

EDITORIAL

Fetal Medicine*Aparna M. Kulkarni**Fetal Medicine Consultant, Deenanath Mangeshkar Hospital, Pune- 411004 (Maharashtra)
India*

Fetal medicine is a new upcoming super specialty branch. It deals with care of the 'un-born baby' where fetus is the patient and ultrasound scan is the main tool used to examine the fetus. As a fetal medicine specialist, our job is to reassure parents that 'their baby looks normal', at the same time we need to help parents take appropriate decision in difficult times.

Detailed ultrasound scan, counseling, screening for chromosomal abnormalities, assessment of growth of the fetus, decision of delivery at appropriate time in growth restricted fetuses, diagnostic procedures such as chorion villous biopsy, amniocentesis, cordoscentesis and therapeutic procedures such as fetal intrauterine blood transfusion form part of this specialty. With amazing advances in the technology and availability of advanced ultrasound scan machines and use of 3D and 4D modality, assessment of fetus has become easier than before.

Screening for common chromosomal abnormality has become an integral part of fetal medicine. The commonest chromosomal abnormality that is screened for is Down's syndrome (Trisomy 21). Along with it other chromosomal abnormalities that are screened for are Edwards' syndrome (Trisomy 18), Patau's syndrome (Trisomy 13) and Turner's syndrome. Incidence of Down's syndrome in India is around 1.4 / 1000 live births [1]. Although the risk of fetus to have Down's syndrome increases with maternal age, every pregnant woman is at a risk of having a child with Down's syndrome. Hence screening is offered to

everyone who is pregnant. Although, various screening tests are available, first Trimester combined screening test is the best test available with sensitivity > 90% when done appropriately. The test involves measurement of nuchal translucency (NT) and assessment of fetal nasal bone by ultrasound scan combined with mother's blood test, done between 11 weeks to 13 weeks 6 days of pregnancy [2, 3]. Appropriate training and certification of the person performing the NT scan is a prerequisite. Pretest and post test counseling are important aspects of any screening test, as the couple needs to understand why the screening is done and implications of a test if the results are screen positive.

Birth defects are known to cause perinatal mortality and morbidity. In India birth defects prevalence varies from 61 to 69.9/1000 live births and congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths [1, 4]. Due to availability and accessibility to healthcare facilities, it has been shown in developed countries, that around 70% of birth defects can be prevented or affected children can be offered care, which is likely to reduce severe disability. US reported 46% decline in the infant mortality rate from birth defects due to of improvement in diagnostic care and prevention [5].

First trimester (11-13 weeks 6 days) is also the time when we assess the fetus for major structural defects. By 11 weeks of gestation, fetal organogenesis is complete and more than 50% of major

structural defects can be identified by a detailed ultrasound scan done during this period. This scan involves a complete head to toe survey of the fetus [6]. Early identification of structural defect gives time for appropriate investigations and decision making in pregnancy.

Certain fetal structures especially fetal brain and heart are better evaluated later in gestation between 18-22 weeks of pregnancy, commonly known as level II fetal anomaly scan/malformation scan. This again involves a complete head to toe survey of the fetus. Nearly 80 % of structural defects can be picked up by a detailed scan during this time.

Some structural defects (eg talipes, PUJ obstruction, cleft lip or cleft palate, some cardiac defects) can be treated postnatally while cure for some defects may not be possible. A multi-disciplinary approach (depending on the type of abnormality) in management of pregnancy involving geneticist, pediatric surgeon, cardiologist, neonatologist, neurosurgeon, is necessary whenever any abnormality is found in the fetus.

Ultrasound scan plays an important role in identification of fetuses with restricted growth. Once a growth restricted fetus is identified, serial sonography and Colour Doppler evaluation helps in deciding the timing of the delivery. Timely intervention taking into consideration various factors such as maternal medical history (e.g. preeclampsia, diabetes etc), fetal activity and fetal behaviour, amount of amniotic fluid around the fetus, can help in avoiding iatrogenic premature delivery in growth restricted fetuses [7].

Colour Doppler evaluation is also helpful in identifying unwell/sick fetuses with severe anemia [8]. With rising incidence of multifetal pregnancy due to advances in assisted reproduction, ultrasound scan is the only modality that helps in identification of chorionicity in twin

gestation [9]. Management of twin gestation with a dichorionic placenta differs from the management of twin gestation with a monochorionic placentation. The role of 'Fetal Medicine Specialist' is to triage such pregnancies into low risk and high risk categories which subsequently helps in planning frequency of scans and identifications of certain complications that are known to be associated with monochorionic placentation [10].

Antenatal interventions are either diagnostic or therapeutic. Chorion villus sampling (done between 11-14 weeks) or amniocentesis (performed after 16 completed weeks) are common diagnostic procedures that are done to rule out or confirm fetal chromosomal abnormality or single gene disorders with known mutation. Prenatal diagnosis of hemoglobinopathy such as Thalassemia major is possible by either chorion villus sampling or amniocentesis. A clinical geneticist plays a crucial role in counseling and deciding need of prenatal diagnosis in parents with previous affected child.

Fetal intrauterine blood transfusion is the therapeutic intervention which is a lifesaving procedure in severe anemic fetuses. Severe anemia in a fetus is identified based on ultrasound scan findings and assessment of Middle Cerebral Artery Doppler Peak Systolic Velocity (MCA PSV) [8]. MCA PSV has 100% sensitivity in identification of significant fetal anemia (needing treatment or delivery) prior to 34 weeks of gestation. The transfusion procedure is done under continuous ultrasound guidance through either fetal intrahepatic portal vein or umbilical cord.

Assisted reproductive techniques are known to cause higher order multifetal pregnancies such as triplets, quadruplets, quintuplets and so on. Fetal reduction is a procedure that is offered to women

pregnant with higher order multiple pregnancies [11]. The procedure is done after 11 weeks when fetal organogenesis is complete. Careful selection of fetus / fetuses to be reduced is crucial. The aim of the procedure is to increase the chance of survival of remaining fetuses (usually twins) by reducing incidence of early preterm delivery (<32 weeks).

With advances in fetal medicine, Fetoscopic therapeutic intervention is being offered to monochorionic twin pregnancies complicated with Twin to twin transfusion syndrome (TTTS).

TTTS is a unique complication of monochorionic placentation due to unbalanced vascular anastomoses. This leads to one fetus (Donor) transfusing its blood to the other fetus (recipient) through placental vascular anastomoses. Fetoscopic LASER coagulation of placental anastomoses gives some hope to parents with pregnancy complicated by TTTS.

The journey in diagnosis and treatment of the fetus in utero has just begun. There is still more to explore in understanding the unborn fetus.

References

1. Rinku Sharma. Birth defects in India: Hidden truth, need for urgent attention. *Indian J Hum Genet* 2013; 19(2): 125–129.
2. Nicolaides KH. Screening for chromosomal defects. *Ultrasound in Obstetrics and Gynecology* 2003; 21(4): 2003, Pages: 313–321,
3. Bindra R, Heath V, Liao A, Spencerand K, Nicolaides KH. One-stop clinic for assessment of risk for trisomy 21 at 11–14 weeks: a prospective study of 15 030 pregnancies. *Ultrasound in Obstetrics & Gynecology* 2002; 20:219–225.
4. Shamnas M, Arya PS, Viji A Thottumkal, MG Deepak. Congenital anomalies: A major public health issue in India. *IJPCBS* 2013, 3(3), 577-585
5. Christianson AL, Howson CP, Modell B. White Plains. New York, USA: March of Dimes Birth Defects Foundation; 2006. [Last accessed on 2012 Feb 24]. March of Dimes Global report on birth defects: The hidden toll of dying and disabled children.
6. ISUOG Practice Guidelines: Performance of first-trimester fetal ultrasound scan. *Ultrasound Obstet Gynecol* 2013; 41: 102–113
7. Ferrazzi E, Bozzo M, Rigano S, Bellotti M, Morabito A, Pardi G, Battaglia FC and Galan HL. Temporal sequence of abnormal Doppler changes in the peripheral and central circulatory systems of the severely growth-restricted fetus. *Ultrasound in Obstetrics & Gynecology* 2002; 19 (2):140–146.
8. Mari G. Middle cerebral artery peak systolic velocity for the diagnosis of fetal anemia: the untold story. *Ultrasound in Obstetrics & Gynecology* 2005; 25 (4): 323–330.
9. Dias T *et al.* First-trimester ultrasound determination of chorionicity in twin pregnancy. *Ultrasound in Obstetrics & Gynecology* 2011; 38(5):530-2.
10. Lewi *et al.* Monochorionic Diamniotic Twins: Complications and management options. *Current Opinion in Obstetrics & Gynecology* 2003; 15(2):177-194.
11. Evans MI, Britt DW. Fetal Reduction. *Seminars in Perinatology* 2005; 29(5) 321-329.

Author for Correspondence: Dr. Aparna Kulkarni, Fetal Medicine Consultant, Deenanath Mangeshkar Hospital, Pune- 411004 (Maharashtra) India Cell: 9922429088 Email: amkulkarni21@gmail.com