ANTENATAL DETECTION OF CONGENITAL ANOMALIES

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SUMMARY

200 pregnant patients were scanned in the 2nd and 3rd trimester of pregnancy, by real time ultrasound. Of them 8 cases had anencephaly, 3 each had encephalocele, hydrocephalus and meningomyelocele. The gastro-intestinal malformations detected were omphalocele, foetal ascites, duodenal and oesophageal atresia in 1 case each. Bilateral pleural effusion was also detected in one case. Two of the cases had multiple congenital anomalies.

Ultrasound played a vital role in the prenatal diagnosis of these congenital abnormalities and their proper management.

INTRODUCTION

One of the most traumatic experiences not only for the gravid woman, but also for the obstetrician is to deliver a congenitally malformed child.

Major malformations occur in 4% to 5% of all births and are associated with 20% to 25% of all perinatal deaths. In view of this it would seem imperative to evaluate the foetus during the antenatal period. Foetal survillance techniques developed in the last decade now permit the prenatal diagnosis of many malformations (Rayburn and Barr 1985).

While ultrasound occupies a central position in the diagnosis of foetal physical defects, it is not yet widely used as a screening tool for the diagnosis of congenital anomalies in all gravidas. As a result the full potential of this modality for the antenatal diagnosis of birth defects, has yet not been achieved.

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METHOD AND MATERIAL

The study was conducted in the Department of Obstetrics and Gynaecology, S.N. Medical College, Agra.

200 pregnant women were scanned in the Second and third trimesters for various indications.

The equipment used was real time sector ultrasound scanner type 1849, Bruel and Kjaer make.

TABLE NO. I

SHOWS THE CONGENITAL ANOMALIES DETECTED

Anomaly	No. of cases detected	% of study group	Diagnosis missed	% of study group
(A) CNS DEFECTS				
1. Anencephaly	8	4	–	–
2. Encephalocele	3	1.5	–	–
3. Hydrocephalus	3	1.5	–	–
4. Meningomyelocele	3	1.5	1	0.5
(B) GASTRO INTENTINAL DEFECTS				
5. Omphalocele	1	0.5	–	–
6. Foetal ascites	1	0.5	–	–
7. Duodenal atresia	1	0.5	–	–
8. Esophageal artesia	1	0.5	1	0.5
THORACIC DEFECTS				
9. Pleural effusion	1	0.5	–	–

OBSERVATIONS AND RESULTS

The observations and results are summarised in Table no.1.

Since anencephaly is incompatible with life, it is desirable to make the diagnosis early in pregnancy. In this study 8 cases (4%) of anencephaly were detected one of our patient had twin pregnancy, of which, one foetus was anencephalic. There were no false positive or false negative cases in the present study.

Hydrocephalus was detected in 3 cases (1.5%). In only one case isolated hydrocephalus was detected.

Open spina bifida had an incidence of 1.5% in this study. In one case it was associated with hydrocephalus and myelomeningocele.

Encephalocele was detected in 3 cases (1.5%) and there was no false positive or false negative case.

Gastro-intestinal abnormalities were the second largest group of anomalies detected. The defects encountered were foetal ascites, omphalocele, duodenal atresia and oesophageal atresia.

Oesophageal atresia was missed in a primigravida, to be diagnosed postnatally. The neonate was operated, but unfortunately did not survive. In general it may be commented that diagnosis of oesophageal atresia is difficult.

Bilateral pleural effusion was detected in one case, who presented with recurrent foetal wastage. The foetus also had anencephaly and small myelomeningocele in the cervical region, which was unfortunately, missed by the ultrasound.

DISCUSSION

The detection rate of neural axis abnormalities, reported by Campbell (1983) was 80% while Mok et al (1985) have reported a detection rate 91.2%. In our study the detection rate of neural tube abnormalities was approximately 94%.

According to Hutson et al (1985) and associates the accuracy rate of ultrasound in gastro-intestinal malformations was 62.5%. In our study the detection rate was 75%.

The overall accuracy of ultrasound in the detection of congenital anomalies, reported by Sabbagha et al (1988) and associate was 96 per cent. In their study the false normal and abnormal diagnosis were 0.5% and 0.6% respectively. In our study the overall accuracy of ultrasound was approximately 91%, with no false abnormal diagnosis.

CONCLUSION

Ultrasound is a boon to obstetrics, for it is a safe, simple, non-invasive procedure easily acceptable to all. It has today, revolutionised the outlook of an Obstetrician. The prenatal diag nosis of congenital anomalies helps th obstetrician in formulating a plan regarding th mode of delivery, timing of delivery, or may offe the parents a safe and simple method of termina tion of pregnancy. In the future, it may play vita role in intra-natal paediatrics.

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