multiple cysts of varying sizes. The diagnosis of infantile polycystic kidney disease (IPKD) could be specifically made on typical sonographic features. Bladder outlet obstruction was detected in 22 fetuses.

Table 2. Distribution of genito urinary anomalies

Anomalies	Number	Percentage
Hydronephrosis	81	9.45
Cystic kidneys	25	2.92
Bladder outlet obstruction	22	2.57
Renal agenesis	14	1.63
Megacystis	05	0.58
Echogenic kidneys	06	0.70
Extrophy	01	0.12
Ureterocele	01	0.12
Total	155	18.09

The skeletal system was involved in 101 fetuses (Table 3). Limb reduction abnormalities seen in 56 were the commonest followed by 16 cases of achondroplasia. A very precise typing of the fetuses with limb shortening was difficult antenatally.

Tabel 3. The distributin of skeletal system anomalies

Anomalies	Number	Percentage
Limb bone shortening	56	6.53
Achondroplasia	16	1.87
Achondrogenesis	08	0.93
Osteogenesis imperfecta	04	0.47
Thanatophoric dysplasia	03	0.35
Thoracic dysplasia	13	1.52
Hypophosphatasia	01	0.12
Total	101	11.79

The cardiovascular system (CVS) was affected in 45 fetuses (Table 4) in the form of major structural abnormalities. Seven fetuses had arrhythmias in addition.

Table 4. The distribution of gastrointestinal anomalies

Anomalies	Number	Percentage
Esophageal atresia	25	2.92
Duodenal atresia	13	1.52
Intestinal atresia	12	1.40
Diaphragmatic hernia	25	2.92
Omphalocele	08	0.93
Gastrochisis	05	0.47
Meconium peritonitis	03	0.35
Total	91	10.51

The gastrointestinal tract (GIT) anomalies were seen in 91 fetuses (Table 5). Atresias (esophageal 26, duodenal 13 and intestinal 13) predominated and were followed by diaphragmatic hernias in 25. All of them presented with considerable degree of polyhydramnios.

Table 5. The distribution of CVS anomalies.

Anomalies	Number	Percentage
Cardiomegaly	08	0.93
Dextrocardia	04	0.47
Ectopia cordis	03	0.35
Atrial septal defect	05	0.58
Ventricular septal defect	03	0.35
Fallot's tetralogy	02	0.23
Hypoplastic left ventricle	03	0.35
Right atrial dilatation	08	0.93
Echogenic mitral valve	02	0.23
Arrhythmias	07	0.82
Total	45	5.24

Note : Some women had more than one anomally (38 women had 45 anomalies)

The others or miscellaneous group included 129 anomalous fetuses (Table 6). Among them, 34 fetuses had classical features of hydrops (non-immune) and 22 had jugular lymphatic obstruction sequence. Multiple systems were involved in 37 fetuses making it difficult to classify the anomalies. One fetus showed the presence of an adrenal mass which was characteristic of a hemorrhage on serial postnatal scanning.

Table 6. Distribution of other or miscellaneous anomalies

Anomalies	Number	Percentage
Hydrops	34	3.97
Jugular lymphatic obstruction sequence	22	2.57
Multisystemic	37	4.32
Amniotic band disruption	07	0.82
Limb body wall complex	03	0.35
Macrosomia	07	0.82
Ovarian cyst	04	0.47
Cystic adenomatoid formation of the lung	03	0.35
Adrenal hemorrhage	01	0.12
Pleural effusion	07	0.82
Pericardial effusion	04	0.47
Total	129	15.05

The postnatal follow up revealed that 10 cases of major anomalies were missed on routine screening. These included two cases each of spina bifida, esophageal atresia and small ventricular septal defect and one case each of bladder extrophy, arthrogryposis multiplex congenita, atrial septal defects, and unilateral facial hypoplasia.

Discussion

Many reports have highlighted the advantage of routine fetal 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 incidence of polyhydramnios among the singleton pregnancies analysed was 1.36%. This is slightly higher than the incidence of 0.93% published after an objective assessment of 9000 subjects [1]. As the present analysis was based on the subjective impression of the author, a true comparison is difficult.

The overall incidence of the major fetal anomalies in this study was 2.59% which falls within the range reported in the literature (Reyneir et al [2], Whiteman and 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. In a multicentric randomized study of 15,151 fetuses, it was found that only 35% of anomalous fetuses could be detected before birth [5].

The CNS was the commonest involved in this study (39.20%). Weston et al [6] reported similar findings. Among CNS anomalies the neural tube defects (NTD) dominated. Of the sonographically identifiable NTDs like anencephaly, cephalocele, meningocele, spina bifida, iniencephaly, and acrania anencephaly has the highest incidence (12.60%) and higher sensitivity and specificity of early diagnosis. Spinal defects contributed for 1.98% of the major anomalies among singletons. This is almost equal to that reported from United States [7]. The incidence among the newborns is decreasing with the widespread use of antenatal ultrasound scanning in the past three decades. Seven fetuses were diagnosed to have acrania which could be easily mistaken for anencephaly in the late first trimester. Hydrocephalus was the second commonest (11.90%) CNS anomaly after anencephaly. This usually manifests in second 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 our study though the former shows an incidence of 1-5% of all pregnancies and the latter has an incidence of 1 in 30,00 births [8,9]. The commonest subtype of holoprosencephaly noted in 25 fetuses was the alobar variety. The incidence of holoprosencephaly decreases by the time of birth because of early spontaneous abortion [8]. Isolated bilateral choroid plexus cysts were seen in only three fetuses (0.35%). This is contrary to the reported incidence of 3-4% in second trimester pregnancies and 1-2% in general population [10]. Microcephaly contributed for a significant share (2.92%). A normal cephalometry in early trimesters doesn't exclude the possibility of microcephaly.

Next to CNS the genitourinary tract was most commonly involved (18.09%). The commonest presentation was as unilateral (pelviureteric junction obstruction) or bilateral hydronephrosis (bladder outlet obstruction) in 12.02%. This is consistent with the report that the commonest genitourinary tract anomaly is hydronephrosis. A certain degree of minimal prominence of the collecting system (less than 8mm before 32 weeks and less than 10mm after wards) was considered insignificant. This is supported by the observation that 43.1% of pelvicalyceal dilation is likely to regress postnatally [12]. Cystic renal diseases diagnosed among 25 fetuses showed overlapping features 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 1.63% in our study compared to an incidence of 0.1 to 0.3 per 1000 live births reported by (Wilson and Baird [14]).

The skeletal system anomalies were the third commonest contributor (11.79%). 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 achondrogenesis could be diagnosed with 100% specificity. Thoracic dysplasias had a significantly noticeable incidence of 1.52%. However, a specific antenatal classification of the skeletal dysplasias is often difficult.

The gastrointestinal tract (GIT) anomalies (10.62%) were mainly in the form of atresias associated with considerable degree of polyhydramnios. The reported incidence of GIT involvement is 5-7% among all anomalies [16]. Few fetuses with esophageal atresias showed a small stomach shadow because of tracheo-esophageal fistula. A recent study has estimated the overall incidence of this anomaly as 1 in 2,200 births[17]. The incidence of abdominal wall defects is also low (1.51%) as reported by a European study [18].

The incidence of cardiovascular system (CVS) anomalies was low (5.24%). 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 of gestation. Since the early part of this study was done with a black and white scanner, it is possible that a few lesions could have been missed. In a recent study involving 22,050 fetuses followed up by clinical and autopsy findings the incidence of cardiac anomalies was 7.6 cases per 1000 live births [19]. 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 transvaginal scanning also enhances the detection rate.

Among the miscellaneous group (15.05%) the majority was diagnosed as immune hydrops (3.98%). There were no cases of immune hydrops in the sample analysed. This was followed by 2.57% of fetuses with classical features of jugular lymphatic obstruction sequence. The incidence of jugular lymphatic obstruction sequence in our population is significant[20]. Because of the fact that these fetuses have an increased chromosomal abnormality and usually undergo spontaneous abortion in early pregnancy, the number of affected fetuses after birth is much less. All tne fetuses with intrapelvic cystic masses were females. In one fetus, the

cyst was occupying the subhepatic region. Postnatal surgery revealed the cysts as simple serous cyst in two fetuses and dermoid in one fetus. Multisystemic anomalies in 37 fetuses hinted at the possibility of chromosomal abnormalities among some fetuses. The figures in this category couldn't be derived since the chromosomal analysis was incomplete. Unilateral cystic adenomatoid malformation of the lung (Type 1) was diagnosed in three fetuses. This anomaly accounts for 25% of congenital lung lesions in the literature [21]. Macrosomia (0.82%) was significantly less as compared to 1-2% incidence in developed countries.

The Medline search didn't yield a similar study of longer duration conducted by a single author in the literature. The results were compared with the other published reports using the test for proportion. It was found that the overall incidence of major anomalies in our study showed highly significant value (P=0.046). This can be explained on the bank of populations characteristris and sample features. When compared to earlier emphsis published by us in 1999, there is slight increase in the insidence 22 that could be due to increased awarness among the doctor and the availability of better machines as the technology adamen. The variabration of the incidence rates of system irroplevent in different ethnic groups demands further properties trials. The samiliarity is considerably affected by the personirralved, the demonstration of the anomalies, the population charactiristic and the method of calculation, adopted 23.

Conclusion

The overall incidence of major anomalies among givinglet9on pregnancies was 2.59% the commonest system affeted was the CNS followed by the genitoinary tract and sheteal system amomalies. Sping bofrdx showing relatively low residence. The CVS showed abwest and modeficate follow up. There incidence alrania and jigular hymphasis obstruction segnance are imp0ortant since both these conditions are immediately left and awareness may lead to earlier deterlar and prevention intervention. At least one antenatal retrasound screening in the late first trimester is recommended.

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