Original article

Prenatal Diagnosis of Congenital Anomalies-Preliminary Report From A New Specialized Ultrasound Unit

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ABSTRACT

Background: Most structural malformations are not detected until birth in less developed countries unlike advanced nations. We decided to assess the role of anomaly scan by specialists in a newly installed, sole specialized unit in a Nigeria town.

Methodology: Pregnant women referred to the unit were counseled and recruited for the study after informed consent. GE Voluson 730 professional ultrasound machine with 2D & 4D probes was utilized to scan the patients at 20 weeks with a repeat at 34 weeks gestation. The newborns were then clinically examined, investigated and or autopsied.

Result: Five thousand fetuses were examined within eighteen months. Twenty anomalies with prevalence rate of 4 per 1000 were discovered. Head/Neck/spinal anomalies were the highest specifically anencephaly .Renal anomalies were also common. The sensitivity and positive predictive values were 100%.

Conclusion: Anomaly Scanning at 20 weeks and 34 weeks reduces perinatal mortality and morbidity, provides opportunity for early treatment and reduces the financial, physical and emotional stress on parents. It is also very reliable and sensitive if (and should be) done by specialists with high resolution 2D & 4D ultrasound machines.

Key Words: Prenatal Congenital Anomalies 4D

INTRODUCTION

First time discovery of termed newborns with gross crippling-sometimes not compatible with life anomalies still occur in our labour wards in Nigeria despite proliferation of prenatal ultrasound centres. It is indeed heart breaking seeing the happiness of the mother turn sour when confronted with malformed baby at termed delivery despite the ''normal pregnancy" verdict of several prenatal ultrasound.

Only the few established institutions have the required skill and equipment to provide quality scanning to patients yet they all thrive due to ignorance on the side of the patients and lack of policy regulating ultrasound practice.

We therefore decided to assess the prevalent rate and sensitivity of (diagnosing) congenital anomaly in our newly opened specialized ultrasound unit (the only one in the town) and compared our results with previous studies.

METHODOLOGY

Ladoke Akintola University Teaching Hospital Ogbomoso was commissioned about two years ago. It's radiology unit is the only specialized radiology centre in the big town located in the northern part of Oyo State. Oyo state is in South western part of Nigeria.

All the patients referred to the radiology department for routine antenatal scanning were recruited for the study. They were counseled and recruited after informed consents. The gestational ages of the pregnancies were confirmed by first trimester ultrasound. Anomaly scans were then carried out between nineteen and twenty-one weeks gestation and repeated at thirty four weeks of gestation using Voluson 730 professional General Electric machine with 4D and 2D abdominal probes.

Patients were informed of the results, counseled on the appropriate means of management. The structures examined were fetal skull and brain including the cavum septi pellucidi, the ventricles, cerebellum and cisterna magna; neck, face, thorax, abdomen, spine and limbs. Both 2D and 4D (especially of the face) were done.

The newborns had detail clinical examination and investiga-

tions while aborted fetuses or stillborn babies had autopsies. Sonographic soft makers like choroid plexus cysts, mild pyelectasis, echogenic bowel and abnormal umbilical cord vessels were not considered in our analysis. The sensitivity and positive predictive values of ultrasound with regard to congenital anomaly detection were made by fetus and not by malformation.

RESULTS

A total number of seven thousand fetuses were scanned during the period. Five thousand were included in the study. Those excluded were due to various reasons which included refusal of the pregnant woman to participate in the study, lost to follow up.

The mean age of the pregnant women was 28 years; two thousand were primigravida and three thousand were multigravida. None of the patients gave past history of congenital anomaly in previous births and no prior ultrasound diagnosis of congenital anomaly (in earlier scans) nor clinical suspicion of such. Only ten of the pregnancies were twin gestations, others were singleton. A total number of twenty congenital anomalies were detected.

I Head/Neck/Spine	
Anencephaly	5
Hydrocephalus	4
Cystic Hygroma	1
Spinal Bifida With Mennogocoele	2
Total	12
II Cardio Pulmonary/Chest	
Hydrothorax	1
III Gastrointestinal	0
IV Renal	
Renal Agenesis	0
Post Urethral Valve	3
Unlateral Hydronephanosis	0
V Skeletal Dysplasia	0
VI Others	
Sacrococcygeal Mass	1
Ascites	3

The prevalence of malformation in this unselected population was 4 per 1000 with 100% sensitivity and positive. There was no false negative diagnosis. Twelve (60%) of the anomalies were seen in the Head, neck and spinal regions while three (15%) of the anomalies were seen in the renal system.
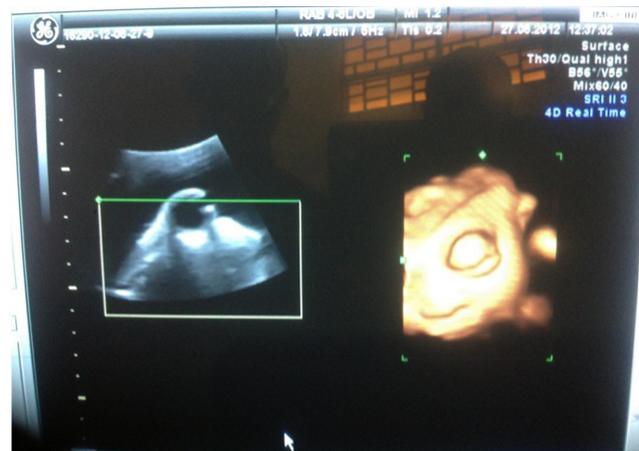
[Table/Fig-1]: Prenatal Ultrasound 2D (Left) and 4D Right showing absent cranium and cerebral hemispheres over the prominent orbits (Anencephaly)

[Table/Fig-2]: 2D (Left) and 4D Right ulltrasound showing the facial features of the fetus to exclude cleft lip and other facial anomalies

[Table/Fig-3]: Prenatal ultrasound showing minimally dilated lateral ventricles at 20 weeks gestation; repeat scan at 34 weeks gastation was normal

[Table/Fig-4]: B mode ultrasound showing the characteristic frog sign in anencephaly

[Table/Fig-5]: Prenatal ultrasound showing dilated fetal posterior urethral and urinary bladder (posterior urethral valve)

DISCUSSION

The improved ultrasound technology offering better resolution and improvement in knowledge and experience of ultrasound examiners have resulted in increased detection rates of fetal malformation since the 1990s [1].

However, in Nigeria like most less developed countries, most fetal anomalies are not detected until birth not because of non availability of ultrasound services but rather due to the fact that ultrasound is arguably one of the most abused instrument: most ultrasound services are provided by poorly trained or untrained medical and non medical personnel's who are benefiting from lack of policy regulating the use of ultrasound.

The non detection of anomalies prenatally has definitely contributed to the high perinatal mortality in Nigeria and has significant psychological effect on the couples. It was noted that all the pregnant women denied history of congenital anomaly in previous births. This might not be unconnected with embarrassment and social stigma that may be associated with birth of a "deformed child" in our environment. Our study was conducted between nineteen and twenty-one weeks of gestation because most authors agreed that ultrasound screening for fetal structural abnormalities is best carried out between 19-21 weeks of gestational age [2, 3, 4].

Also, like most of the authors we repeated the ultrasound scan in the third trimester since it has been found that most structural anomalies are increasingly detected with advancing gestation [5]. The sensitivity rate in our study was 100%. This was rather high although, sensitivities as high as 85-90% had been reported, they were in studies conducted in populations at specific risk and at centres of excellence by expert operators [2]. Most studies conducted in general population like we did showed widely varied detection rates with ranges between 8.7% to 85% [6]. The wide differences were said to reflect varying criteria for definition of malformation, post natal examination, selection of study population, prevalence of specific anomalies within a population and other methodology issues (e.g. single hospital versus multi center setting, expertise and skill of operations, use of standardized protocols for ultrasonograhic examination) [7].

The high sensitivity rate in our study was multifactoral; fewer number of study population, the high resolution of ultrasound used with a combination of B mode 2D and 4D scanning and conduction of the scanning by a consultant radiologist.=Also, some anomalies missed on ultrasound might not be recorded post delivery because cultural issues, ignorance and mood of most parents made them to refuse thorough investigations and or pathological examination of the aborted fetuses and stillborns.

Furthermore, children with some anomalies may remain "normal" for several years without symptoms [2] for example infants with congenital hydronephrosis may appear normal for months or even years before development of symptoms or eventual diagnosis [2]; infant with mild/borderline ventriculomagaly may later presents with neurological deficits [2].

Ramosan et al., [1] reported a sensitivity/detection rate of 68% in their study of the larger population of 16,775 fetuses while Carole A Lack 3 reported sensitivity and specificity rates of 85% and 99.9% in a study of 8849 deliveries though the scans were performed by three radiographers with diploma in medical ultrasound and minimum of four years experience in obstetrics scan. Rosendah and Kwenen reported 58.1%, 99.9%, 91.5% and 99.6% sensitivity, specificity, positive predictive and negative predictive values respectively in a general population study of 9012 fetuses [8].

Comparison of sensitivity or detection rates between studies was said to be meaningless because the variation in detection rate reflects differences in examination skill and quality of equipment; differences in definitions of malformation, detection rate and quality of follow up 1. Boyd et al., [9] went ahead to discuss the problem of defining agreement between prenatal and postnatal diagnoses. There was eventually no cases of false positives in our study because of the three cases (of) dilated ventricle discovered at 20 weeks gestation that regressed at 34 weeks gestation scan were not included.

The issue of false positive diagnosis of fetal malformations have been fairly well [10] or very well described and discussed [9, 11]. Reported false-positive rates with regard to fetal malformations (defined as 1-specificity) are usually < 0.1% 9,10 but in two studies, the false positivity rate was as high as 0.48% and 0.40% [12, 13].

This variation in false positive rates may be partly due to differences in definitions e.g. whether or not ultrasound findings that were subsequently refuted or that regressed during pregnancy (such as dilated kidney pelvis) were included or excluded [1]. Such findings were excluded in our study.

Several other studies [1, 5, 10] reported refuted or regressed false positive diagnoses of hydonephrosis [1], hygromas, ascites, intra abdominal cysts, heart malformation [9], esophageal atresia [5], abnormally shaped head and cleft lip [1].

Gaglioti et al., [14] emphasized the fact that some diagnosis classified as false positives: for example substantial proportion of fetuses with mild ventriculomegaly discharged home as 'normal newborn' developed psychomator problems later in life.

Our prevalent rate of 4 per 1000 (20/5000) was comparable to that of Romosan et al., [1] found in chromosomally abnormal fetuses but quite lower than 18.0 per 1000 and 13.2 per 1000 they found in chromosomally normal fetuses with major and minor malformations respectively.

Sixty percent (12 anomalies) of the anomalies seen were in the Head/neck and spinal region [Table/Fig-1,2,3,4] with Anencephaly responsible for the highest number [5]. This was followed by 15% of the anomalies seen in the renal system [Table/Fig-5]. All the five cases of Anencephaly accepted termination of pregnancy after counseling.

Grandjean et al., [5] reported the highest prevalence in musculo skeletal anomalies in their study of 4615 malformations while renal anomalies constituted the highest number in Carole A Luck study [3].

In conclusion, anomaly scan should be a must for all pregnant women. The sensitivity rate and predictive values are good if carried out by well trained individuals using good quality, high resolution 2D and 4D ultrasound machines.

The detection of crippling anomalies and possible termination of the pregnancies is not only cost effective but spares the parents from the emotional and physical stress.

Early detection of anomalies also provide opportunity for treatment which may be life saving. However, there is a need to regulate the use of ultrasound, provide specialized training to prevent patients (and spouses) from emotional torture of false positive diagnoses.

REFERENCES

[1] Romosan G., Henriksson E, Rylander A, Valentin L. Diagnostic performance of routine ultrasound screening for fetal abnormalities in an unselected Swedish population in 2000-2005. *Ultrasound Obstet Gynecol.* 2009; 34: 526-33.

[2] Tochos T, Capuzzo E, Gaglioti P. Prenatal diagnosis of congenital anomalies. *Images Paediatr Cardiol.* 2001 Apr- June; 3 (2): 3-18.

[3] Carole A Luck. Value of routine ultrasound scanning at 19 weeks: a four year study of 8849 deliveries. *BMJ.* 1992; 304: 1474-8

[4] Todrus T, Faggiano F, Chiappa E, Gaglioti P, Mutola B, Sciarrone A. Gruppo Piemontese for Prenatal Screeening of congenital Heart disease. Accuracy of routine ultrasonography in screening heart disease prenatally. *Prenat Diagn.* 1997; 17: 901-06.

[5] Grandjean H, Larroque D, Levi S. Eurofetus study Group. The performance of routine ultrasonographic screening of pregnancies in the Eurofetus study. *Am J Obstet Gynecol.* 1999; 181: 446-54.

[6] Romero R.Routine obstetric ultrasound. *Ultrasound obstet Gynecol.* 1993; 3:303-07.

[7] Crane J.P, Lefevre ML, Winborn RC, Evans J K, Evingman BG, Bain RP, Frigoletto F D, Mc Neillis D. Radius study Group. A randomized trial of prenatal ultrasono-graphic screening: Impact on the detection, management, and outcome of anomalous fetuses. *Am J obstet Gynecol.* 1994; 171: 392-99.

[8] Rosendahl H, Kivenen S. Antenatal detection of congenital malformations by routine ultrasonography. *Obstet Gynecol.* 1989 June, 73(6): 947-51.

[9] Boyd PA, Chamberlain P, Hicks NR. 6 years experience of prenatal diagnosis in an unselected population in Oxford, UK. *Lancet.* 1998; 352: 1577-81.

[10] Nalding J, Backe B. Routine ultrasound screening and detection of congenital anomalies outside a university setting. *Acta obstet Gynecol Scand.* 2005; 1042-48.

[11] Shirley I.M, Bottomley F, Robinson V.P., Routine radiographer screening for fetal abnormalties by ultrasound in an unselected low risk population. *Br J Radiol.* 1992; 65: 564-69

[12] Saari kemppainen A, Karjalainen O, Ylostalo P, Heinonen O.P. Fetal anomalies in a controlled one-stage ultrasound screening trial. A report from the Helsinki ultrasound Trial. *J Perinat Med.* 1994; 22: 279-89.

[13] Saltvedt S, Almstrom H, Kublickas M, Valentin L, Grunewald C. Detection of malformations in chromosomally normal fetuses by routine ultrasound at 12 or 18 weeks of gestation-a randomized controlled trial in 39572 pregnancies. *BJOG.* 2006; 113: 664-74.

[14] Gaglioti P, Danelon D, Bontempo S, Mombro M, Cardaropoli S, Todros T. Fetal cerebral ventrculomegaly; outcome in 176 cases. *Ultrasound obstet Gynecol.* 2005; 25: 372-77.

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