


A new scheme for risk stratification of pregnancy complicated by fetal anomalies

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ABSTRACT

Fetal anomalies are being increasingly detected during pregnancy with the increased adoption of sonological screening, advances in fetal imaging and genetic testing. The optimal use and factual interpretation of fetal diagnostic modalities and therapeutic decision-making constitute a significant challenge to the concerned specialists. This article aims to review the topic of management concerns in pregnancy complicated by fetal anomaly (PCFA), with the objective of proposal of a risk stratification system that can guide evaluation and therapy. The general management principles in common fetal disorders, the concept of 'soft markers' and the importance of a multidisciplinary approach to therapy are also analyzed. A narrative review of the management principles in PCFA on the basis of analysis of current literature, clinical guidelines and recommendations, in the light of clinical experience, is presented here. A novel risk stratification approach to PCFA is proposed that would reflect the severity, prognosis and possible outcome. The critical parameters in characterization of a prenatally detected fetal anomaly are its physiological/ pathological nature, the severity and associated risk factors, possible progression and complications, and its effect on the gestation and the fetus. These factors determine the need for detailed fetal imaging, invasive fetal diagnosis, fetal therapy, changes in perinatal management and even the option to terminate pregnancy in severe anomalies. All these key parameters are integrated in the new system of risk stratification of PCFA proposed here. The need for categorization of PCFA arises from the variegated nature of fetal disorders that range from benign physiological alterations or minor defects to major, multi-systemic anomalies. Though the management of fetal anomalies has to be highly individualized, proper risk stratification would help bring clarity to the decision-making process.

Keywords: Congenital abnormalities, prenatal diagnosis, fetal therapies, pregnancy, high-risk

INTRODUCTION

Background

Fetal anomalies are being increasingly detected during pregnancy with the increased adoption of sonological screening, advances in fetal imaging and genetic testing. The newer genetic tests and markers (like cell free fetal DNA testing, and biomarker assay) have made it possible to screen for fetal disorders even from a maternal blood sample. Similarly, advances in fetal imaging have made it possible to detect, evaluate and follow up fetuses with structural anomalies of variable severity.¹⁻⁴ The clinical aspects of management like the universal applicability of maternal screening tests, the indications for invasive fetal diagnosis/ fetal therapy and termination of pregnancy for severe anomalies continue to be grey areas clouded by medical and ethical concerns.

Objective: This article aims to review the topic of management concerns in pregnancy complicated by fetal anomaly (PCFA), with the objective of proposal of a risk stratification system that can guide evaluation and therapy. The general management principles in common fetal disorders, the concept of 'soft markers' and the importance of a multidisciplinary approach to therapy are also analyzed.

Methods: A narrative review of the management principles in PCFA on the basis of analysis of current literature, clinical guidelines and recommendations, in the light of clinical experience, is presented here. A novel risk stratification approach to PCFA is proposed that would reflect the severity, prognosis and possible outcome.



The Importance of Risk Stratification of Fetal Anomalies

Many of the prenatally detected fetal anomalies are generally followed up on the basis of the organ system-specific grading systems of severity (as in the case of fetal hydronephrosis). But many of the other common organ system anomalies do not have the benefit of a severity scoring system (as in the case of many anomalies of heart, lung or nervous system). It is neither tangible nor practically feasible to have an individual risk stratification for each and every anomaly.

The wide spectrum of these defects range from minor aberrations to major genetic or structural anomalies. Moreover, many of the fetuses have disorders affecting more than one system, each with variable severity and prognosis. The natural history of the anomalies are also highly variable with possibility of spontaneous resolution in some and the propensity to progress or develop complications during pregnancy in the others. Progression during prenatal period can have an adverse effect on the fetus, the mother and the pregnancy.³⁻⁵

It has been observed in clinical practice that many minor physiological/anatomical alteration(s) in the fetus could be misinterpreted as more sinister conditions, leading to unnecessary investigations, interventions and unwarranted distress. This would be particularly true for anomalies of organ systems without any specific severity scoring. Conversely, there is also the likelihood of a disorder that ideally requires aggressive prenatal testing/intervention, not benefitting from the same in the absence of a proper risk stratification. Delay or failure in the appropriate decision-making regarding management would result in unwarranted complications.³⁻⁵

The Objectives of Risk Stratification of Fetal Anomalies

It is vital to differentiate an anomaly with low likelihood of complications, high probability of spontaneous resolution and scarce need for therapy from those where the converse is true. Once a fetal anomaly is detected on the prenatal scan, it is crucial to have clarity in this distinction, from the perspective of the obstetrician, concerned specialist(s) and the parents. This distinction has the greatest bearing on the decision-making process regarding any prenatal, perinatal or postnatal intervention.

The critical parameters in characterization of a prenatally detected anomaly are the physiological/pathological nature of the anomaly, severity of the disorder, associated problems/risk factors, possible progression, likely complications, and its effect on the gestation and life of the fetus. These factors, in turn decide the need for detailed fetal imaging, invasive fetal diagnosis, fetal therapy, perinatal therapy, changes in obstetric management and the option to terminate pregnancy in severe anomalies.^{1,3,5,6}

All these parameters and factors are integrated in the new risk stratification of PCFA proposed here. The categorization should help in structuring crucial management decisions regarding the investigative or therapeutic modalities in pregnancy. The present stratification is not intended to substitute the available organ-specific grading systems for fetal anomalies. Such grading systems can be integrated into

the risk stratification to supplement the data. The classification systems can thus serve to be mutually complementary, not mutually exclusive.

Risk Stratification for Prenatally Detected Fetal Anomalies based on Severity of the Anomaly and the Necessity for Crucial Management Decisions in Pregnancy Complicated by Fetal Anomaly (PCFA)

Prerequisites of categorization of fetal anomalies: The diagnosis should be based on an Ultrasound performed by a sonologist with expertise in prenatal sonology. But the categorization should be done by the maternal-fetal medicine specialist/paediatric surgeon (with inputs from other specialists where required) and not by the untrained sonologist or obstetrician. This is because the anomaly/anomalies detected have to be assessed for severity and prognosis and management options are to be decided upon by specialists with expertise in management of these conditions (**Figure**).

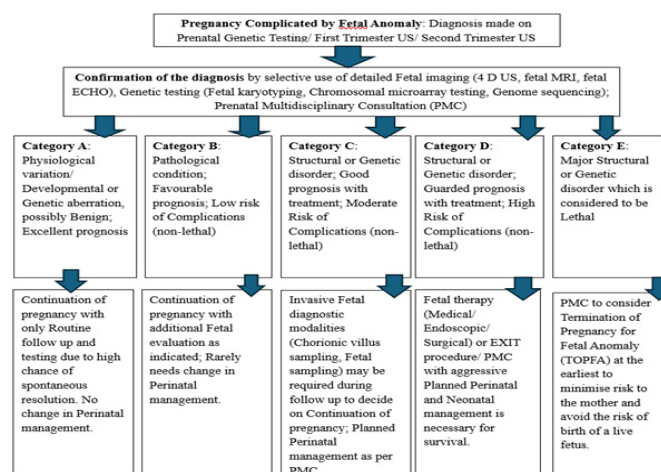


Figure. The general scheme of risk stratification approach to pregnancy complicated by fetal anomalies

If organ-specific grading systems are available for a particular anomaly, it can be integrated with the risk stratification. When more than one anomaly is detected, the severity should be assessed individually. The categorization will have to be revised, on the basis of evolution of the disorder during pregnancy/ test results of specific studies/ follow up investigations, whenever necessary (**Table**).

THE RISK STRATIFICATION OF PCFA

Category A

Physiological variation/developmental or genetic aberration; possibly benign/incidental finding; excellent prognosis; high likelihood of spontaneous resolution; low risk of complications*.

Plan: Continuation of pregnancy with only routine follow up and testing; Selective use of additional fetal evaluation**; No change in perinatal management.

Category B

Pathological condition with less likelihood of spontaneous resolution; favourable prognosis; low risk of complications* (non-lethal).

**Table.** The general scheme of risk stratification approach to pregnancy complicated by fetal anomalies, with examples of each category

Category of risk strata	A	B	C	D	E
Nature of the anomaly	Physiological/minor; possibly benign	Pathological condition	Major defect	Major defect	Major defect
Chance of spontaneous resolution	High	Low	Nil	Nil	Nil
Risk of progression/ complications	Low	Low (non-lethal)	Moderate (non-lethal)	High (non-lethal)	High (lethal)
Prognosis/ Possible Outcome	Excellent	Favourable	Good, with therapy	Guarded, with therapy	Poor
Plan of evaluation	Only selective use of evaluation other than routine tests	Additional evaluation is required	Invasive fetal diagnostic modalities are required	Requires use of fetal therapy/ EXIT procedure	Evaluate to confirm diagnosis
General management plan	Continuation of pregnancy	Continuation of pregnancy	Continuation of pregnancy based on risk evaluation	Continuation of pregnancy based on risk evaluation	Termination of pregnancy for fetal anomaly
Changes in Perinatal management	Nil	May be required	Yes	Yes	-
Examples for each category	Low grade urinary tract dilatation, choroid plexus cysts	Unilateral hydronephrosis, gonadal cyst	Genetic or Syndromic disorders, posterior urethral valves	Large neck lesions, large teratoma, fetal lung lesions	Anencephaly, Trisomy 13, Infantile polycystic kidney

Plan: Continuation of pregnancy with the additional fetal evaluation as indicated** and rarely need changes in perinatal management.

Category C

Structural defect/genetic disorder; good prognosis; moderate risk of complications* (non-lethal)

Requires the use of invasive fetal diagnostic modalities** to decide on continuation of pregnancy; planned perinatal management.

Category D

Major structural defect/genetic disorder; guarded prognosis with treatment; high risk of complications* (non-lethal)

Requires fetal therapy (medical/surgical/fetal endoscopic)/ex-utero intrapartum treatment (EXIT) procedure, with planned obstetric/ perinatal management.

Category E

Major structural defect/genetic disorder (lethal) that requires termination of pregnancy for fetal anomaly (TOPFA)

***Complications:** Fetal/gestational/maternal (narrated below)

****Fetal evaluation:** Non-invasive testing includes serial ultrasound and Doppler, fetal echocardiogram, fetal MRI etc. Invasive, diagnostic testing includes amniocentesis, chorionic villus sampling (CVS), fetal blood/urine sampling etc.

Examples of each category⁵⁻⁸:

A: Isolated findings of low grade urinary tract dilatation, echogenic bowel, intracardiac echogenic focus, choroid plexus cysts etc. (Low probability of being pathological in isolation)

B: Higher grades of urinary tract dilatation; Gonadal cyst; Isolated systemic anomalies (cardiac, lung etc.) that generally carry a good prognosis etc.

C: Complex defects/ multisystemic anomalies/ Genetic disorders that require Amniocentesis/ Chorionic villus sampling/ fetal blood sampling; Posterior urethral valves requiring fetal urine sampling.

D: Fetal lung lesions requiring prenatal maternal steroids, large Teratoma requiring fetal surgery, poor risk Myelomeningocele that is a candidate for fetal surgery, large neck lesions requiring EXIT Procedure.

E: Anomalies like Anencephaly, Trisomy 13, Infantile Polycystic Kidney Disease etc.

Complications in prenatally detected fetal anomalies:

The complications pertaining to prenatally detected fetal anomalies can be classified as fetal, gestational and maternal. Fetal complications will include conditions like growth retardation, fetal anemia, organ damage from progressive disease, cardiac failure, hydrops fetalis, intrauterine death etc.⁴⁻⁶ Gestational (pregnancy-related) complications include amniotic fluid abnormalities, placentomegaly, chorioamnionitis, rupture of protective sacs as in exomphalos etc. and maternal complications include preeclampsia, maternal mirror syndrome, premature labour, dystocia etc.⁶⁻⁹

DISCUSSION

The Concept of Sonological “Soft Markers” Revisited

The categorization proposed here does not consider or include the concept of sonological “soft markers”. The findings labelled so, as those of cerebral ventricular dilatation, thickened nuchal fold, single umbilical artery, shortened humerus/femur length and hypoplastic nasal bone could be markers of significant disorders that may mandate detailed evaluation. The other such findings like mild renal pelvic dilatation, choroid plexus cysts, echogenic bowel and intracardiac echogenic focus could be benign findings with high chance of spontaneous resolution, especially when found in isolation.^{1,4,7,8} But still, they



mandate a detailed anatomical survey and even additional evaluation, especially when they occur along with other abnormalities. Hence it would be detrimental to group together these diverse conditions of variable significance under a common umbrella, that risks undermining their possible significance.⁹⁻¹²

Commonly Detected Anomalies and General Management Principles

The defects that usually need only prenatal follow up and postnatal therapy include conditions like hydronephrosis, isolated congenital diaphragmatic hernia and oesophageal atresia, intestinal atresias, anorectal malformation, enteric cysts, small and intact abdominal wall defects, small sacrococcygeal teratoma, small lymphovascular malformations, benign cysts and uncomplicated craniofacial/limb/chest wall abnormalities.¹¹⁻¹⁴ The defects that may require induction of preterm delivery include ruptured exomphalos, gastroschisis, progressive hydrops fetalis, progressive hydrocephalus, progressive hydrothorax, fetal arrhythmias and severe growth retardation. The defects that need planned caesarian section include large sacrococcygeal teratoma, severe hydrocephalus, large myelomeningocele, conjoined twins, large exomphalos/gastroschisis, large cervical lympho-vascular malformation and the presence of inadequate labor or fetal distress. The defects that may require an ex-utero intrapartum treatment (EXIT) procedure include the conditions that require emergency upper airway access like congenital high airway obstruction syndrome (CHAOS), large neck lesions and conditions requiring immediate extracorporeal membrane oxygenation (ECMO) cannulation. The defects that require TOPFA include severe neurological defects like Anencephaly, chromosomal anomalies like Trisomy 13, bilateral renal agenesis and infantile polycystic kidney disease, metabolic disorders like Tay-Sach's disease and lethal bone dysplasias like recessive Osteogenesis imperfecta.¹³⁻¹⁶

The Importance of a Multidisciplinary Approach to Treatment of Fetal Anomalies

A crucial role in the management of prenatally detected fetal anomalies rests on the paediatric surgeon, who is generally responsible for the management of the majority of the congenital structural disorders that occur in the neonate. It is also important to involve other specialists like the geneticist, paediatrician, nephrologist, cardiologist, neurologist etc. in situations that require their specific expert opinion. The scarcity of well-trained maternal-fetal medicine specialists, especially in limited resource scenarios, only augments the responsibility of the paediatric surgeon to coordinate the opinions and take management decisions. Hence, it is vital for the paediatric surgeon to be well trained and updated about the concepts in evaluation and treatment of these conditions. They should have a dynamic interaction with the obstetrician, to facilitate and streamline therapy.¹⁵⁻¹⁸

The optimal use and factual interpretation of fetal diagnostic modalities and therapeutic decision-making constitute a significant challenge to the concerned specialists. There is

a need for objective guidelines, multidisciplinary integration and evidence-based approach to the management of PCFA. The aspect of informed decision-making by the parents is equally vital for the optimal management of fetal anomalies. A risk stratification approach and multidisciplinary involvement can greatly help the concerned specialists and parents involved in the decision-making process.¹⁹⁻²²

The science of prenatal diagnosis has evolved tremendously over the past few decades with the availability of newer genetic testing, early and advanced fetal imaging, maternal testing, advances in genetic diagnosis like chromosome microarray and genome sequencing and advanced fetal therapy. These advances have necessitated the stratification of anomalies, both isolated and multiple, for the purpose of institution of appropriate therapy.

The novel risk stratification approach proposed here has the objective of segregation of anomalies based on their severity, need for further evaluation/ therapy and possible outcome. The categorization may need revision in some cases based on the evolution of the disorder during pregnancy. The stratification proposed here needs evidence-based validation in prospective cohort studies in the next phase for clinical application. The primary purpose of this structured approach to evaluation and to ensure apt treatment at the right time where necessary and to avoid testing and therapy where it is unwarranted. This would provide a basic and broad framework for prognostication, evaluation, management and follow up.

CONCLUSION

The importance of categorization of prenatally detected fetal anomalies pertain to the need to determine the severity of the disorder, predict outcome, decide on therapy and to facilitate communication with the parents. The need for classification also arises from the variegated nature of these disorders that range from physiological changes or minor defects to major anomalies. A proper risk stratification approach would hence facilitate the diagnostic process, guide evaluation and aid prognostication and formulation of crucial management decisions.

ETHICAL DECLARATIONS

Peer Review Process

This review was externally peer-reviewed.

Conflict of Interest

The authors declare no conflicts of interest.

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Author Contributions

The author is solely responsible for the conception, data collection, analysis, and writing of this manuscript.



REFERENCES

1. Bianchi DW, Crombleholme TM, D'Alton ME, Malone FD. Fetology: diagnosis and management of the fetal patient. 2nd ed. New York, NY: McGraw-Hill Education/Medical; 2010.
2. Corsello G, Giuffrè M. Congenital malformations. *J Matern Fetal Neonatal Med.* 2012;25(Suppl 1):25-29. doi:10.3109/14767058.2012.664943
3. Li G, Yang N, Xie M, et al. Perinatal and follow-up outcome study of fetal anomalies with multidisciplinary consultation. *Ther Clin Risk Manag.* 2017;13:1303-1307. doi:10.2147/TCRM.S138808
4. Remez L, Mayall K, Singh S. Global developments in laws on induced abortion: 2008-2019. *Int Perspect Sex Reprod Health.* 2020;46(Suppl 1): 53-65. doi:10.1363/46e0920
5. Mohaupt SM, Sharma KK. Forensic implications and medical-legal dilemmas of maternal versus fetal rights. *J Forensic Sci.* 1998;43(5):985-992.
6. Kilby MD, Pretlove SJ, Bedford Russell AR. Multidisciplinary palliative care in unborn and newborn babies. *BMJ.* 2011;342:d1808. doi:10.1136/bmj.d1808
7. Luks FI, Carr SR, Feit LR, Rubin LP. Experience with a multidisciplinary antenatal diagnosis and management model in fetal medicine. *J Matern Fetal Neonatal Med.* 2003;14(5):333-337. doi:10.1080/jmf.14.5.333.337
8. Raboei EH. The role of the pediatric surgeon in the perinatal multidisciplinary team. *Eur J Pediatr Surg.* 2008;18(5):313-317. doi:10.1055/s-2008-1038641
9. Mangla M, Anne RP. Perinatal management of pregnancies with fetal congenital anomalies: a guide to obstetricians and pediatricians. *Curr Pediatr Rev.* 2024;20(2):150-165. doi:10.2174/1573396318666221005142001
10. Craig SD. Indicated preterm birth for fetal anomalies. *Semin Perinatol.* 2011;35(5):270-276. doi:10.1053/j.semperi.2011.05.011
11. Bijma HH, Schoonderwaldt EM, van der Heide A, Wildschut HI, van der Maas PJ, Wladimiroff JW. Ultrasound diagnosis of fetal anomalies: an analysis of perinatal management of 318 consecutive pregnancies in a multidisciplinary setting. *Prenat Diagn.* 2004;24(11):890-895. doi:10.1002/pd.883
12. Heaney S, Tomlinson M, Aventin Á. Termination of pregnancy for fetal anomaly: a systematic review of the healthcare experiences and needs of parents. *BMC Pregnancy Childbirth.* 2022;22(1):441. doi:10.1186/s12884-022-04770-4
13. Wataganara T, Grunebaum A, Chervenak F, Wielgos M. Delivery modes in case of fetal malformations. *J Perinat Med.* 2017;45(3):273-279. doi:10.1515/jpm-2015-0364
14. Goel P, Agrawal V, Srinivasan RB. Management, outcome, risk, and expectation classification for structural fetal anomalies to aid antenatal counseling: a systematic review. *J Indian Assoc Pediatr Surg.* 2024;29(4): 309-318. doi:10.4103/jiaps.jiaps_53_24
15. Sadlecki P, Walentowicz-Sadlecka M. Prenatal diagnosis of fetal defects and its implications on the delivery mode. *Open Med (Wars).* 2023;18(1): 20230704. doi:10.1515/med-2023-0704
16. Fung ACH, Kan ASY, Chung PH, Shek NWM, Chan IHY, Wong KKY. Antenatal counselling of congenital surgical anomalies: a decade of experience in a local tertiary centre. *J Paediatr Child Health.* 2021;57(5): 710-714. doi:10.1111/jpc.15321
17. Edwards L, Hui L. First and second trimester screening for fetal structural anomalies. *Semin Fetal Neonatal Med.* 2018;23(2):102-111. doi:10.1016/j.siny.2017.11.005
18. Teefey CP, Soni S, Khalek N. Maternal fetal surgery: intervention and management. *Clin Obstet Gynecol.* 2020;63(2):455-467. doi:10.1097/GRF.0000000000000534
19. Rabie N, Magann EF, Gardner L, Wendel PJ. Arkansas fetal diagnosis and management: identifying and coordinating care for anomalous fetuses. *J Matern Fetal Neonatal Med.* 2016;29(1):46-50. doi:10.3109/14767058.2014.986448
20. Jacobs AR, Dean G, Wasenda EJ, et al. Late termination of pregnancy for lethal fetal anomalies: a national survey of maternal-fetal medicine specialists. *Contraception.* 2015;91(1):12-18. doi:10.1016/j.contraception.2014.10.005
21. Kaplan R, Adams S. Incidental fetal ultrasound findings: interpretation and management. *J Midwifery Womens Health.* 2018;63(3):323-329. doi:10.1111/jmwh.12754
22. Sewell EK, Keene S. Perinatal care of infants with congenital birth defects. *Clin Perinatol.* 2018;45(2):213-230. doi:10.1016/j.clp.2018.01.007