CONTEMPORARY REVIEW ARTICLE

Ultrasound for Screening Fetal Malformations—Has 3-D Made a Difference?

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Abstract

Ultrasonography as a universal screening tool augers well for detection of most of *in utero* congenital abnormalities, particularly with help of color Doppler, 3-D and 4-D scanning. Argument against universal screening is existing anomalies, not detected by USG, manifesting during childhood. Despite many variables of cost and ethics, poorly trained sonographer, it can be ardently stated that a routine scan is warranted for each pregnancy but only if well-performed at the least, level scan II.

Keywords: Ultrasonography, 3-D and 4-D scan, screening, congenital abnormalities, management.

INTRODUCTION

Ultrasound has revolutionized obstetric practise world over, there is no doubt about this. With good resolution machines, color Doppler, 3-D and 4-D scanning it is now possible to make a prenatal diagnosis of many structural anomalies, which are lethal, life-threatening and debilitating.

All pregnancies are at risk of having fetal malformations. Some pregnant women are at a greater risk. The world consensus on whether all pregnancies should be screened by ultrasound for anomalies and when, is still devided.

Presence of anomalies and their undesirable consequences for the affected neonate, family and medical faternity is a very convincing argument by many experts on universal screening. The prevalence of fetal malformations is 6.5% through only 2-2.5% are potentially life-threatening, lethal or represent a major cosmetic defect.[1]

Regardless whether a woman is in low-risk (majority cases) or high risk category (genetic, diabetes, etc.) the risk of fetal malformation is always there and because there are no symptoms and these pregnancies may be uneventful. Ultrasound routine screening is a very valuable tool for detecting congenital anomalies.

A lot of questions on the various aspects of machine technology, expertise, natural history, embryology, treatments available and of course ethical aspects have to be addressed critically and answered before, we declare that routine ultrasound is an absolute necessity.

a. **Is routine screening justified?**
Screening to be justified should fulfil many criteria, the procedure should be safe, reliable, reproducible, easily available and cost effective. For a "at risk" population an ultrasound scan is justified but in developing countries like India where still almost half our pregnant woman have no access to a proper antenatal care, a routine ultrasound currently may not be practically feasible test for screening even though its utility and efficacy are beyond doubt.[2]

b. **Is incidence of fetal malformation is it high enough to merit screening?**
According to Heinonen (1977), approximately 1,50,000 children are born with malformations annually in USA where almost 100% pregnant women have antenatal care and institutional deliveries.[1] In developing countries the incidence is higher due to inability for detection, screening and more exposure to teratogens.

c. **Is outcome of undetected congenital malformations detrimental enough to warrant a routine screening?**
Out of an incidence of around 6% congenital malformations almost half (2.5%) are lethal, life-threatening and have a major cosmetic defect.[3]

Major congenital defect mostly manifest in fetal intrauterine life (ultrasound detectable), sometimes in fetal life (ultrasound suspicion) and occasionally in childhood (ultrasound undetectable) for this reason some experts question the need of routine prenatal ultrasound screening.[4]

Fetal medicine is still not advanced to treat potential life threatening conditions like open neural tube defects and cardiac defects where death is the expected outcome after delivery, occasionally these defective babies survive and are severely handicapped. Diagnosis of such conditions during pregnancy can give the couple an option of termination. Current technology enables detection of over 60% fetal malformations.[5,6]

d. **Can prenatal diagnosis of anomalies ease emotional pain?**
An antenatal diagnosis of congenital anomaly whether lethal, life-threatening or even lesser serious anomaly can still help couples and doctors prepare themselves to the challenge to come.[7] There is a definite benefit of screening for both patients and physicians. A normal ultrasound scan is usually a good news to the parents to be because of the relative low prevalence of anomalies in general population and relative low incidence of false positive results by ultrasound.[8]

If the ultrasound screening is positive for anomaly then counseling and discussion of all options can be done and choice left open to the parents to be.[9]

e. **Is ultrasound prenatal screening cost effective**?
It is difficult to asses cost effectiveness of screening and there are only a few studies on this. Certain costs like purchase, maintainance of equipments, and salary of well trained technicians and doctors can be assessed and is expensive.[10] Emotional costs of family disorganisation and suffering cannot be calculated. Because of the many options for handling anomalies available from termination to plastic major surgery it is again difficult to asses whether it is cost effective to detect an anomaly. Helsinki ultrasound trial (1996)[11] has shown that second trimester screening for anomalies by ultrasound is cost effective.

f. **How does prenatal anomaly scan for screening, influence infant health?**
Ultrasound screening is not primary prevention because it cannot prevent the anomaly, it can detect the problem and if the anomaly is lethal, it give the parents 'to be' an option to terminate pregnancy – secondary prevention. Also in many cases severe but curable defects (cardiac) can be managed by treating newborn without delay, if the pediatric surgery unit is prepared. Expertly performed prenatal ultrasound screening and autopsy reports corelate and provide accurate information.[12]

g. **What are the options after diagnosis of congenital malformations?**
The options for managing congenital malformation pregnancy have to be discussed with the parents 'to be' and the final choice lies with the parents. A team of specialists should provide all information and counseling. This team should consist of obstetrician, sonologist, geneticist, neonatologist, pediatric surgeon and a psychologist.

Options depend on severity of the anomaly and can be:

1. Termination of pregnancy.
2. Intrauterine treatment.
3. Maternal transport to tertiary care center.
4. Premature delivery.
5. Immediate specialized neonatal care.
6. Additional diagnostic tests.
7. Extensive monitoring.

h. **Alternatives or adjuncts to ultrasound?**
There are various blood tests like MSAFP, triple test, quadruple tests and many interventional procedures like CVS and amniocentesis, cordocentesis and fetal biopsy which can help to directly karyotype and chromosomally analyse the fetus but these are expensive, not easily available and carry a procedure related risk of miscarriage.

Noninvasive MRI is definitely not a cost effective method for screening.

Ultrasound advances have made this technology for screening an ideal test because it is:

- Relatively low cost
- Ease to perform
- Real time display
- Acceptable to all
- Widely available
- Accurate
- Safe
- Reproducible
- Available as office investigation
- Can now be applied for late first trimester also.[13]

i. **How long does it take?**
A primary screening ultrasound exam is a systemic analysis of fetal growth and fetal morphology system wise and will take 10 to 20 minutes to scan. The screening will stop if everything appears normal in all significant organs and structures (Figs 1 to 5).

Depending on image quality, maternal obesity, gestational age, type of anomaly, color Doppler or 3-D scan, still the total scan duration rarely exceeds 30 minutes. For subtle defects or solitary markers or inexperienced

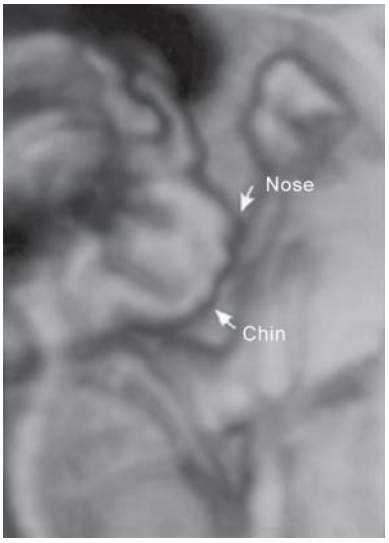

Fig. 1: Facial profile

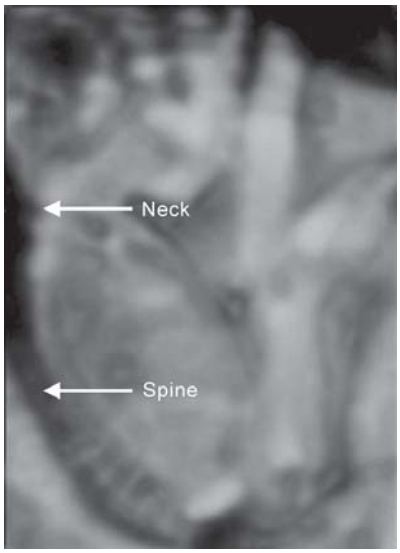

Fig. 2: Fetal profile

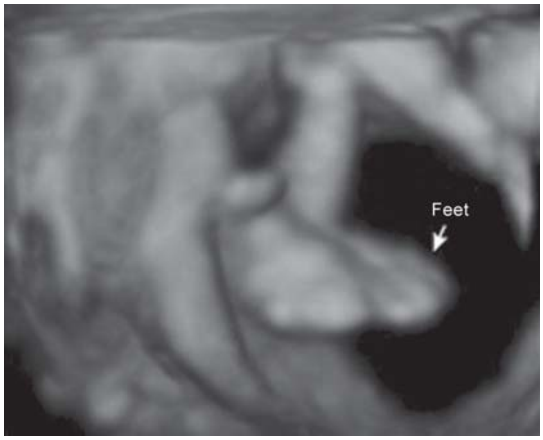

Fig. 3: Limbs

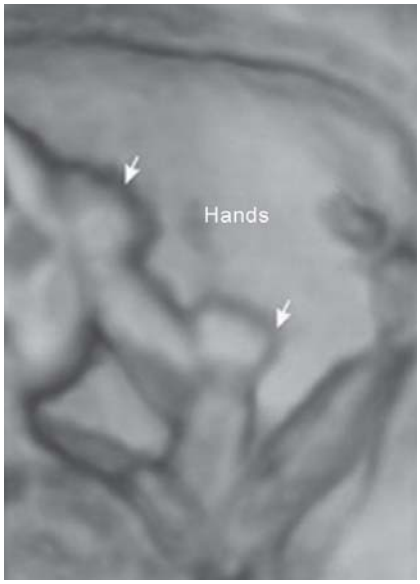

Fig. 4: Limbs

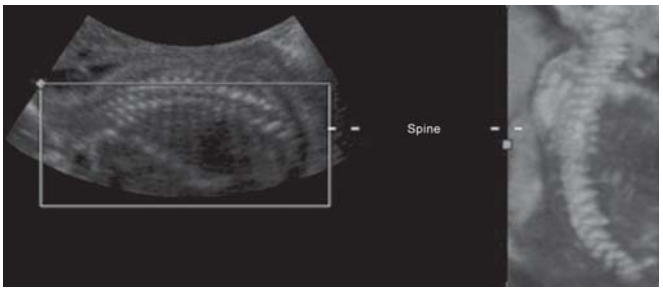

Fig. 5: 3-D spine

sonologists a second opinion scan might be required by an expert, which will take another 30 minutes.

j. **What does a prenatal ultrasound scan show**?

Depending on the gestational age the defects can be seen and identified, e.g. nuchal translucency in first trimester, duodenal atresia, GI defects, neural tube defects and some cardiac defects in second trimester.[14]

When we don't see the expected image of the fetus we suspect a defect. Sometimes we have to look for soft markers and signs of chromosomal anomalies, e.g. banana sign, lemon sign, etc.

Ultrasound can also pick-up functional abnormalities and abnormal fetal biophysical profile and abnormal fetal behaviors.

- Abnormal fetal activity
- Rapid uncoordinated fetal movements
- Fetal arrhythmia
- Fetal vomiting.
- Fetal GI stenosis

k. **When should a screening prenatal scan be done?**

Nicolaides has suggested a 11 to 14 weeks scan for screening for chromosomal anomalies, trisomy 21 by looking at the nuchal translucency and nasal bone ossification[14] other workers have suggested addition of biochemical markers.[15] The detection rate for trisomies varies from 80 to 89% with a false positive rate of 5% by using multiple markers study in first trimester scan (11 to 14 weeks).

A second trimester anomaly scan should be done between 18 to 22 weeks and a detailed fetal echocardiography and color Doppler uterine artery and ductus venosus should be done.

Third trimester screening should not be delayed more than 32 weeks gestation and is mainly done for growth and color Doppler studies for hypoxia detection. Late anomaly screening for GI and urinary tract anomaly is usually done at 32 weeks.

Ideal time for ultrasound screening for each and every gravida should be a monthly ultrasound but as this is not practical and feasible atleast each pregnancy should have two scans one 11 to 14 weeks scan and one second trimester scan.[16,17]

l. **Ultrasound : How sensitive it is for malformation detection?**

In a major study on 5,00,000 cases 11,000 (2.2%) were found

to be malformed fetus with a range of sensitivity from 14 to 80% (mean 45.5%)

In another study on 1,70,000 pregnant women, 4000 malformed fetus were detected with a sensitivity of 61%.[17]

m. **What counts as success in genetic counseling?**
Whenever anomaly is detected for some people abortion and termination of pregnancy is a matter of course response and no ethical dilemma arises, however, among certain religous groups objections to termination. Pose an ethical dilemma.

n. **Advances in fetal surgery?**
This option is still a research tool and there is an ethical aspect that many of these fetal surgical procedures are still experimental and of uncertain value and to give or not to give this option to couples carrying a malformed fetus is a dilemma.

o. **3-D and 4-D scans for screening are they useful or gimmicks?** There is now an increasing availability 3-D ultrasound. The benefits of 3-D/4-D are now a matter of debate. 3-D and 4-D help in maternal fetal bondage and also help for recognition and better confirmation of certain anomalies like cleft lips, polydactyly, micrognathia, malformed ears, club foot, vertebral malformations and other exterior surface anomalies. The development of TVS 3-D probes have further enhanced its value in early diagnosis of malformations.

p. **Reassurance scans – How reassuring?**
Proposed by Prof Stuart Campbell, a 3-D routine scan to reassure the parents and to rule out anomalies. But criticized as entertainment scans and consumerized for unprecedented profit marketing particularly after 4-D ultrasound.

CONCLUSION

To conclude a comprehensive state of art information on the routine obstetric ultrasound controversy and the ethical benefits of ultrasound screening has been presented. We conclude that a routine scan is warranted for each pregnancy but only if well performed at least level scan II. The scientific and economic benefit of such a scan will always remain a controversy but it is our ethical obligation to offer to a woman a methodology of prenatal diagnosis wherever it exists because in absence of prenatal diagnosis for malformations, many opportunities for optimal management can be lost. The psychological benefit of reassurance that the baby is normal is also of considerable value.

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