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The Prenatal Diagnosis~ What should we do? What can we do?

Hideaki S[1,2*], Jyoji Y[2], Tomokazu N[2], Katsufumi O[3], Motoichiro S[4], Yoshifusa A[5] and Yu W

[1]Department of Pediatric Surgery, Showa University School of Medicine, Japan

[2]Division of Pediatric Surgery, Showa University Kotou Toyosu Hospital Children's Medical Centre, Japan

[3]Department of Obstetrics and Gynecology, Showa University Koto Toyosu Hospital, Japan

[4]Division of Neonatology, Children's Medical Center, Showa University Koto Toyosu Hospital, Japan

[5]Division of Pediatrics, Children's Medical Center, Showa University Koto Toyosu Hospital, Japan

1. Abstract

Purpose: Prenatal diagnostic accuracy has improved; however, the pediatric surgeon's role remains unclear. This paper aimed to determine the prenatal diagnoses made and the role of pediatric surgeons.

Methods: A 6-year retrospective review of 904 pregnancies managed at our institute was conducted. They were classified as a normal pregnancy (NP, n=194), abnormal pregnancy maternal factor (MF, n=449), or abnormal pregnancy fetal factor (FF, n=261).

Results: In the FF group, the identified conditions were twin pregnancies (n=75), intrauterine growth restriction (IUGR) (n=49), breech presentation (n=26), arrested development (n=19), hypoamnion (n=42), fetal distress (n=16), hydramnios (n=10), abnormal heart sounds (n=5), meconium staining (n=5), surface anomaly (n=4), calcification (n=2), fetal hydrops (n=2), fetal death (n=2), bowel dilatation (n=2), abdominal mass (n=1) and diaphragmatic hernia (n=1). Case of twin pregnancies, breech presentation, arrested development, IUGR, hypoamnion, abnormal heart sounds, meconium staining and fetal hydrops did not require surgery. Of the 16 cases of fetal distress, 1 had biliary atresia. Of the 10 cases of hydramnios, 1 had meconium peritonitis. Of the 4 with surface anomalies, 3 had gastroschisis. Of the 2 with calcification, 1 had meconium peritonitis. Of the two fetal deaths, 1 had anal atresia suggesting a chromosomal abnormality. Of the 2 cases of bowel dilatation, 1 had bowel atresia. The abnormal mass was caused by adrenal bleeding. These diagnoses were made at an average of 27.4 gestational weeks; however, 2 cases of gastroschisis, suggesting a body stalk anomaly; diaphragmatic hernia; and brain cysts were diagnosed before 20 weeks and were aborted. After delivery, the mortality rate was 0% in neonates treated by pediatric surgeons and neonatologists.

Conclusion: Fetal abnormalities are rare; however, early aggressive management with the cooperation of obstetricians and parents is crucial for pediatric surgeons to minimize the effects of anomalies.

2. Keywords: Prenatal Diagnosis; Fetal Anomaly; Prenatal Therapy; Prenatal Medicine

3. Abbreviations: MRI: Magnetic Resonance

*Corresponding author: Hideaki Sato, Division of Pediatric Surgery, Kotou Toyosu Hospital Children's Medical Centre, Showa University, 5-1-38, Toyosu, Koutou-Ku, Tokyo, 135-8577, Japan, Tel: +81-3-6204-6000; E-mail: shide2929@gmail.com

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Imaging; CVS: Chorionic Villus Sampling; cfDNA: Cell-free DNA; NIPT: Non-Invasive Prenatal Testing; NIPS: Non-Invasive Prenatal Screening; NP: Normal Pregnancy; MF: Abnormal Pregnancy Maternal Factor; FF: Abnormal Pregnancy Fetal Factor; IUGR: Intrauterine Growth Restriction

4. Introduction

There has been significant progress in making prenatal diagnose over the resent decades with advances in research, technology and skill [1]. Advances in sonographic techniques have enabled the detection of heart anomalies and/or hydrops in the fetus [2] and the advanced resolution of magnetic resonance imaging (MRI) enables the detection of anomalies on the surface as well as those of internal organs [3]. Genetic testing techniques have fundamentally changed the practice of prenatal diagnosis in the recent decades [4]. Routine blood investigations available to all pregnant women can determine the risk of common chromosome abnormalities; chorionic villus sampling (CVS) and amniocentesis have been used to diagnose almost all conditions with a known genetic cause [4]. The acceptance of greatly improved non-invasive risk assessment for common fetal aneuploidies such as cell-free DNA (cfDNA) testing technologies of the maternal plasma (non-invasive prenatal testing (NIPT) or non-invasive prenatal screening (NIPS)) has resulted in a significant decline in invasive testing requiring CVS or amniocentesis [5]. The term "fetal anomalies diagnosis" can be replaced by the term "prenatal medicine," which includes everything from diagnosis to treatment [6]. The importance of counseling for surgical anomalies has been the focus for the last 2 decades since Crombeholme et al. described these anomalies [7]; however, in the era of prenatal medicine, the role of the pediatric surgeon in prenatal care remains unclear. This study aimed to investigate the prenatal diagnoses that are made and the role of the pediatric surgeon.

5. Materials and Methods

Study design

This single-center study included a retrospective review of 904 pregnancies managed by obstetricians at our institute between January 2014 and December 2019. The pregnancy course was classified as a normal pregnancy (NP), abnormal pregnancy maternal factor (MF) or abnormal pregnancy fetal factor (FF) (Figure 1).

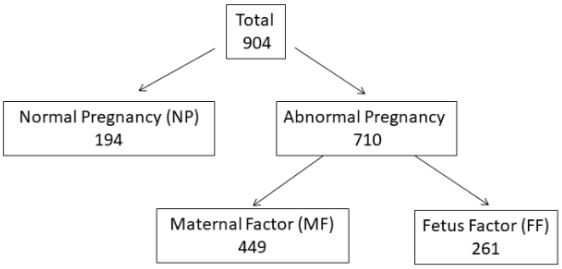

Figure 1: Flow chart of patients included in the study.

All pregnancies were routinely evaluated by the obstetrician using ultrasound.

The cases referred from other hospitals and requiring urgent cesarian delivery were excluded. All pregnancies were followed until delivery and the outcomes of neonates treated by the neonatologist were reviewed.

In the case of an abnormal pregnancy, a team comprising of the obstetrician, neonatologist, nursing staff of the ward and clinical psychologist discussed the management of each pregnancy once per week. In the case of a fetus expected to require surgery after delivery, the pediatric surgeon was included in the above team.

The cause of the abnormality, including the maternal and fetal factors, were evaluated and the likely effects of the abnormalities in the neonates were evaluated based on the clinical course and prognosis. To investigate the relationship between the fetal condition and the neonatal outcomes, the data were summarized into NP and FF groups.

6. Results

The inclusion of patients is shown in Figure 1. In total, 904 pregnancies were identified; of these, 194 pregnancies (21%) met the NP criteria, 710 met the MF or FF criteria, of which 449 had MF (49%) and

261 had FF (28%).

In the NP group, 28 neonatal abnormalities were identified as follows: hyperbilirubinemia (n=21, 75%), respiratory distress (n=4, 15%), urogenital abnormalities (n=2, 7%) and anal atresia (n=1, 3%). After delivery, the mortality rate in neonates was 0% (Table 1).

Table 1: Postnatal condition of neonates in the normal pregnancy group.

Prenatal Diagnosis	No.	Postnatal Condition	No.
Normal	194	Normal	166
		Hyperbilirubinemia	21
		Respiratory Distress	4
		Hydronephrotic Kidney	1
		Megalocystis	1
		Anal Atresia	1

Table 2: Pre- and post-natal condition of neonates in the abnormal pregnancy caused by a fetal factor group.

Prenatal Diagnosis	No.	Postnatal Condition	No.	Intervention by Pediatric surgeon
Twin	75	Normal condition	75	
IUGR	49	Normal condition	22	
		Hyperbilirubinemia	9	
		Low birth weight	5	
		Respiratory distress syndrome	4	
		Heart anomaly	4	
		Asphyxia	2	
		Coiling umbilical cord	2	
		Meconium related ileus	1	Enema
Hypo amnion	42	Normal condition	27	
		Hyperbilirubinemia	9	
		Heart anomaly	2	
		Respiratory distress syndrome	2	
		Asphyxia	2	
Breech Presentation	26	Normal condition	26	
Arrested development	19	Normal condition	19	
Fetal Distress	16	Normal	9	
		Hyperbilirubinemia	3	Operation for Biliary Atresia :1
		Asphyxia	1	
		Apnea	1	
		Meconium Aspiration	1	
		Coiling umbilical cord	1	
		Megalocystis	1	

		Anal Atresia	1	Operation
Hydramnios	10	Normal condition	4	
		Cleft lip and palate	3	
		Chromosomal aberration	1	
		Meconium peritonitis	1	Drainage, operation
		Floppy infant	1	
Abnormal heart sound	5	Normal condition	3	
		Asphyxia	1	
		Tortion of umbilical cord	1	
Meconium staining	5	Normal condition	3	
		Meconium Aspiration	1	
		Apnea	1	
Surface anomaly	4	Gastroschisis	3	Operation:1
		Cerebral aneurysm	1	
Calcification in abdominal cavity	2	Meconium peritonitis	1	Operation
		Brain calcification	1	
Fetal hydrops	2	Fetal hydrops	2	
In utero fetal death	2	Normal condition	1	
		Anal Atresia, Chromosomal aberration	1	
Bowel dilatation	2	Normal condition	1	Enema
		Bowel atresia	1	Operation
Abdominal mass	1	Adrenal hematoma	1	
Diaphragmatic hernia	1	Diaphragmatic hernia	1	

In the FF group (Table 2), the identified fetal conditions were as follows: twin pregnancies (n=75), intrauterine growth restriction (IUGR) (n=49), hypoamnion (n=42), breech presentation (n=26), arrested development (n=19), fetal distress (n=16), hydramnios (n=10), abnormal heart sounds (n=5), meconium staining (n=5), surface anomaly (n=4), calcification in the abdominal cavity (n=2), fetal hydrops (n=2), in-utero fetal death (n=2), bowel dilatation (n=2), abdominal mass (n=1) and diaphragmatic hernia (n=1). Cases of twin pregnancy, breech presentation, arrested development, IUGR, hypo-amnion, abnormal heart sounds, meconium staining and fetal hydrops did not require surgical intervention or consultation by the pediatric surgeons after birth. Of the 16 cases of fetal distress, 1 had biliary atresia and 3 had hyperbilirubinemia after birth. Of the 10 cases of hydramnios, 1 had meconium peritonitis that required abdominal drainage after birth. 2 cases suspected with esophageal atresia in the 10 cases of hydramnios, 1 had a cleft lip and palate and the other revealed suffering floppy infant after birth. Of the 4 cases with surface anomalies, 3 had gastroschisis that required treatment after birth and the other case had a cerebral aneurysm. Of the 2 cases with calcification in the abdominal cavity, 1 had meconium peritonitis that required laparotomy after birth and the other had a calcification in the brain. Of the 2 in utero fetal deaths, 1 had anal atresia suggesting a chromosomal abnormality. Of the 2

cases with bowel dilatation, 1 bowel atresia and required laparotomy, while the other had no abnormalities in the bowel. An abnormal abdominal mass was found to be an adrenal hematoma. The diaphragmatic hernia on the left side was found at 19 weeks of gestation and the parents opted for an abortion. These diagnoses were made at an average of 27.4 gestational weeks. Two cases of gastroschisis suggesting a body stalk anomaly, 2 cases of in utero fetal deaths, diaphragmatic hernia, calcification in the brain and cerebral aneurysm were diagnosed before 20 weeks of pregnancy and were aborted (Table 3).

Table 3: Post-natal condition of terminated pregnancies.

Prenatal Diagnosis	Postnatal Condition	No.
In utero fetal death	Anal Atresia, Chromosomal aberration	1
	Normal condition	1
Surface anomaly	Gastroschisis, Body stalk anomaly	2
	Cerebral aneurysm	1
Calcification in abdominal cavity	Brain calcification	1
Diaphragmatic hernia	Diaphragmatic hernia	1

After delivery, the mortality rate was 0% in neonates who were treated by pediatric surgeons and neonatologists.

7. Discussion

The importance of teamwork in prenatal care has been known for decades [6-11]. Kokila described the importance of a multidisciplinary team comprising of obstetricians, neonatologists, geneticists, pediatricians, pediatric surgeons and occasional other specialists with expertise to deal with all maternal and fetal complexities related to the diagnosis of a structural defect. This team should be able to provide information to the prospective parents on the fetal outcomes; possible interventions; appropriate setting, time and route of delivery and expected postnatal outcomes. The role of the surgical consultant in this team is to present the information regarding the prenatal and the natural postnatal history of an anomaly, its surgical management and the long-term outcomes [8]. Several papers describe the role of the pediatric surgeon in the perinatal management. Surgeons influence the time of delivery, mode of delivery and termination of pregnancy [7-10]. Loren et al. suggests the importance of exposure to prenatal consultation during pediatric surgery residency [11].

The prenatal diagnosis of surgically correctable congenital malformations has remarkably improved due to the development of imaging technologies as well as new genetic testing techniques in the past decade [1]. In genetic testing, cell-free DNA testing such as NIPT or NIPS has enabled non-invasive risk assessment for common fetal aneuploidies [4,5]. For structural testing, ultrasound scanning is now routinely performed at 18–20 weeks of gestation [2]. Benachi et al. reported that when the fetal malformation is diagnosed in the first trimester, consultation with the pediatric surgeon is important [12]. However, some papers reported that better resolution and increased experience with ultrasound scans has increased the detection rate of fetal anomalies but at the expense of higher false-positive rates [13]. Hence, repeated consultations should be recommended for precise information regarding changes seen on ultrasound and/or MRI images, especially in cases where a poor prognosis is expected [12].

In our series, 6 cases in the FP group (2.2%) and 1 case (0.5%) in the NP group required surgery after a consensus on the maternal and fetal condition between the multidisciplinary team. This resulted in a 0% mortality rate. Seven patients opted for termination of pregnancy due to a severe

chromosomal aberration, body stalk anomaly, or central nervous system disease in the fetus. However, one case of congenital diaphragmatic hernia opted for abortion based on information obtained from the internet without adequate information from the pediatric surgeon.

Although the importance of the role of the pediatric surgeon for counseling and as a member of the maternal care team has been suggested, few papers referred to the parental anxiety [14].

Marokakis et al. stated in a systematic review of prenatal counseling, including anxiety and distress in parents, that parents desired written, visual and web-based information resources [14]. In this era where information is easily available, the internet allows parents to search for the pathologic conditions and prognosis of their fetus. Reliable sources are easily available on forums such as "Pregnancy and childbirth-family doctor.org," supported by the American Academy of family Physicians (https://familydoctor.org/family-health/pregnancy-and-childbirth/). These tools are useful and provide easily accessible information to the parents about the pregnancy, fetal health, childbirth and caring for the newborn. However, this easily available information has its pros and cons.

There is a risk of incorrect comprehension by the parents about their fetus' condition and prognosis. Precise and reliable information on the pathologic and prognostic condition for surgically correctable congenital malformations of the fetus must be offered to the parents by the maternal care team, including pediatric surgeons, through face-to-face counseling. Thus, the pediatric surgeon`s role has changed in the information-driven era of prenatal medicine.

7. Conclusion

Fetal abnormalities are rare; however, early aggressive management with the cooperation of obstetricians and parents is crucial for pediatric surgeons to minimize the effects of anomalies.

8. Acknowledgement

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9. Ethics Approval and Consent to Participate

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards. The study was approved by the Bioethics Committee of Showa University. Informed consent was obtained from all individual participants included in the study.

10. Availability of Data and Material

The datasets generated and analyzed during the current study are not publicly available to protect the privacy of the study participants but are available from the corresponding author on reasonable request.

11. Authors' Contribution

HS: Conceptualization, Methodology, Formal Analysis, Investigation, Data Curation, Writing-Original Draft, Visualization; JY: Review & Editing; TN: Investigation, Resources; YW: Review & Editing, Supervision, Project administration, Guarantor. All authors have read and approved this manuscript.

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