Ultrasound Screening for Fetal Abnormalities

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The aim of the study was to review the recent trends in the incidence, risk factors, pattern, diagnosis and management of various congenital anomalies in our population. *This* cross-sectional, observational study included sixty pregnant women aged 20-35 years. Mode of delivery was decided after considering all these aspects. Total eight (13.3%) fetuses had congenital malformations. The follow-up of these ladies revealed that 26 (43.3%) were lost on follow-up, so the incidence of congenital malformations at birth was found to be 5.8%.
Key words: Screening, ultrasonography,

The antenatal (prenatal) diagnosis of fetal abnormality is, perhaps, one of the most difficult or contentious aspects of modern obstetrics. Professor Ian Donald introduced ultrasound in obstetrics in 1958. The first report of the prenatal diagnosis of fetal malformation by ultrasound (anencephaly) with consequent termination of pregnancy appeared only in 1972. So ultrasonography during pregnancy was incorporated into routine obstetric care before firm scientific evidence was available to justify it. Ultrasound of that era produced images that were extremely crude by modern standards, but the rapid improvements in electronics has made the equipment cheaper and easy to use. Examination may be repeated and are apparently without hazard[1].

The rapid improvements in imaging capabilities has not only made the diagnosis of structural abnormalities much easier, but has also permitted easier performance of invasive, diagnostic (and therapeutic) procedures like amniocentesis, chorion villous sampling etc. During the same time, there have also been extraordinary advances in genetics, and especially molecular genetics, which have greatly broadened and facilitated the prenatal diagnosis of inherited diseases.

Advantages of routine screening have included less frequent labour induction for post-term pregnancy, detection of fetal growth retardation, and identification of malformed fetuses. Luck[2] performed routine ultrasound scanning at 19 weeks in over 8800 British women and concluded that this approach reduced perinatal morbidity and mortality because of pregnancy termination in 25 fetuses with crippling or lethal malformations.

There are many purposes of antenatal diagnosis detection of abnormalities, termination of pregnancy, planning of time, place and method of delivery. Many detected abnormalities are not isolated but they occur either as a syndrome or as major chromosomal abnormalities[(3)]. The fetal chromosomes (the karyotype) may be investigated by amniocentesis, or, more rapidly, by cordocentesis or placental biopsy. Some abnormalities suggest the possibility of infection e.g. fetal hydrops (parvovirus), microcephaly (cytomegalovirus), hydrocephalus (Toxoplasma) indicate the need for appropriate investigation.

The techniques of prenatal diagnosis can be divided into three broad groups, i.e, clinical examination, screening tests like ultrasonography and maternal serum biochemical sampling and invasive procedures like amniocentesis, chorion villus sampling, embryoscopy and MRI. MRI can be used in case of abnormal with pelvic tumours, fetal nervous anomalies and placental haemorragic lesions[3]. However, antenatal diagnosis of many fetal structural abnormalities is possible only by ultrasound and its availability in the last decade has allowed many women to continue through pregnancy, knowing that their fetus would not be handicapped by the same abnormality as a previous baby.

In recent years, several randomized controlled studies[4] and metaanalysis[5] on the routine use of ultrasonography in pregnancy have been published. A decrease in inductions for postdatism has been demonstrated, due to more accurate gestational age determination with ultrasonography.

The effect of routine mid-trimester ultrasonography on perinatal mortality[5], adverse perinatal outcome[4,5] and low birth weight babies is still uncertain. Timely detection of fetal abnormalities by routine ultrasonography has been reported to reduce perinatal terminations for fetal anomalies, thus removing abnormal fetus from the calculations of perinatal mortality rather than improving the positive outcomes of pregnancy. population based studies demonstrate that the general use of obstetric ultrasonography has probably only a modest impact on the number of abnormal babies being born[6]. However, ultrasound imaging is an operator dependent technique. The quality of reports is dependent on the experience and expertise of the ultrasonographer.

Patients and methods

The purpose of this study was to describe our experience of routine ultrasonography at Jinnah Hospital, Lahore and to review the recent trends in the incidence, risk factors, pattern, diagnosis and management of various congenital anomalies. The study was conducted at Radiology Department, Jinnah Hospital, Lahore, from 15.12.2002 to 15.2.2003 in collaboration with the Department of Obstetrics and Gynaecology. Jinnah Hospital is a 1100

bedded teaching hospital affiliated with Allama Iqbal Medical College. The study included sixty pregnant women who were sent for ultrasound examination for a variety of reasons. Majority was referred by the Department of Obstet./Gynae, Jinnah Hospital while others were referred by other hospitals and doctors.

The ultrasound equipment used was Hitachi, (Japan) EUB-310 with a convex electrical transducer of 3.5 mHz. Detailed history including age, LMP, parity was recorded at the time of examination, followed by physical examination and US examination. Both booked and unbooked patients were examined. The age was between 20-35 years. Mode of delivery was decided after considering all these aspects. The information recorded on US examination included: no of fetuses, gestational sac size, CRL, BPD, FL, presentation of the fetus, location and morphology of placenta, amount of liquor, sex, gestational age, FCA and any fetal abnormality.

Results

The total females included in study were sixty. The total fetal abnormalities detected were eight. Hence, the incidence of fetal abnormalities was 13.3%. Fifteen women were primigravida, while 8 women were grandmultipara. The risk factors are mentioned in Table 1. Fifty-eight patients had a singleton pregnancy (96.7%) while two twin pregnancies were encountered (3.3%). The changes in liquor volume and associated anomalies are elaborated in Table 2. The location of placenta and associated abnormalities are described in Table 3.

Table 1. Risk factors

1. History of Diabetes
2. History of Hypertension
3. Previous history of Congenital Malformations
4. History of drug intake
5. History of Infection
6. History of 1st cousin marriage
7. Clinically oligohydramnios
8. Clinically polyhydramnios
9. Any other illness (suspected pituitary tumor, right sided ptosis)
10. Reasons not specified (routine screening)

Table 2: Amount of liquor on clinical examination

Amount of liquor on clinical examination	Ultrasound findings	n=
Suspected oligohydramnios	Oligohydramnios	2
Normal amount	Normal/ adequate	56
Suspected polyhydramnios	Polyhydra-mnios	2

Different malformations, associated anomalies and their percentages are mentioned in Table 4. The follow-up of these ladies revealed that 26(43.3%) were lost on follow-up, so the incidence of congenital malformations at birth was found to be 5.8% (2/34 patients).

Table 3 Location of placenta

Location of placenta	No.	% age	Any associated abnormality
Anterior, upper	27	45	Intraplacental bleeding (43x21mm hypoechoic area anterior to placenta)
Fundal	6	10	
Posterior upper	21	35	Subamniotic hematoma
Low lying reaching lower segment	1	1.6	
Placenta praevia (Type 1)	4	6.6	
Placenta praevia (Type V)	1	1.6	

Table 4. Major fetal abnormality

Major fetal abnormalities	Associated abnormalities	No	Gestational age at the time of diagnosis	%age
Anencephaly	Polyhydramnios and breech	2	25 wks 26 wks	3.4
Omphalocele				
Hydrothorax				
Polyhydramnios				
Microcephaly	Oligohydramnios	2	35 wks	3.4
Right sided holoprocencephaly	-	1	23 weeks	1.67
Bilateral renal Hydronephrosis	Microcephaly Oligohydramnios	1	37weeks	1.67
Right sided hydrocele	-	1	37 weeks	1.67
Fetal hydrops	Breech placenta previa	1	26 weeks	1.67
Total		8	23-37 wks	13.3

Discussion

Ultrasound is invaluable in the diagnosis of fetal malformations. However, some malformations such as anencephaly, large meningo/encephalocele, omphalocele, are diagnose, whereas borderline cases of microcephaly, and abnormalities such as harelip, cleft, palate, polydactyl, and small spina bifida may be overlooked on a routine ultrasound examination. Ultrasound is very helpful diagnosing malformations of the internal structure of the fetal head , thorax and abdomen, as these may not be obvious at the clinical examinations of the neonate, and prenatal diagnosis would prepare the obstetricians and the paediatric surgeon for appropriate intervention.

The diagnosis of a lethal anomaly of the fetus can affect a pregnant woman in a traumatic way. Literature shows that such a loss is very difficult to cope with, and can lead to social isolation and depression. Only a few studies are available regarding length, course and severity of grief in this case[7]. Ultrasound can be employed in three ways to assist the identification of fetal malformations : to visualize the malformations; to facilitate other diagnostic techniques, amniocentesis and chorion villus sampling and to allow fetal measurements (thereby maximizing the

performance of other tests that require accurate knowledge of gestational age). Genetic counseling prior to prenatal testing is important. It is preferable for counseling to be given earlier, to allow time for couples to think carefully without feeling pressured into reaching a decision. The couple should feel free to exercise whatever options they choose. The woman's preferences must be respected[8].

The knowledge of placental appearance (texture guarding) can result in clinical action (fetal assessment techniques and elective delivery) to improve pregnancy outcome. There is compelling evidence of the positive value women attach to being able to 'see their baby' in utero. The RCOG working party on ultrasound screening for fetal abnormalities reported that the objectives of a routine scan include 'psychological support[9,10].

The advent of umbilical artery Doppler examination has been shown to predict and prevent fetal compromise. Doppler ultrasound seems to be safer than CTG, in the management of SGA infants[11]. Scoring system derived from multivariate analysis of Doppler indices demonstrate the potential of being able to identify, in early pregnancy, a group of women at increased risk of developing pre-eclampsia and premature delivery[12].

Regarding the safety of ultrasound, no association could be found between exposure to ultrasound in early fetal life (before 19 weeks) and impaired vision or hearing or growth among 8-9 years old children[13].

The commonest infections implicated in fetal hydrops include cytomegalovirus and parvovirus B-19. In both conditions maternal infection is usually asymptomatic, and the suspicion of fetal infection is usually raised after the ultrasonographic demonstration of fetal hydrops and microcephaly or hydrocephalus[14].

In our study, 5(62.8%) fetal anomalies belonged to central nervous system. Two were neural tube defects (NTD) i.e, anencephaly. Two fetuses had microcephaly and one had holoprosencephaly. Two abnormalities involved genitourinary system. One fetus had omphalocele and fetal hydrops. The results of our study were similar to another study conducted in central Auckland[15].

There is a high incidence of chromosomal abnormalities (50-60%) in trisomy 13 and also in trisomy 18. A 200 fold increased incidence of holoprosencephaly has been described in diabetic mothers. The prognosis is uniformly poor, so termination of pregnancy should be considered throughout pregnancy[16].

Omphalocele is an extra embryonic hernia due to failure of a portion of abdominal wall to form normally, quoted incidence being 1:2500-1: 5000. In our study, the omphalocele was associated with anencephaly, fetal hydrops and polyhydramnios.

Conclusion

Understanding how parents manage the resurgence of mental anguish during a subsequent pregnancy permits healthcare workers to assist these parents. Facilitating parents efforts to develop emotional armor, limit disclosure, delay attachment to the baby, and attach to health care worker and support groups can mitigate social and psychologic discomfort for these parents[18].

The incidence of malformations is quite high in our study (13.3%) as compared to the literature (2-3%). This can be due to referral of high-risk group to the local teaching hospital. The malformed babies (after birth) were 5.8%, excluding 26 patients who were lost to follow-up. Thus, this cannot be a true evaluation of the incidence of malformations, although we may gain valuable information regarding the pattern and common types of fetal malformations occurring in our population.

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