

Detection of Congenital Malformations in Healthy Pregnant Ladies Undergoing Routine Scan

SADIA NAZIR, NASEEM NOOR ELAHI

ABSTRACT

Aim: To detect the congenital malformations in healthy pregnant ladies on routine ultrasonography.

Material and methods: This purposive sampling study was carried out in DHQ Teaching Hospital, Dera Ghazi Khan from October 2011 to October 2012. A total of 480 healthy pregnant ladies attending outpatients department at 18-20 weeks of gestation were screened for congenital malformations with the help of USG.

Results: The incidence of major congenital malformations was found to be 7.1%. The central nervous system was the commonest involved (61.6%), followed by genitourinary tract (11.7%) and gestational 9%. A significant number of fetuses showed anencephaly 4.5% and hydrocephalus 2%. False positive rate was 2.9% while one case of meningocele, spina bifida occulta and bilateral renal agenesis were missed i.e. false negative rate was 5.8%. termination of pregnancy was done in 79% of cases with severe malformations and 21% underwent spontaneous miscarriage or death in utero.

Conclusion: It is concluded that mid-trimester ultrasound examination is an effective and useful method for screening anatomical congenital malformations.

Keywords: Mid trimester ultrasonography, Antenatal screening, fetal malformations.

INTRODUCTION

Ultrasound has number of valuable functions in obstetrics that include dating pregnancy, confirming of viability, counting number of fetuses placental localization and evaluation of certain foetal malformations. It is safe, accurate and cost-effective procedure for detection of foetal malformation. With the improvement in ultrasound imaging equipment the diagnosis of congenital malformation is possible in early pregnancy¹. The optimal gestational age to screen for fetal malformations appear to be between 18-20 weeks². Around three quarters of fetuses with major malformations can be detected on routine scan³.

Most common cardiovascular abnormalities are hypoplastic ventricles aortic stenosis, cardiac hydrops and fetal arrhythmias. Detection of hydrop fetalis, trisomy, multiple congenital anomaly complex with normal chromosomes are also possible in mid-trimester scan⁴. Ultrasound scan also detect soft markers. These are features which themselves are not important for the health and development of fetus but can indicate some underlying problems like chromosomal anomalies. Screening of Down syndrome by mid-trimester ultrasonography shows that detection rate was 63% in women aged less than 35 years⁵. Termination of pregnancy is offered to the women with major malformations where death is

inevitable and with the malformation associated with long term handicapped⁶.

For potentially correctable abnormalities, parents can be prepared to initiate related investigations and appropriate management. Foetal conditions potential for intrauterine therapy as obstructed foetal bladder, which can lead to renal damage without insertion of vesicoamniotic shunts, can be referred to the specialized centres⁷. Early diagnosis of foetal abnormalities which require postnatal management as congenital dislocation of hip joint can facilitate the decision making regarding the mode of delivery and the place of delivery accordingly⁸.

Foetal malformations are responsible for 30% of perinatal in developed countries⁹. Early diagnosis of malformation provides information for decisions during pregnancy, appropriate treatment at birth and prompt transfer to specialized centres for care of newborn. Therefore, perinatal detection should help to reduce perinatal mortality and morbidity. It should be a part of routine care that will improve foetal outcome¹⁰.

MATERIAL AND METHODS

This purposive sampling study was carried out in DHQ Teaching Hospital, Dera Ghazi Khan from October 2011 to October 2012. A total of 480 healthy pregnant ladies attending outpatients department at 18-20 weeks of gestation were screened for congenital malformations with the help of USG.

Department of Obstetrics & Gynaecology, DHQ Hospital, Dera Ghazi Khan
Correspondence to Dr. Sadia Nazir, District Gynaecologist

RESULTS

The incidence of major congenital malformations was found to be 7.1%. The central nervous system was the commonest involved (61.6%), followed by genitourinary tract (11.7%) and gastrointestinal 9%. The incidence of neural tube defect was 44.3%. A significant number of fetuses showed anencephaly 4.5% and hydrocephalus 2%. False positive rate was 2.9% while one case of meningocele, spina bifida occulta and bilateral renal agenesis were missed i.e. false negative rate was 5.8%. Termination of pregnancy was done in 79% of cases with severe malformations and 21% underwent spontaneous miscarriage or death in utero. Out 480 patients, 149 (31%) were primigravida and 331 (69%) were multigravida. On ultrasonography, 34 (7.1%) patients were found abnormal whereas 446 (92.9%) were normal.

Table 1: Age distribution

| Age (years) | Frequency | %age |
|-------------|-----------|------|
| 16-20 | 163 | 34 |
| 21-25 | 115 | 23.9 |
| 26-30 | 158 | 33 |
| 31-35 | 44 | 09.1 |

Table 2: CNS anomalies (n=21)

| Type | Frequency | %age |
|------------------|-----------|------|
| Anencephaly | 9 | 26.7 |
| Spina bifida | 3 | 08.8 |
| Hydrocephalus | 4 | 11.8 |
| Encephalocele | 3 | 08.8 |
| Microcephaly | 1 | 02.9 |
| Hydroanencephaly | 1 | 02.9 |

Table 3: Congenital anomalies associated with polyhydramnios (n=15)

| Type | Frequency | %age |
|---------------------|-----------|------|
| Anencephaly | 9 | 60 |
| Encephalocele | 2 | 13.3 |
| Hydrops | 2 | 13.3 |
| Oesophageal atresia | 1 | 06.7 |
| Cystic hygroma | 1 | 06.7 |

Table 4: Outcome of malformed fetuses (n=34)

| Outcome | =n | %age |
|--------------------------|----|------|
| Termination of pregnancy | 27 | 79 |
| Death in utero | 03 | 09 |
| Spontaneous abortions | 04 | 12 |

DISCUSSION

One stage screen is recommended in UK. This is best done at 18-20 weeks of gestation¹¹. Structural malformations including neural tube defects, gastrointestinal, renal, cardiac and limb defects can be detected by ultrasonography¹².

Out of 480 cases, 34 women (7.1%) had malformed fetuses. A similar increased detection rate of 53% was seen in a study¹³, While in another study the detection rate was quite low (2.3%)⁵. The results of our study are comparable with a study. In that study the most commonly detected anomalies were CNS (39.2%) followed by genitourinary system (18%) and skeletal system (11.7%)¹⁴. But these results vary from a study in which most commonly detected malformations were of genitourinary system (88.5%) and CNS (88.3%)¹⁵. The CNS was the most frequently involved system in our present study as reported by the other major series¹⁶.

The incidence of spina bifida aperta in the form of meningocele and meningocele was 5.9% while spina bifida occulta was 2.9%. If the overall incidence is considered, it contributed for 8.8% of the major malformations in low risk population that was quite high compared to that reported in other studies^{17,18}. In present study 2 cases of hydrocephalus has associated spina bifida (5.8%) which is comparable with European study¹⁵.

One case (2.9%) of microcephaly was noted that was comparable in the a study¹⁴. However, a normal cephalometry in early second trimester does not exclude the possibility of microcephaly¹⁹. The skeletal system malformations were the 4th commonest contributor having 6% of fetal abnormalities that was quite low as compared with the another study¹⁴.

When severe malformations are detected in mid trimester, patient can be offered termination of pregnancy after counseling the couple and discussing prognosis of abnormality. In this way they can be spared of the burden of carrying pregnancy that would result in stillbirth or liver birth of handicapped infants²⁰. In present study, among the malformations detected by UDG, 12% underwent spontaneous abortions and TOP was done in 79% of cases. In 9% of cases IUD of malformed fetus was diagnosed on routine scan. While in a study with severe malformations, there was spontaneous abortions and IUDs in 15% and elective terminations in 41% of fetuses¹⁵. Six patients were diagnosed to have minor structural defects postnatally, even though they had normal anomaly scan in mid trimester. Out of those, 1 was having hypospadias, 1 with phimosis, 1 with low set ears and 3 with talipes. It was observed that the detection of major malformations was better than minor one²¹.

CONCLUSION

It was concluded from the study that mid-trimester ultrasound examination is an effective and useful method for screening of structural malformations.

REFERENCES

1. Simpson JL. Genetic counseling and prenatal diagnosis. In: Gabbe SG, Niebyl JR, Simpson JL editors. Normal and problem pregnancies. New York. Churchill Livingstone 1996; 215-48.
2. Harman SR. In: James DK, Steer PJ, Weiner CP, Gonik B editors. Mid trimester scan 2nd ed. London, England WB Saunders Co. 1997; 175-82.
3. Hunter S, Heads A, Wylie J, Robson S. Prenatal diagnosis of congenital heart disease in northern region of England. *Heart* 2000; 84(3): 294-8.
4. Stoll C, Game E, Clement M. Evaluation of prenatal diagnosis of associated congenital heart diseases. *Prenat Diagn* 2001; 21(4): 243-52.
5. Lee K, Kim SY, Choi SM, Kim JS, Lee BS, Seo K et al. Effectiveness of prenatal ultrasonography in detecting foetal anomalies. *Unse Med J* 1998; 39(4): 372-82.
6. Pardeep K, Junejo MA. Hydrocephalic, pivotal rate of ultrasonography and its management. *Pak Med J Res* 1998; 37(4): 145-7.
7. Charles H, Redeck P, Pandya PP. Perinatal diagnosis of fetal abnormalities. In: Chamberlain G, Steer PJ, Breart G editors. *Tumbull's obstetrics*. 3rd ed. London. Churchill Livingstone, 2001; 170-8.
8. Qureshi AM, Awan TSA, Khan F, Shafique F, Khan MN. An uptodate on congenital diaphragmatic hernia and its management. *J Coll Phys Surg Pak* 1997; 8(2): 75-81.
9. American College of Obstetricians and Gynaecologists. *Ultrasonography in pregnancy*. Washington DC. Am Coll Obstet Gynaecol 1993.
10. Ultrasound screening for fetal abnormalities. Report of RCOG working Party. London RCOG Press 1997.
11. Anthony MV, Cande V, John CS, Beazoglou T. Routine 2nd trimester ultrasonography. *Am J Obstet Gynaecol* 2000; 182(3): 215-20.
12. Skupski DW, Newman S, Edershein T, Milton HJ, Udom RI, Chervenak F et al. Ultrasonography screening of low risk population. *Am J Obstet Gynaecol* 1996; 175: 1142-5.
13. Caroly D [Clinical preventive services} screening ultrasonography in pregnancy [online] 2000 [cited 20 Aug 2004].
14. Balakumar K, Antenatal ultrasound screening for anomalies among singleton. *Cal Med J* 2004; 2(1): 9.
15. Grandjcan H, Carrogue D, Levi S. The performance of routine ultrasonographic screening of pregnancies. *Am J Obstet Gynaecol* 1999; 181: 820-7.
16. Weston MJ, Porter HJ, Andrews WS, Berry PJ. Correlation of antenatal USG and pathological examination. *J Clin Ultrasound* 1993; 21(6): 387-92.
17. Stein SC, Feldman JG, Friedlander M, Klein RJ. Is myelomeningocele a disappearing disease? *Pediatr* 1982; 69: 511.
18. Lorber J, Ward AM. Spina Bifida: a vanishing nightmare? *Arch Dis Child* 1985; 60(11): 1086-91.
19. Bromley B, Benaceraff BR. Difficulties in the prenatal diagnosis of microcephaly. *J Ultrasound Med* 1995; 14(4): 303-6.
20. Luck CA. Value of routine ultrasound scanning. *Br Med J* 1992; 304: 1474-8.
21. Suwanarath C. Detection of fetal anomalies by ultrasonography. *Songkia Med J* 1998; 16: 179-85.