

Congenital Malformations of Fetus: Obstetricians Perspective

Alka B Patil¹, Sayli Thavare², Bhagyashree Badade³

Author's Affiliation:

¹Professor and Head, ^{2,3}Junior Resident, Department of Obstetrics and Gynecology, ACPM Medical College, Dhule, Maharashtra 424002, India.

Corresponding Author:

Alka B Patil, Professor and Head, Department of Obstetrics and Gynecology, ACPM Medical College, Dhule, Maharashtra 424002, India.

E-mail: alkapatil@rediffmail.com

Received on 25.07.2019,

Accepted on 12.10.2019

Abstract

Fetus has been given individual status and rights. Congenital malformations of fetus are diagnosed antenatally through ultrasound and prenatal testing. Congenital anomaly is defined as abnormality of structure, function or metabolism that is present at birth. The gestational age at which severe malformation is diagnosed is crucial for the further management of pregnancy. Prenatal diagnosis of congenital malformation provide information for decision during pregnancy and treatment perinatally. Decision regarding aborting a malformed fetus is challenging in view of human rights, balancing rights against each other is essential. The right to the life of mother and same right of the fetus.

Keywords: Fetus; Malformation; Ethics; Perinatal Mortality.

How to cite this article:

Alka B Patil, Sayli Thavare, Bhagyashree Badade. Congenital Malformations of Fetus: Obstetricians Perspective. Indian J Matern Fetal Neonatal Med. 2019;5(2):117-121.

Introduction

Fetus has been given individual status and rights. With advent of modern technologies and emerging branch of fetal medicine, obstetricians have to face ethical dilemmas related to many fetal issues. In this article we highlight ethical issues related to congenital fetal malformations.¹

Ethics has always been the basis of good practice in medicine. Medicine when practiced at its best, seeks to do what is right and good, and ethics helps in defining and achieving that goal.² Obstetrics is a specialty dealing with two lives, closely linked, whose interest always may not coincide. Congenital malformations account for about 20% of perinatal deaths and a substantial proportion of spontaneous abortions in the first trimester of

pregnancy. Defects in CNS account for 50% of all serious malformations.

Congenital malformations may be easily recognized at birth e.g. Spina bifida or may not be revealed until symptoms and signs indicate its presence.³ A congenital anomaly is defined as an abnormality of structure, function or body metabolism that is present at birth and results in physical or mental disability or is fatal.⁴

The finding of some 'abnormality' in pregnancy transforms what was previously a joyful event into distressing time. The greatest care should be taken in explaining findings of congenital malformations to parents. Sympathetic approach, understanding and reassurance are important.

Aims of prenatal diagnosis of congenital malformations are:-

The identification at an early gestation of abnormalities incompatible with survival or likely to result in severe handicap, to prepare parents and offer the option of termination of pregnancy.

Identification of conditions which may influence

- Timing
- Site
- Mode of delivery.

Identification of fetuses who will benefit from early pediatric intervention.

The identification of fetus who may benefit from in utero treatment.

The parents must decide what action they wish to take. They have to live with the decisions we place in front of them. It is our role to advise, guide and respect their final wishes, irrespective of our personal view.⁵

In obstetrics practice, the strength of counseling for fetal benefit should vary according to presence and severity of anomalies. When lethal anomalies such as anencephaly is diagnosed, what is ethical? Such fetuses are dying patients and counseling is important.⁶ As anencephalous infants do not survive, these women are offered the option of MTP if diagnosed before 20 weeks. Despite their infrequent occurrence, the consequences of birth defects on perinatal morbidity and mortality, infant mortality and ultimate quality of life are significant.

Clinical Implications

Major impact of antenatal diagnosis of malformations is related to the severity of malformations detected. Most severe defects are detected earlier than minor one, which is relevant in countries where MTP is authorized by law before viability. The gestational age at which severe malformation is diagnosed is crucial for further management of pregnancy. Prenatal diagnosis of congenital malformation provides information for decisions during pregnancy and treatment perinatally.³

Gestational age and diagnosis of anomaly

- Anencephaly- 10 to 14 weeks.
- Omphalocele, limb anomalies-diagnosed in first trimester.
- Urinary tract anomalies detected later in pregnancy.⁷

Malformation Sequence

One malformation eg: renal agenesis has led to other malformations eg: oligohydramnios and pulmonary hypoplasia. An association in which various malformations are statistically associated but there is no known or predisposing cause.

Clinical Observations, Indications of High Incidence of Fetal Malformations

- History of an affected sibling or of a previous abortion or stillbirth
- Polyhydramnios / Oligohydramnios
- Slow fetal growth
- Single umbilical artery
- Maternal uncontrolled diabetes
- Advanced maternal age
- History of exposure to teratogenic drugs
- History of maternal rubella in first trimester.³
- High risk pregnancy.

Pregnant Women who are at High Risk for Fetal Malformations are

- Women who have previously delivered a baby with malformations.
- Women aged ≥ 35 yrs
- Women with diabetes mellitus, thyroid dysfunction, epilepsy, phenylketonuria or congenital heart disease.
- Women exposed to drugs with known teratogenic potential.
- Women infected with TORCH group of infections or treponema palladium, varicella or parvovirus in the first 12 weeks of pregnancy.
- Women who are heavy smokers, consume large amount of alcohol or are known substance abusers
- Women with multifetal pregnancies, breech presentation at ≥ 34 weeks of gestation, face/brow presentation.
- Pregnancies complicated by polyhydramnios or oligohydramnios.
- Pregnancies with severe IUGR or macrosomia.
- In women with congenital malformation of uterus or leiomyomata leading to fetal constraint in utero.⁸

Previously large no. of malformations which had bad prognosis are now treated successfully by fetal

and neonatal surgery.

The obstetric decisions for fetal malformations include

- The decisions for MTP
- Antenatal monitoring in a tertiary center
- Fetal therapy
- Decision for the optimum time and mode of delivery
- Optimum neonatal management with neonatologist and pediatric surgeon.⁸
- Despite their low prevalence, fetal malformations are responsible for approximately 30% of perinatal deaths in addition to considerable infant morbidity in developed countries. The gestational age at which a severe malformation is diagnosed is therefore crucial to further management of pregnancy.

The question around appropriate management in case of sonographically established fetal abnormality have risen.

- Should we apply all means to keep alive a fetus with a very poor prognosis?
- Do medical professionals in the field of perinatal medicine agree on fetal prognosis after ultrasound diagnosis of fetal abnormality?
- How should obstetric and neonatal management be done?
- How do parents view end of life decisions concerning their unborn?⁹

Right to Life

- I will maintain the utmost respect for human life, from the time of conception.
- Termination of pregnancy brings out conflicts between the rights of two persons: the rights of mother and the rights of the child in the womb.
- Has the mother the right to have the child in the womb destroyed?
- Does the unborn child have right to life?
- Has the doctor the right to kill the child in the womb at the request of the mother?
- Parents have the desire to have a child of certain quality.
- Fetus in utero is not able to consent to or refuse medical treatment to sustain life or allow its termination.

- Are the parents and doctors free to choose for babies?
- Can death be a legally valid choice in case of severely disabled or malformed baby?¹⁰

Late termination of pregnancy for fetal abnormalities is permitted on different grounds in different countries depending on:

- Type of malformation
- Gestational age at diagnosis
- Abortion legislation
- Severity of structural anomalies directly correlated with abortion rates of anomalous fetuses. USG should be repeated to assess the evolution of the anomaly and attempt to detect other anomalies not previously identified.

Follow up After Termination of Pregnancy:

- Close follow up of parents for early detection of adverse pathological outcomes.
- Referral to psychiatrist and to parent support group is helpful.
- Attempt should be made to detect genetic familial disease for proper counseling.⁸
- Fetal autopsy is important. This can provide additional information or clarification in approximately 15% of cases. This will help in recurrence-risk counseling. Autopsy should only be performed with written informed consent of the women and/or her husband.¹¹

Management

Fetal anomalies.

Cesarean section has been advised for fetal gastroschisis to avoid contamination of fetal abdomen with maternal secretions during labor.¹²

Meningomyelocele and cesarean delivery

The major cause of morbidity in babies with meningomyelocele is the neurological deficit. Exposure of the neural tube to amniotic fluid and to intrauterine pressure and mechanical stresses associated with labor and delivery can injure spinal cord and worsen neurological outcome. Scheduled delivery by cesarean section before the onset of labor should be considered for the fetus with meningomyelocele particularly if prenatal USG and karyotype rule out:

- Presence of severe hydrocephalus
- Chromosomal abnormality
- Multiple systemic anomalies

Medicine is a science, but also an art; medical ethics concern the doctors knowledge as well as behavior.¹³

Patient may present with acute fetal distress requiring emergency CS. Parents should be counseled about possibility of fetal malformation. Destructive operation, if indicated should be done by obstetrician experienced in these procedures.¹⁴

Discussion

Parents have a desire to have a child of certain quality. Congenital anomalies contribute a significant proportion of infant morbidity and mortality as well as fetal mortality. A debate regarding aborting a malformed fetus still exists.¹⁵ Despite their infrequent occurrence, the consequences of birth defects on perinatal morbidity and mortality, infant mortality and ultimate quality of life are significant.³ Because USG can detect associations of specific anomalies, detection of pattern of anomalies may help to make a diagnosis or determine which pregnant women should be offered invasive testing. It has not been established to what extent information provided by MRI may require changes in patient counselling and management. So further studies are needed to assess how additional information from MRI may affect outcome.¹⁶ Decision regarding aborting a malformed fetus is challenging. Counseling to the parents is very important. When parents are informed that fetus has Down's syndrome they may take decision to continue pregnancy. Pregnant women may think that "It is God's wish, I will look after the baby." She may refuse termination of pregnancy. Principle of autonomy demands that her wish to continue pregnancy should be respected.

Delivering and raising a severely malformed and disabled baby may have an impact on physical, mental and social life of family. Women should have the opportunity to consider an option of not continuing the pregnancy. The decision to continue or terminate the pregnancy should always rest with woman.

During the antenatal care visits, the obstetrician is frequently faced by a common question regarding presence of congenital fetal anomaly.¹

Fetal malformations represent important public health issue because of:

- Severe disabilities
- Social consequences
- Financial burden for family
- Effect on community¹⁷

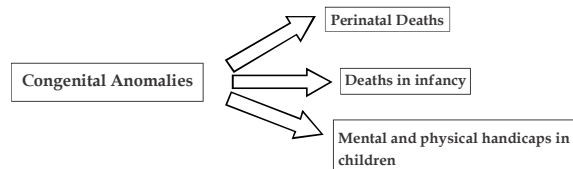


Fig. 1:

Conclusion

Obstetrics is unique speciality dealing with two lives. Congenital malformations of the fetus is diagnosed antenatally through ultrasound and prenatal testing. Obstetrician should disclose the information to pregnant women in a sensitive, sympathetic way. Informed decision of whether to continue or interrupt a pregnancy is quite crucial. In view of human rights, balancing rights against each other is essential. The right to the life of the mother and same right of the fetus.

References

1. Patil AB, Singh S. Fetus and bioethics, Indian journal of maternal-Fetal and Neonatal medicine. 2016 Jul-Dec;3(2):101-06.
2. Sanjay Gupte. Ethical issues in gynecology and obstetrics. Das Gupta, Recent advances in obstetrics and gynecology. Jaypee 2003
3. Daftary, Shirish N and Chakravarti, Sudip and Pai, Muralidhar V and Kushtagi, Pralhad (2016) Holland & Brews Manual of Obstetrics. Reed Elsevier India Pvt. Ltd, New Delhi. ISBN 978-81-312-4240-7; e-book ISBN:978-81-312-4241-4.
4. Al-Alayiyan S, Al Faleh KM, Aborting a malformed fetus: A database issue in Saudi Arabia J elin Neonatal. 2012;1(1):6-11.
5. Ajit Virkud Modern Obstetrics, APC Publishers 1st edition.
6. Frank A Chervenak, Laurence B- McCullough. Ethical issues in Obstetric and Gynaecological practice John Studd Seang Lin Tan, Frank Chervenak. Progress in obstetrics and gynecology Elsevier Vol 17.
7. AV Kannere. Study of congenital anomalies of

- fetus and its outcome in a tertiary care centre. www.ejmanager.com.
8. B. Shakuntala Baliga. Obstetrics Decisions for fetal malformations Usha Krishna and Duru Shah, Vinita Salvi Nozer Sheriar Kaizad R Damania. Pregnancy at risk. Jaypee. 5th edition.
 9. Mishra R. Patil A. Ultrasonographic detection of fetal anomalies. Impact, prognosis and management Indian Journal of Maternal, fetal and neonatal medicine. 2017 Jul-Dec;4(2):191-200.
 10. CM Francis. Medical Ethics. second edition. Jaypee Brothers Medical Publishers (P) Ltd, 2007.
 11. Dickinson JE, Prime DK, Charles AK. The role of autopsy following pregnancy termination for fetal abnormalities. Aust N Z J Obstet Gynaecol. 2007 Dec;47(6):445-9.
 12. Nagendra Sardeshpande. Indications for cesarean delivery. Ranjit Akolkar The cesarean birth. National Mumbai. 2010
 13. Sankarnarayanank Mondkar. Neonatal considerations in cesarean birth. Ranjit Akolkar The cesarean birth. National Mumbai. 2010
 14. Marden PM, Smith DW, McDonald MJ. Congenital anomalies in the newborn infant, including minor variations. A study of 4412 babies by surface examination for anomalies and buccal smear for chromatin. J Pediatr. 1964;64:357-71
 15. Patil AB, Dode P, Ahirrao A. Medical ethics in abortion. Indian Journal of Clinical Practice, 2014 Nov;25(6):544-48.
 16. Todros T, Capuzzo E, Gaglioti P. Prenatal diagnosis of congenital anomalies Images paediatric Cardiol. 2001 April-June;3(2):3-18.
 17. Pinto V, D'Addario V. The accuracy of ultrasound in screening for fetal anomalies. Donald School Basic Textbook of Ultrasound in Obstetrics & Gynecology. JP Medical Ltd. 2014.
-