

A review of the management of prenatally detected fetal anomalies: the need for structured evaluation and a multidisciplinary approach

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ABSTRACT

The evaluation and management of prenatally detected fetal anomalies continue to be a challenging problem. The judicious use of screening tests, determination of severity of a particular disorder and the decision-making regarding prenatal and perinatal management pose unique difficulties. This narrative review aims to address the critical aspects with regard to prenatally detected fetal anomalies with respect to their diagnosis, evaluation and management. They are presented in the light of clinical experience and observations with regard to clinical management of these disorders. The key information pertaining to the clinical use of prenatal diagnostic modalities, maternal screening and prenatal and perinatal management are summarized here. The wide spectrum of fetal anomalies range from physiological phenomena of uncertain significance to complex, lethal anomalies. The practical issue with the clinical management of a pregnancy complicated by fetal anomaly lies in the lack of universally accepted guidelines in management. The use of invasive fetal diagnosis, fetal therapy and termination of pregnancy for fetal anomalies continue to be matters of medical, social and ethical concern. The management of prenatally detected fetal anomalies should follow a structured and scientific approach to yield optimal results. The prenatal, perinatal and postnatal management can be thus streamlined. Establishment of standard clinical guidelines, multidisciplinary teams and specialized centers will help to optimize the management of fetal anomalies.

Keywords: Prenatal diagnosis, fetal anomaly, maternal screening, fetal therapy, perinatal management

INTRODUCTION

The evaluation and management of prenatally detected fetal anomalies continue to be a challenging problem. The determination of severity of a particular disorder, selection of the appropriate tests for evaluation and institution of the most appropriate management in a particular scenario require consideration of multiple factors. The new and evolving methods of genetic testing, invasive fetal diagnostic techniques and advances in fetal imaging have contributed to an early and more precise diagnosis. But these advancements are also complicated by concerns regarding their interpretation, significance and appropriate, scientific use.

The practical issue with the clinical management of a pregnancy complicated by fetal anomaly lies in the lack of universally accepted guidelines in the diagnosis, evaluation and therapy. The wide spectrum of these disorders range from physiological phenomena of uncertain significance

to complex, lethal anomalies, that can not be fitted under a single umbrella. The use of invasive fetal diagnosis, fetal therapy and termination of pregnancy for fetal anomalies (TOPFA) continue to be matters of medical, social and ethical concern. Moreover, the best possible outcome ideally requires the input of multiple specialists, a scientific and structured approach and multidisciplinary teams. These facts point to the need to establish universal standard treatment guidelines and clinical practice principles. There is also a need for institution of medical boards and specialized centers to streamline the management of fetal anomalies.

Aims

This article aims to review the various aspects of the diagnosis, evaluation and management of prenatally detected fetal anomalies. The essential objective is to provide a comprehensive overview of prenatal screening, fetal therapy,



prenatal and perinatal management. The article also aims to propose future directions towards standardization of the practice of fetal medicine.

Methods

A narrative review of the present literature, clinical guidelines and practice recommendations is performed here in the light of clinical experience and observations with regard to clinical management of these variegated group of disorders. Current literature including PubMed, Scopus, Embase and Medline database articles, clinical guidelines and practice recommendations from 2004-2024 were reviewed. The focus mainly rested on articles pertaining to the 'prenatal diagnosis', 'TOPFA', 'fetal therapy' and evaluation and management of 'fetal anomalies'. An attempt has been made to synthesize and organize the said data about various aspects of fetal anomalies for easy reference. The key observations pertaining to the clinical application of these principles are also summarized here.

Main Text

When faced with the issue of treatment of fetal anomalies, there are numerous concerns to be addressed by the concerned specialists. These pertinent factors with regard to diagnosis, evaluation and management of fetal anomalies are summarized in the **Table 1**. The various aspects of this subject is discussed here under the following headings:

- The optimal use of presently available maternal screening tests
- The use of invasive fetal diagnosis, fetal therapy and EXIT procedure
- The decision-making and communication with regard to fetal anomalies
- The perinatal management with respect to fetal anomalies
- Fetal anomalies and medical termination of pregnancy (MTP)
- The need for a multidisciplinary and integrated approach to management
- The need for specialized organizational entities for fetal anomalies

THE OPTIMAL USE OF PRESENTLY AVAILABLE MATERNAL SCREENING TESTS

First trimester screening is performed with maternal serum biomarkers, pregnancy-associated plasma protein A (PAPP-A) and free beta-human chorionic gonadotropin (hCG). Second trimester screening is performed with alfa fetoprotein (AFP), hCG, estriol, and inhibin-A, the "quad screen". These are useful in the detection of chromosomal disorders, neural tube defects (NTD) etc. Carrier genetic screening is also available to detect inherited disorders for high-risk cases (pre-implantation genetic testing).¹⁻³

Cell free fetal DNA study by isolation of fetal cells in the maternal circulation using advanced sorting techniques permit genetic testing from a maternal blood sample. It can be done from 10 weeks of pregnancy and beyond. This can be used for the screening of trisomy 21, 18, 13 and sex-chromosome aneuploidy. This is only a screening tool and a positive cell-free DNA test result should be followed by a diagnostic test with amniocentesis or chorionic villus sampling (CVS). The high cost factor, technical complexity, the lack of standardization and non-uniform insurance cover for testing limit the widespread adoption of this test.⁴⁻⁷

Prenatal genetic testing: Prenatal genetic testing techniques to detect aneuploidy include QF-PCR and FISH for rapid aneuploidy testing, G-banded karyotyping and chromosomal microarray analysis (CMA). The adoption of next generation sequencing (NGS) techniques in prenatal diagnostics, including whole exome sequencing (WES) and whole genome sequencing (WGS) enable a much greater diagnostic yield.⁸⁻¹¹

The prenatal diagnosis of a lethal genetic disorder enables appropriate prenatal, perinatal and postnatal management, and also termination of pregnancy where indicated. A proper genetic diagnosis also permits counselling about the risk of recurrence and facilitates prenatal testing or pre-implantation genetic diagnosis in subsequent pregnancies.⁹⁻¹¹

Prenatal genetic screening (serum screening with or without nuchal translucency (NT) US/ cell-free DNA screening) and diagnostic testing (CVS or amniocentesis) options should be discussed with all parents during pregnancy, regardless of maternal age or risk of chromosomal abnormality. The

Table 1. The 'six pertinent questions each' with regard to the prenatal diagnosis, evaluation and therapy of a pregnancy complicated by fetal anomaly

Prenatal diagnosis	Prenatal evaluation	Prenatal and perinatal therapy
The need for carrier screening and use of maternal screening tests	The reliability of the diagnosis and the need for confirmatory testing	Indication for non-invasive/ medical therapy (steroids for fetal lung maturation)
The nature of anomaly: developmental aberration/ significant defect	The need for only routine scans/use of serial testing	Indication for invasive fetal therapy: endoscopic/ surgical
Severity of the anomaly and grading of the defect	The need for detailed fetal evaluation (fetal echocardiogram, fetal MRI etc.)	Indication for EXIT procedure
Isolated defect or part of a complex association	The need for invasive prenatal diagnosis (amniocentesis, CVS etc.)	Indication for termination of pregnancy for fetal anomaly
Likely natural history, progression and prognosis	The need for invasive fetal testing (fetal cord blood or urine sampling etc.)	Indication for referral to a dedicated perinatal centre
Likely complications: prenatal, perinatal and postnatal/fetal (hydrops fetalis, IUGR etc.), maternal (PIH) and gestational (oligohydramnios)	Maternal evaluation for specific risk factors like pre-eclampsia.	Indication for change in perinatal therapy: preterm delivery/caesarean section

MRI: Magnetic resonance imaging, CVS: Chorionic villus sampling



parents retain the right to pursue or decline prenatal genetic screening and diagnostic testing, after proper counselling. If screening is accepted, patients should have one prenatal screening approach, and should not have multiple screening tests performed simultaneously. The sensitivity of first trimester and second trimester screening tests are 80-84% and 80-82% respectively. The specificity of first trimester and second trimester screening tests are both about 95%.^{9,11-13}

First trimester ultrasound scan (US) at 11 to 13 weeks is very useful as it aids in precise dating, detection of twin chorionicity, and the early detection of major structural abnormalities. Nuchal translucency (NT) measurement on US at 11 to 14 weeks is a marker for fetal chromosomal disorders, NTD, structural anomalies (cardiac) and genetic disorders.¹⁴⁻¹⁶

Second trimester US is vital to assess major defects of the heart, brain and spine, kidneys, abdomen, craniofacial region, limbs etc. This is to be done at 18 to 22 weeks of pregnancy. Detailed fetal imaging using 4D US, fetal echocardiogram, fetal MRI and serial US are indicated in complex anomalies.¹⁴⁻¹⁷

Parents with a positive screening test result for fetal aneuploidy should undergo genetic counselling and a comprehensive US evaluation with an opportunity for diagnostic testing (CVS, amniocentesis) to confirm results. Parents with a negative screening test result should be informed that the result indicates significantly lower risk of the specific aneuploidy. But this does not fully ascertain that the fetus is unaffected. Fetal structural defects may occur with or without fetal aneuploidy. Hence all patients should be offered a second-trimester US between 18 and 22 weeks of gestation (with or without second-trimester maternal serum AFP).^{1,8,11,15,17}

THE USE OF INVASIVE FETAL DIAGNOSIS, FETAL THERAPY AND EXIT PROCEDURE

Invasive fetal diagnosis relies on CVS in the first trimester and amniocentesis in the second trimester. These are selectively

used in the case of a positive screening test and also in high-risk cases, for the sake of a definitive diagnosis. These are complicated by the attendant risks to the mother and fetus. CVS is mainly used for diagnosis of genetic disorders. Amniocentesis aids in the evaluation of chromosomal, biochemical, histopathological, and infective disorders.¹⁸⁻²¹ The common prenatal investigations, their ideal timing and their general significance are summarized in the **Table 2**.

Fetal therapy: Most of the advanced fetal therapy is still experimental and generally conducted only as a part of clinical trials, in selected high-risk cases. The benefits of intervention should outweigh the risks posed to the mother and fetus. Fetal endoscopy (FETENDO), fetal transfusion and open surgery are used in select cases. The maternal administration of drugs like steroids benefit the fetus in certain situations like lung disorders.^{2,16,18,21}

EXIT procedure is reserved for fetal anomalies that benefit from intrapartum intervention. These include conditions like congenital high airway obstruction syndrome (CHAOS) that mandate early airway access and conditions requiring immediate cannulation for ECMO. The EXIT procedure is associated with a higher maternal risk than a caesarean. There are also considerable logistical challenges due to the need for multidisciplinary involvement. The decision regarding the use of invasive fetal diagnosis, fetal therapy and EXIT procedure need careful risk-benefit assessment. This decision should be taken by a team involving all the concerned specialists.^{2,16,19,21}

THE DECISION-MAKING AND COMMUNICATION WITH REGARD TO FETAL ANOMALIES

The salient points in the maternal history with relevance to fetal anomaly include the following points: a. history of consanguinity, b. affected siblings, c. previous fetal loss, d. advanced maternal age, e. treatment for infertility, f. bad obstetric history, g. maternal illness/carrier state, h. history of genetic disorder and i. risk of recurrence of anomaly in the subsequent pregnancies. These should be surveyed, analysed and documented. In case of detection of multiple anomalies,

Table 2. Summary of the common prenatal investigations, their timing and general significance

Prenatal testing	The investigation	Significance
Carrier testing	Genetic testing done before pregnancy.	Detection of inherited disorders in high risk cases
Prenatal genetic testing to detect aneuploidy	QF-PCR and FISH, G-banded karyotyping CMA, NGS: WES and WGS	Diagnosis of genetic defects
First trimester screening	Maternal serum biomarkers, PAPP-A and free beta hCG	Detection of chromosomal disorders, neural tube defects, nuchal translucency measurement on US etc.
Cell free fetal DNA testing	Genetic testing from maternal blood sample from 10 weeks and beyond	Used for the screening of trisomy 21, 18, 13 and sex-chromosome aneuploidy.
First trimester scan	Ultrasound at 11-13 weeks	Precise dating, detection of twin chorionicity, and early detection of major structural anomalies.
Second trimester screening	Quad screen with AFP, hCG, estriol, and inhibin-A	Detection of chromosomal disorders, neural tube defects etc.
Second trimester scan	Ultrasound at 18-22 weeks	Assess major defects of the heart, brain and spine, kidneys, abdomen, craniofacial region, limbs etc.
Fetal diagnostic testing	Chorionic villus sampling in first trimester and amniocentesis in the second trimester	Selectively used for a positive screening test and high-risk cases for definitive diagnosis
Fetal sampling	Fetal cord blood sampling fetal urine sampling	Diagnosis of TORCH infections, prognostication PUV etc.

PAPP-A: Pregnancy associated plasma protein A, hCG: Human chorionic gonadotropin, QF-PCR: Quantitative fluorescence polymerase chain reaction, FISH: Fluorescence in situ hybridization, CMA: Chromosomal microarray, NGS: Next generation sequencing, WES: Whole exome sequencing, WGS: Whole genome sequencing, PUV: Posterior urethral valves



the management decisions should be based on the major issue with the most significant bearing on the outcome and issues that affect perinatal therapy (major cardiac defects).^{2,16,19-21}

The parents should receive education, counselling and psychological support during the prenatal and postnatal period. The communication should be compassionate, empathetic and open. The objective is to help them understand the situation and arrive at an informed decision. The important factors with regard to this communication pertain to the following points: a. nature of the anomaly and its severity, b. the likely prognosis and outcome, c. the need for further investigations, d. the option of TOPFA, where indicated, e. the need for invasive diagnostic/therapeutic modalities and, f. the need for change in mode, time or venue of delivery.^{2,16,19-21}

The obstetrician is usually the first contact of parents in case of prenatally detected anomalies. They should refer the mother to the appropriate center for the fetal specialist to arrive at a diagnosis, assess the prognosis and decide on the need for further evaluation. The initial communication regarding the possible nature of the anomaly and management should ideally be made by the fetal specialist. The sonologist/obstetrician may not have received specific training in fetal medicine to provide a definite diagnosis and prognosis. The management plan should be formulated by the team of specialists. A continuous and dynamic interaction should be maintained between the obstetrician and the fetal specialist during the follow up and serial re-evaluation. This enables to update decisions regarding antenatal and perinatal management, keeping the parents abreast with the progress.^{12,13,16,19-21}

THE PERINATAL MANAGEMENT WITH RESPECT TO FETAL ANOMALIES

After the diagnosis of a fetal anomaly, the appropriate perinatal management may require transfer to a specialised centre/involvement of multiple specialists. Though broad guidelines can be followed in the management, decision-making in each case of fetal anomaly has to be individualised. The ideal timing and mode of delivery are also to be decided upon based on risk-benefit assessment. Transporting a pregnant woman before delivery to a hospital that can provide the necessary level of care is a much safer option than transferring an unstable neonate who requires urgent intervention in the immediate postnatal period.²²⁻²⁵

Anomalies in the fetus can affect gestation and vice versa, highlighting the vital need for close interaction and continued communication between the obstetrician and the fetal specialist during the follow up. Lack of such coordination and planned decision-making exposes both the mother and the child to avoidable stress and complications.¹¹⁻¹⁵ The timing and mode of delivery are important considerations in all major fetal anomalies. But these are especially vital in conditions characterized by a complicated perinatal period like cardiac defects, congenital diaphragmatic hernia (CDH), oesophageal atresia, abdominal wall defects, NTD etc. Spontaneous vaginal delivery should be the primary

option in most fetal anomalies. This is associated with lowest maternal morbidity and mortality.²⁴⁻²⁷

In case of lethal anomalies that have not undergone TOPFA, and have reached late gestation, term vaginal delivery is to be preferred, as this is the safest option for the mother. Caesarean delivery is generally indicated if a fetal anomaly is associated with the risk of dystocia, bleeding, or disruption of a protective sac. Examples of the same include anomalies like large exomphalos, severe hydrocephalus, large myelomeningocele (MMC), and teratomas.^{22,24-26}

In many situations with prenatal diagnosis of fetal anomalies, there is a tendency for Obstetricians to proceed with delivery immediately after completion of 36 weeks. But it is ideal to wait for delivery till 38-39 weeks to optimise the risk factors, unless gestational factors supervene. It should be remembered that the fetus in utero is at lesser risk and better protected than a preterm neonate in the intensive care unit. The fact that mortality and morbidity in preterm neonates with anomalies is higher than term neonates is worth consideration here. Due to the severe risks related to it, preterm delivery should be reserved for few selected cases of fetal anomalies, the most common indication being the worsening of fetal status.^{22,23,25,27}

Planned labour is of utmost importance in a neonate with a major structural anomaly (CDH, oesophageal atresia, cardiac defects, abdominal wall defects etc.). If an unplanned delivery occurs outside regular hours in these conditions, the well-being of the neonate is critically endangered. This is primarily due to logistical constraints pertaining to the availability of neonatologists and paediatric surgeons. Planned induction of labour is also beneficial for mothers who reside far away from a tertiary care centre. A broad outline of the evaluation and management pathways in pregnancy complicated by fetal anomalies is summarised in the **Figure**.^{23,26-28}

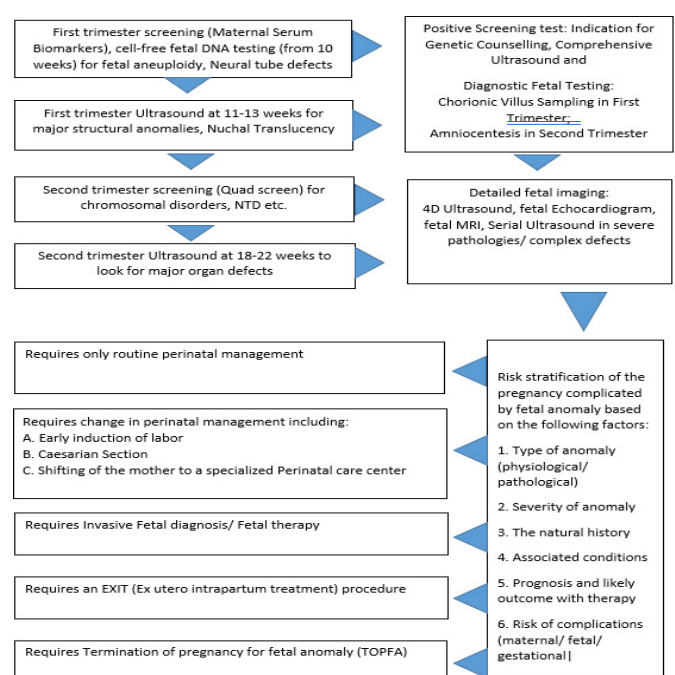


Figure. A broad and general outline of the pathway for evaluation and management in pregnancy complicated by fetal anomalies



FETAL ANOMALIES AND MEDICAL TERMINATION OF PREGNANCY (MTP)

Widespread use and advancements in prenatal diagnostic techniques have resulted in increased detection of anomalies, with an increase in demand for TOPFA. In case of major anomalies that are detected in the first trimester (anencephaly, pulmonary agenesis, body stalk anomaly etc.), TOPFA should be considered at the earliest. The relaxation in the legal gestational age limits for TOPFA in various countries has eliminated an important barrier in this regard. There have also been directions for a medical board to be established for special permission with regard to MTP in specific situations. In many countries, the gestational upper limit has been increased to 24 weeks and beyond 24 weeks, with the permission of the medical board, with no gestational upper limit in the cases of major fetal congenital malformations. Fetal anomalies may sometimes be recognised only after 20 weeks of gestation and continuation of such a pregnancy could be detrimental to the mother and the foetus. In such situations, the woman can approach the medical board for an MTP.^{2,16,29-32}

Delay in diagnosis of a fetal anomaly due to factors like delay in seeking antenatal care, delayed diagnosis, irregular follow-up, inadequate counselling and late referral are the major reasons for the delay in the decision for abortion. Abortion-related complications increase with gestational age. Also, the pregnancy termination late in gestation has the inherent risk for the occurrence of a live birth. This can result in great anguish for parents and also become a severe drain on healthcare resources.³⁰⁻³² Hence, in late gestation, a feticide procedure may be required for prevention of occurrence of a live birth. Feticide must be considered before MTP for gestation that has progressed beyond 24 weeks. It is the responsibility of the health care sector to facilitate access to early and effective prenatal diagnosis. This would help to reduce the number of cases where TOPFA needs to be considered late in the gestation.^{19-21,29-32}

THE NEED FOR A MULTIDISCIPLINARY AND INTEGRATED APPROACH TO MANAGEMENT

The essential objective in the case of prenatal detection of a fetal anomaly is to provide the pregnant woman with comprehensive specialized care. A fetal specialist (maternal-fetal medicine specialist/paediatric surgeon) confirms the anomaly and determines the prognosis and provides therapeutic options. The opinion regarding severity and prognosis of a fetal anomaly can be organ-specific and may need the input of multiple specialists.³³⁻³⁶

The paediatric and neonatal surgeon is in a pivotal position to opine regarding the diagnosis, prognosis and therapy with regard to most of the surgically treated structural anomalies. The opinion of other specialists should be integrated in the decision-making process. The use of technology for image sharing and collaboration can help to overcome the logistical challenges of garnering multiple opinions. It is imperative that the fetal specialist/paediatric surgeon be abreast with all the new diagnostic/therapeutic techniques, clinical

guidelines and the current medico legal developments in this regard.^{2,16,32-36}

The input of other specialists like cardiology, neurology, nephrology, genetics, orthopedics and paediatrics is vital in certain specific disorders involving specific organ systems. The team of specialists should decide on the evaluation, frequency of follow-up, the mode, timing and venue of the delivery. The obstetrician, in consultation with the paediatric surgeon and neonatologist, suggests the ideal prenatal, perinatal and postnatal management in each case.³³⁻³⁶

THE NEED FOR SPECIALIZED ORGANIZATIONAL ENTITIES

The factors which are narrated in this article reiterate the need for establishment of two separate organizational entities: 1. Specialized centers for facilities for antenatal diagnosis, evaluation and follow up, with multidisciplinary teams: Advanced facilities for fetal diagnosis should be available at these centers. The involvement of concerned specialists, whenever necessary, will help to improve diagnostic accuracy and streamline management. The paediatric surgeon/fetal medicine specialist could act as the central person coordinating evaluation, communications and decisions. This group can constitute the medical board that has to be involved in contentious decision making in pregnancy complicated by fetal anomaly. The prenatal follow up by the surgeon who is responsible for postnatal care can ease communication, facilitate decision-making and improve confidence of the parents.^{2,16,33-36}

2. Specialized perinatal centers with the advanced facilities for EXIT, neonatal medicine and neonatal surgery: Mothers with fetus having major anomalies with high risk of complications need specialist medical/surgical therapy during perinatal period. They can be referred at the appropriate time during gestation to such centers for advanced care. Delivery can also be conducted at hospitals in close proximity to these specialized centers, so that transport-related issues can be minimized.^{2,16,19-21,33-36}

DISCUSSION

The rapid advancements and widespread use of prenatal genetic testing and fetal imaging have greatly enhanced the diagnostic yield of fetal anomalies. Newer NGS techniques like WES have greatly improved diagnostic accuracy. But these have also resulted in detection of anomalies of uncertain clinical significance. This should be an important consideration while dealing with all fetal anomalies. The decisions regarding the use of screening studies (aneuploidy screening) should be taken considering factors like maternal risk, parental request, medical indications and local regulations, in consultation with concerned specialists whenever necessary. The role of a pediatrician and geneticist is crucial in this regard. It is also vital that all the concerned specialists involved in the management of fetal anomalies be well versed with the latest guidelines and diagnostic options.

The advances in fetal therapy have not generally kept pace with the rapid strides in fetal diagnosis, thus limiting the



options of prenatal therapy. These facilities may be available in only select centres, for high-risk cases, as a part of clinical trials. But the advancements in therapeutic options and fetoscopic techniques may lead to increased adoption of fetal therapy in the future. The present scenario dictates the need for institution of the ideal perinatal therapy for each case, especially with regard to the venue, timing and mode of delivery. The issues pertaining to MTP, TOPFA, invasive fetal diagnosis and fetal therapy have medico legal, ethical and social ramifications, in addition to the medical considerations. Moreover, the issue of MTP is associated with cultural concerns and legal framework-related implications which can be locally variable. These facts reiterate the need for the involvement of a multidisciplinary team in taking management decisions.

The optimal use of fetal diagnostic and therapeutic modalities and scientific perinatal management depend on the accurate estimation of severity of the disorder and the likely complications. The inherent issues with regard to the management of fetal anomalies can only be addressed by the formulation of standard guidelines and adoption of a multidisciplinary approach. The establishment of global registries and collaborative network of fetal specialists will help to set universally accepted standards of care.

CONCLUSION

The prenatal diagnosis of a fetal anomaly should prompt the involvement of a multidisciplinary team to decide on a comprehensive treatment plan. The paediatric surgeons have a pivotal role to play in this regard in consideration of their expertise in the treatment of congenital disorders. The primary objective of the collaborative approach is to ensure structured evaluation and timely prenatal, perinatal and postnatal management. The establishment of specialized prenatal and perinatal centers with diagnostic and therapeutic facilities and trained specialists can go a long way towards achieving this objective.

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