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Principles and Practice of

Fetal Medicine



Raju R Sahetya
Jaideep Malhotra
Hema Purandarey

Foreword
Rustom P Soonawala

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CHAPTER

2 Multidisciplinary Approach for Prenatal Screening and Diagnosis

Apoorva Pallam Reddy, Narendra Malhotra, Jaideep Malhotra

INTRODUCTION

The exhilaration at the thought of being a mother is more often than not plagued by the worries of the well being and normalcy of the fetus. "Is my baby doing alright?" is the most common question fired at any obstetrician. In spite of the extensive line up of investigations available, there are always asterisk (*) signs saying that 'all risks cannot be ruled out.'

Apart from the most frequently occurring fetal aneuploidies like trisomies 21, 18, and 13 there are many more structural defects that can be detected by prenatal screening. Various congenital cardiac anomalies, abnormalities of the chest and gastrointestinal tract, urinary tract abnormalities, anomalies of the central nervous system, externally visible body defects and skeletal abnormalities can be detected by antenatal ultrasonography.

The diagnosis of these anomalies is essential not only to detect the fetuses that are not viable but more importantly identify the ones that need extra support for viability. Under such circumstances having a multidisciplinary approach that involves adequate counseling and active involvement of the parents in their baby's care is the only way forward.

In this chapter, we discuss about all the members that need to come together to form an ideal prenatal management team and regarding their role in improving the fetal outcome. (Flow chart 2.1).

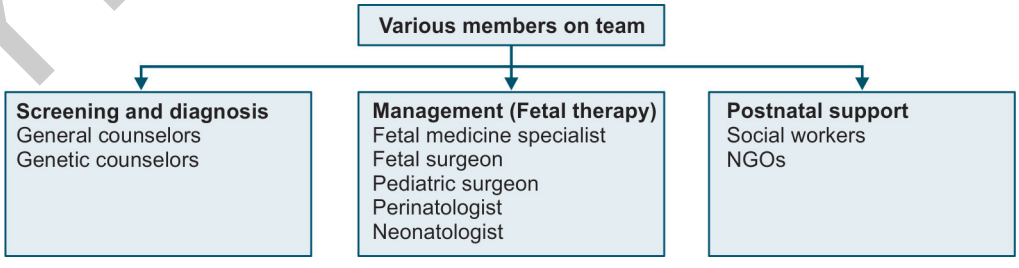
PRENATAL SCREENING COUNSELOR

Prenatal screening should not be considered as yet another routine antenatal investigation, added to the pre-existing long list of tests, as the results can potentially change the course of pregnancy. It is important for health care professionals to both ***offer*** the screening tests available and ***discuss*** their implicatiosn with all pregnant women. Information pamphlets, in their native language, can also be helpful in explaining the purpose and limitations of the tests. The decision for screening is entirely voluntary and should be documented.

Tips for Prescreening Counseling[1]

- Counsel your patient that all pregnant women have some risk of having a fetus affected by trisomy 21, 18, or 13 or other structural defects.
- Explain that prenatal screening will use specific maternal and pregnancy factors (e.g. age, ethnicity) to assess her individual risk of having an affected fetus with maternal age being an important effector for risk.
- Emphasize that a positive screen will mean that her risk is above a predetermined cutoff (set by regional programs), not that her fetus is necessarily affected. Distinguish clearly between a screening test and a diagnostic test.

Flow chart 2.1 Essential members of the multidisciplinary team for prenatal screening and management



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- Give simple examples while explaining the specific and personalized risk. For example, a risk of 1:100 means that for one woman carrying an affected fetus, there are 99 women carrying a fetus that is not affected.
- Discuss options and the invasive prenatal testing that may need to be considered after screening.
- *Screen negative:* Explain that if screening is negative, no further testing is indicated other than routine second trimester ultrasound screening but does not guarantee the birth of a healthy baby.
- *Screen positive:* Inform your patient that if the screening test is positive she is eligible for diagnostic testing.
- *Diagnostic testing:* Ensure your patient is aware that diagnostic testing is entirely voluntary. This testing would be done by amniocentesis in 2nd trimester or late 1st trimester, which carries a procedural risk of 1/100 to 1/175 for loss of the pregnancy or by chorionic villus sampling, in early 1st trimester which carries a procedural risk of 1/100 for loss of the pregnancy.
- *Decide for or against termination:* Ensure your patient knows that she has the option of continuing or terminating the pregnancy, if the test shows that her fetus has an aneuploidy or another significant chromosomal anomaly. It is a common misconception that screening is offered only to patients who would terminate a pregnancy if an anomaly were found. It is important to tell patients who may choose to continue the pregnancy that their fetus will be given the best care possible.
- In the case of chromosomal anomalies that have an extremely poor prognosis, this may include referring the patient to a neonatologist to discuss palliative.

GENETIC COUNSELORS[2]

Genetic counselors work as part of a healthcare team, providing information and support to families affected by or at risk of a genetic disorder. They help to identify families at possible risk of a genetic disorder, gather and analyze family history and inheritance patterns, calculate risks of recurrence, and provide information about genetic testing and related procedures.

Genetic counselors also provide supportive counseling services, serve as patient advocates, and refer individuals and families to other health professionals and community or state support services. They serve as a central resource of information about genetic disorders for other healthcare professionals, patients, and the general public.

The most common indications for genetic counseling include advanced maternal age, family history of a genetic condition, and suspected diagnosis of a genetic condition.

The post-test session involves more than the provision of medical information and often focuses on helping families cope with the emotional, psychological, medical, social, and economic consequences of the test results. In particular, psychological issues such as denial, anxiety, anger, grief, guilt, or blame are addressed and, when necessary, referrals for in-depth counseling are offered. Information about community resources and support groups are provided to the patient/family.

If the genetic test is positive, testing should be considered in additional relatives of this individual.

At the conclusion of the genetic counseling sessions, the patient should be offered a written summary of the major topics discussed. The summary is often provided in the form of a letter which serves as a permanent record of the relevant information discussed, as well as relaying additional information that may have become available after the final counseling session.

Fetal Therapy

Fetal therapy was introduced as early as 1952 by Brevis as amniocentesis for management of rhesus isoimmunization and is currently a very well-established field.

Definition

A therapeutic intervention for the purpose of correcting or treating a fetal anomaly or condition. In almost every case, the fetus is at risk of intrauterine death from the abnormality. Advances in fetal therapy have enabled us to prevent and treat potentially life-threatening conditions and improve outcome.

FETAL MEDICINE PHYSICIAN

Fetal medicine specialists use ultrasound, blood tests, and procedures such as fetoscopy to evaluate the developing fetus.

They perform diagnostic procedures like chorionic villous sampling, amniocentesis and fetal blood sampling as well as therapeutic interventions like laser ablation of AV anastomosis in twin to twin transfusion syndrome, twin reversed arterial perfusion syndrome or selective fetal reduction of the anomalous twin.

Tracheal occlusion in fetuses with congenital diaphragmatic hernia, aims at blocking the outlet of the lung-fluid, which accumulates within the lungs increasing the transpulmonary pressure, promoting lung growth. Percutaneous fetal endoscopic balloon tracheal occlusion (FETO) is being evaluated for the same. Fetuses with tracheal occlusion must be delivered by EXIT procedure (partial delivery of the fetus, removal of the tracheal occlusion,

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administration of surfactant and institution of assisted ventilation while the infant is still on placental support).

In very severe cases of erythroblastosis fetalis fetal intrauterine transfusion can be performed to treat the hemolytic anemia. A pediatric hematologist can be involved in decision making.

FETAL SURGEON[3]

Rare fetal tumors like congenital cystic adenomatoid malformation of the lung (CCAM) and sacrococcygeal teratoma (SCT) cause almost 100% fetal mortality when associated with hydrops fetalis. These tumors cause hydrops by either venous obstruction due to mediastinal shift (CCAM) or high output heart failure (SCT). Operative removal of these tumors has resulted in survival of ~50% of the affected fetuses.

Intrauterine repair of myelomeningocele decreases the incidence of hindbrain herniation and shunt-dependent hydrocephalus in infants with spina bifida, but increases the incidence of premature delivery [Management of Myelomeningocele Study (MOMS)].

Aspiration of fetal ovarian cysts larger than 30 mm to prevent torsion and avoid neonatal surgery is being evaluated in France.

PEDIATRIC SURGEON

- *Diaphragmatic hernia:* The outlook for babies born with CDH is increasingly positive with new surgical techniques and ways to support babies as they heal. Surgery is performed after delivery as soon as the lungs can maintain good blood flow through the vessels prior to the operation.
- *Duodenal atresia:* Duodenal atresia is often characterized by a "double bubble" on prenatal ultrasound. One bubble represents air in the stomach and the other bubble represents the first part of the duodenum, with no air past that. Surgery is required to treat duodenal atresia, usually within the first few days of life.
- *Esophageal atresia:* VACTERL association because of the possible involvement of vertebral column, anorectal, cardiac, tracheal, esophageal, renal (kidney), and limbs require early management of neonate for better prognosis.
- Management of nonfatal cosmetic conditions like cleft lip, cleft palate, congenital tallipes should be discussed clearly as soon as the diagnosis is made.

PERINATOLOGIST AND NEONATOLOGIST

In cases where antenatal screening has not been done or has not picked an anomaly, pediatricians often face the responsibility of revealing the diagnosis to the parents and dealing with the emotional overture. They also have to deal with complications in the neonatal period (IUGR, congenital anomalies) and various comorbidities (hypothyroidism, recurrent otitis media, atlantoaxial instability, transient myeloproliferative disorders). The perinatologist should be involved in the decision making of mode of delivery for anomalous fetuses along with the couple.

SOCIAL WORKERS AND SUPPORT GROUPS[4]

Despite the best efforts and elaborate investigations 1 in 1000 live births result in a Downs baby. For a couple waiting to hold their newborn close, the idea of an 'abnormal' fetus is always shattering. Lack of knowledge on the routine management and anticipated medical problems only compounds the emotional turmoil. Thankfully there are a lot of Down's syndrome support groups both nationally and internationally helping many Downs babies lead a life as close to normalcy as possible. A case report of a 25-years-old married Downs lady delivering a healthy term baby only emphasizes the importance of familial and social support.

NONVIABLE TO VIABLE: YES WE CAN!!

Since 2012, donation possibilities for babies, specifically babies with anencephaly, have expanded and currently there are more potential for donation than ever before. Although each particular case is unique, and each potential donation has various criteria that must be met, donation is a very viable option for a baby with anencephaly, if families wish to pursue this option. Donation can be used either for transplant or research.

The recent story of Teddy, the anencephalic twin, donating kidney and heart valves to an adult suffering from renal failure is no less than a heroic act.

Currently, India does not have any legislation over organ donation in anencephalus fetuses. With an extremely poor national organ donation rate of 0.26 per million population, it is indeed essential to tap into the possibility of organ donation from anomalous fetuses.

Where Do You, As An Obstetrician, Stand in All This?

Just like the conductor of a symphony orchestra unifies the performers and executes clear preparations to shape the sound of the ensemble, the obstetrician plays the key role of coordinating all the events in prenatal screening, diagnosis and management and bringing out the entire team as one unit.

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CONCLUSION

A multidisciplinary yet integrated approach for prenatal screening and diagnosis with expert counseling and close follow-up after delivery for appropriate medical support not only improves the chance of healthy viable fetus but also decreases the anxiety levels for the parents.

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Salient Features

- The field of prenatal diagnosis is rapidly expanding. Because of this, we have made every effort to include the most recent, up-to-date knowledge available.
- Major source of reference and guidance to a wide range of professionals.
- Useful for the Obstetricians, Geneticists and Genetic Counselors, Ultrasonologists, Pediatricians and the laboratory people, to provide information, guidance and opinions to prospective parents on prenatal diagnosis.
- The book is divided into 10 sections:
  - *General Aspects of Prenatal Diagnosis:* Revolution in Prenatal Diagnosis, Multidisciplinary Approach for Prenatal Screening and Diagnosis, Goals and Issues in the Delivery of Genetic Services in India, Prenatal Diagnosis: Challenges and Opportunities Obstetrician Perspective
  - *Prenatal Genetic Counseling:* General Counseling, Premarital and Preconception Counseling, Genetic Counseling in Prenatally Diagnosed Chromosomal Abnormalities, Preconceptional Counseling and Management, Role of Genetics in Reproductive Failure, Genetics and Counseling of Female Hereditary Breast and Ovarian Cancers
  - *Prenatal Screening—Noninvasive:* Global Rainbow Healthcare, First Trimester Screening, Maternal Serum Screening for Chromosomal Abnormalities, Newborn Screening, Screening and Diagnosis of Inborn Errors of Metabolism, 11 to 13+6 Weeks Scan: A Window to Fetal Abnormalities, Fetal Anomaly Scan, Fetal Growth Restriction, Color Doppler in Fetal Growth Restriction
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  - *The Future of Prenatal Diagnosis:* Current Status of Fetal Genomic Diagnosis in Fetal Medicine, Progress and Challenges, Array-based Comparative Genomic Hybridization: Future in Prenatal Diagnosis, Future of Prenatal Diagnosis and Screening
  - *Guidelines:* FIGO: Good Practice Advice, Maternal-Fetal Medicine



Raju R Sahetya
MD DGO DFP FCPS FICOG
Mumbai, Maharashtra,
India



Jaideep Malhotra
MD DGO FICOG
Agra, Uttar Pradesh,
India



Hema Purandarey MS
Mumbai, Maharashtra,
India

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