A Five–Year Review of Cases Referred to the Genetics Clinic, PGH for Findings of Congenital Anomalies on Prenatal Ultrasound

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ABSTRACT

Background. Prenatal diagnosis in the Philippines has been limited to standard two-dimensional ultrasonography for antenatal screening of high risk pregnancies. Once findings of fetal anomalies are confirmed, a referral to the Genetics Clinic for prenatal counseling is recommended.

Objectives. To review the prenatal cases referred to the Genetics Clinic for counseling and its neonatal outcome and to evaluate the diagnostic accuracy of two-dimensional ultrasonography performed in detecting congenital malformations in the fetus.

Methods. This was a retrospective study based on clinical histories, prenatal ultrasonographic findings and neonatal outcome of 128 pregnancies referred to the Genetics clinic for prenatal counseling from January 1, 2003 to December 31, 2007.

Results. The most frequently encountered congenital anomalies were malformations of the central nervous system, abdominal wall defects, hydrops fetalis and their associated anomalies. The diagnostic accuracy was 100% for abdominal wall defects and associated anomalies, 92.7% for CNS malformations and 92.3% for hydrops fetalis. Overall, the diagnostic accuracy was 91.6% with a false-positive rate of 2.8%.

Conclusion. Prenatal ultrasound findings of congenital anomalies have increasingly become a major reason for referral to the Genetics Clinic. Although these fetal anomalies can be diagnosed accurately in the majority of cases by standard ultrasonography, neonatal mortality and morbidity remain high.

Key Words: congenital anomaly, prenatal counseling, 2D ultrasonography

Introduction

Prenatal diagnosis in the Philippines has generally been limited to maternal serum screening and standard twodimensional (2D) ultrasonography for congenital anomaly screening. Invasive techniques like amniocentesis, chorionic villi sampling and fetal blood and tissue samplings are rarely performed because of the cost and the risks for fetal loss, bleeding, infection and fetal injury. Furthermore, if fetuses are found to have anomalies, religious practices and Philippine laws do not allow termination of pregnancy.

In many countries, ultrasound is offered routinely to all

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College of Medicine-Philippine General Hospital University of the Philippines Manila pregnant women despite controversies over its benefits. However, in the local setting, it is reserved for high-risk pregnancies and is usually done during the second and third trimesters of pregnancies. If a fetal anomaly is detected, a detailed congenital anomaly scan is requested for a more thorough evaluation and in cases with congenital heart defects, a fetal echocardiography is also recommended.

At the Philippine General Hospital (PGH), the prenatal care of both mother and fetus with findings of congenital anomalies on prenatal ultrasound is shared by many specialists in the field of obstetrics, surgery and pediatrics depending on the type and the severity of the malformations seen on ultrasound. Prenatal genetic counseling is carried out after a diagnosis of structural fetal anomalies is made. During the counseling process, the couples are informed about the natural history of the birth defect, its etiology, pattern of inheritance and genetic risks if known. As well, the approach to the neonatal diagnosis and appropriate management are discussed. Correct identification of congenital anomalies coupled with good prenatal counseling reduces parental anxiety and help expectant parents prepare for the birth of an affected child.

The general objective of this paper was to review the prenatal cases of women referred to the Genetics Clinic, PGH for findings of congenital anomalies on prenatal ultrasound and to evaluate the diagnostic accuracy of the two-dimensional ultrasonography done in detecting congenital malformations in the fetus. The specific objectives of this study were: 1) to determine the clinical profile of the pregnant women referred to the Genetics Clinic for significant findings of congenital anomalies on ultrasound according to age, gravidity and parity, and the gestational age at which the ultrasound was done; and 2) to determine the type of congenital malformations identified among neonates and to determine the neonatal outcome of these high risk pregnancies.

Materials and Method

Patients who were referred for prenatal genetic counseling to the Section of Genetics from January 1, 2003 to December 31, 2007 were identified and the following data were obtained retrospectively from their medical records: age, gravidity and parity, age of gestation, history of consanguinity, risk factors, sonographic findings and

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fetal outcome. The neonatal outcomes were taken from the records of each neonate and the dysmorphologic findings were correlated with the prenatal sonographic findings. All data were drawn from the medical records of the Genetics Clinic, Neonatal Intensive Care Unit (NICU), Perinatology Unit and Medical Records Section of the Philippine General Hospital. The descriptive data are presented as percentages.

Results

Demographic Features

From January 1, 2003 to December 31, 2007, a total of 129 cases were referred to the Section of Genetics for prenatal genetic counseling. All pregnancies were from non-consanguineous union. Eighty six percent (n=111) of the cases were seen at the Genetics Clinic of the Outpatient Department and only 14% (n=18) were referred from the General Obstetrics Ward and Intensive Maternal Unit of the hospital.

The demographic data of the patients are presented in Table 1. Majority of the patients were between 20 and 35 years old, with the youngest being 15 years old while the oldest was 44 years old. The gravidity ranged from 1 to 7. In many instances, the anomalies were detected during the last trimester of pregnancy. Risk factors were identified in more than half of the cases and these were advanced maternal age (defined as an age of at least 35 years at the expected date of confinement), high gravidity, infection and use of an abortifacient during the first trimester of pregnancy.

All of the women counseled were pregnant at the time of consultation except for one who had just delivered a child with Trisomy 18 and was contemplating on another pregnancy. As seen in Table 2, seven of the 128 pregnant

Table 1. Demographic Data of Patients Referred to the Genetics

 Clinic for Prenatal Counseling

	Cases (n=129)	Percentage %
A. Age		
< 20 years	11	8.5
20-35 years	96	74.5
>35 years	22	17
B. Gravidity		
Primigravid	45	35
Segundigravid	27	21
Multigavid	57	44
C. Parity		
Nulliparous	56	43.4
Primiparous	27	21
Segundiparous	21	16.2
Multiparous	25	19.4
D. Age of Gestation		
First trimester	2	1.6
Second trimester	24	18.6
Third Trimester	102	79
Postpartum	1	0.8
E. Risk Factors		
With identifiable risk factor	85	66
Without identifiable risk factor	44	34

women seen had normal sonographic findings and the main reason for referral to the Genetics Clinic was because of a previous birth of a child with congenital anomalies. Overall, the top indication for prenatal counseling was detection of a fetal anomaly on standard 2D ultrasonography.

Table 2. Sonographic Profile of the Prenatal Cases

0 1				
Not Pregnant			1	
Pregnant			128	
Normal ultrasound	7	(5.5%)		
Positive congenital scan	121	(94.5%)		
Total Cases			129	

Sonographic Findings

The types of malformations seen on prenatal 2D ultrasonography are listed in Table 3. Isolated malformations of the central nervous system were by far the most frequently encountered anomalies in 45.4% of cases (55/121). In almost 19.8% of cases (24/121), defects in the abdominal wall (isolated and in combination with other anomalies) were seen; among these, omphalocoele was the most frequent abdominal wall defect detected on sonography, with 10 cases reported. Findings of hydrops fetalis, a serious fetal disorder characterized by abnormal fluid accumulation in more than two compartments of the fetal body, comprised 10.7% (13/121) of the cases. There were 7 twin pregnancies with one case having no fetal abnormalities. There were 13 pregnancies that had associated fetal anomalies.

Outcome

Out of the 121 cases with significant findings on congenital anomaly scan, 88.4% (107/121) were delivered in PGH and 11.6% (14/121) were delivered in other birthing centers. Table 4 summarizes the outcome of all cases with significant findings on congenital anomaly scan and the accuracy of these prenatal findings.

A comparison of prenatal ultrasound findings with postnatal physical examination and diagnostic findings was done. The correlation of the prenatal and postnatal findings is shown in Table 5. Only those pregnancies delivered in PGH (n=107) were included in the data analysis. Of the 107 cases, 91.6% (n=98) had malformations correctly identified on ultrasound. Of these, 12.2% (12/98) had additional dysmorphologic findings detected upon postnatal evaluations. Anomalies not identified on ultrasound in these patients included limb abnormalities such as talipes equinovarus, amnion bands and macrodactyly; facial hemangioma, myelomeningocoele and cleft lip and/ or palate. Abdominal wall defects and gastro-intestinal tract defects were diagnosed accurately by ultrasound. The diagnostic accuracy for malformations in the central nervous system (CNS) was 92.7%, 92.3% for hydrops fetalis, 80% for renal anomalies and 50% for skeletal dysplasias. In this report, the overall diagnostic accuracy of standard ultrasonography in detecting fetal anomalies is 91.6% (98/107).

Table 3. Malformations Observed on Prenatal Sonography

Types of Congenital Anomalies	Number of Cases n=121	Percentage	
A. Primary CNS Malformations	n=55	45.4	
Hydrocephalus/Ventriculomegaly	37		
Anencephaly	11		
Encephalocoele	3		
Hydrocephalus and Holoprosencephaly	2		
Holoprosencephaly	1		
	1		
Microcephaly R. Abdominal Well Defeate	=	19.0	
B. Abdominal Wall Defects	n=22	18.2	
Gastroschisis	6		
Omphalocoele	6		
Congenital Diaphragmatic Hernia (CDH)	3		
CDH plus Omphalocoele	1		
CDH plus Hydrops fetalis	1		
Omphalocoele plus Ectopia cordis	1		
Gastroschisis plus clubfeet	1		
Limb body wall complex	2		
Pentalogy of Cantrell	1		
C. Gastrointestinal Malformations	n=3	2.5	
Esophageal atresia	1		
Tracheo-esophageal atresia	1		
Duodenal atresia	1		
D. Hydrops Fetalis	n=10	8.2	
E. Renal Anomalies	n=6	5	
		5	
Polycystic Kidney Disease	4 2		
Hydronephrosis		2.2	
F. Skeletal Dysplasia	n=4	3.3	
Achondroplasia	2		
Osteogenesis Imperfecta	1		
Cardiomegaly T/C Skeletal dysplasia	1		
G. Others	n=8	6.6	
Fetal Trisomy	4		
Cleft Lip	2		
Sacrococcygeal teratoma	1		
Cystic hygroma	1		
H. Multiple Fetal Anomalies			
Anhydramnios, renal agenesis, hepatic cyst, TGA			
Renal agenesis, omphalocoele, skeletal dysplasia	n=6	5	
Hydrocephalus, omphalocoele, myelomeningocoele	1	0	
Cleft lip, esophageal atresia, Dandy Walker	1		
malformation	1		
Hydrocephalus, esophageal atresia, atrial septal	1		
	1		
defect	1		
Anencephaly, esophageal atresia, meningocoele	1		
I. Twin Gestation	_		
Twin gestation, no anomalies	n=7	5.8	
FDIU one twin, normal other twin	1		
Hydrocephalus 1 twin, FDIU other twin	1		
Hydrocephalus, both twins	1		
Hydrops fetalis in one twin, normal other twin	1		
Hydranencephaly/meningocoele one twin,	2		
Anencephaly other twin	1		

Table 4	. Outcome	of	the	Cases	with	Significant	Findings	on
Ultrasou						U	0	

Correct Diagnosis Incorrect Diagnosis	98 (91.6%) 9 (8.4%)	
Total Cases	107	

Postnatally, malformations in the central nervous system (CNS) were by far the most frequently encountered type of anomalies accounting for half of the cases. Congenital hydrocephalus both isolated and in combination with other anomalies, made up 38.7% of all malformations and 77.4% of all CNS anomalies. This was followed by neural tube defects (NTD), abdominal wall defects and hydrops fetalis which accounted for 10.4%, 19.8% and 12.3% of all cases,

Table 5. Correlation of Prenatal and Postnatal Findings

Type of Congenital Anomaly	Prenatal Findings Number of Cases (n=107)	Postnatal Findings Number of Cases (n=98)	Percentage (%) Ave 91.6%
A. Primary CNS Malformations	(n=48) 36	(n=44) 33	91.7
Hydrocephalus/Ventriculomegaly Anencephaly	50 7	6	91.7 85.7
Encephalocoele	2	2	100
Hydrocephalus and Holoprosencephaly	2	2	100
Holoprosencephaly	1	1	100
B. Abdominal Wall Defects	(n=19)	(n=19)	
Gastroschisis	5	5	100
Omphalocoele	5	5	100
Congenital Diaphragmatic Hernia (CDH)	3 1	3	100
CDH plus Omphalocoele CDH plus Hydrops fetalis	1	1 1	100 100
Omphalocoele plus Ectopia cordis	1	1	100
Gastroschisis plus clubfeet	1	1	100
Limb body wall complex	2	2	100
C. Gastrointestinal Malformations	(n=2)	(n=2)	
Esophageal atresia	1	1	100
Tracheo-esophageal atresia	1	1	100
D. Hydrops Fetalis	(n=10)	(n=9)	90
E. Renal Anomalies	(n=5)	(n=4)	
Polycystic kidney Disease	3	3	100
Hydronephrosis	2	1	50
F. Skeletal Dysplasia	(n=4)	(n=2)	
Achondroplasia	2	0	0
Osteogenesis Imperfecta	1	1	100
Cardiomegaly T/C Skeletal dysplasia	1	1	100
G. Others	(n=6)	(n=5)	
Fetal Trisomy	2	2	100
Cleft Lip	2	2	100
Sacrococcygeal teratoma Cystic hygroma	1 1	1 0	100 0
			0
H. Multiple Fetal Anomalies	(n=6)	(n=6)	100
Anhydramnios, renal agenesis, hepatic cyst, TGA Renal agenesis, omphalocoele, skeletal dysplasia	1 1	1	100
Hydrocephalus, omphalocoele, myelomeningocoele	1	1	100
Cleft lip, esophageal atresia, Dandy Walker malformation	1	1	100
Hydrocephalus, esophageal atresia, atrial septal defect	1	1	100
Anencephaly, esophageal atresia, meningocoele	1	1	100
. Twin Gestation	(n=7)	(n=7)	
Twin gestation, no anomalies	1	1	100
FDIU one twin, normal other twin	1	1	100
Hydrocephalus 1 twin, FDIU other twin	1	1	100
Hydrocephalus, both twins Hydrops fetalis in one twin, normal other twin	1 1	1	100 100
Hydrops fetalis in one twin, normal other twin Hydrops fetalis in both twins	1 1	1 1	100
Hydrops letans in boar twins Hydranencephaly/meningocoele one twin, anencephaly other twin	1	1	100

respectively.

In 8.4% of cases (9/107), the postnatal diagnoses were discordant with the prenatal findings. Three patients had normal neonatal findings giving a false positive rate of 2.8% (3/107). A structural fetal anomaly was considered false-positive if the suspected defect was not confirmed postnatally or on postmortem examination. Table 6 enumerates the

malformations that were identified incorrectly.

Table 7 presents the survival outcome of the neonates who were found to have congenital malformations prenatally. As mentioned earlier, only 107 out of the 121 pregnant mothers with significant findings of congenital anomalies on ultrasound were delivered in PGH; seven of these were twin pregnancies giving a total of 114 babies

Table 6. Malformations Identified Incorrectly (n=9)

2D ultrasound findings	Postnatal Outcome		
Hydrocephalus	Normal		
Cystic hygroma	Normal		
Hydronephrosis	Normal		
Anencephaly	Missed abortus		
Hydrocephalus	Hydranencephaly		
Hydrocephalus	Holoprosencephaly		
Hydrops fetalis	Hydrocephalus		
Achondroplasia	Thanatophoric dysplasia		
Achondroplasia	Thanatophoric dysplasia		

Table 7. Outcomes of Neonates Delivered at PGH

	n=114	Percentage (%)
Stillborn	20	17.5
Early neonatal death	54	47.4
Late neonatal death	4	3.5
Discharged with morbidity	19	16.7
Discharged well	11	9.6
Normal neonates	6	5.3

evaluated postnatally. Fifteen percent (17/114) of these babies was discharged alive and well; 6 were normal babies, 5 cases with isolated gastroschisis, 3 cases of omphalocoele, 2 with cleft lip and 1 with hydronephrosis. Sixty-eight percent (78/114) of these cases diagnosed prenatally with congenital anomalies were stillbirths, neonatal or infant deaths. Majority of the stillbirths did not have any postmortem investigations. There is no long-term followup of babies who were discharged well but of the 19 cases discharged with morbidities, 3 (15.7%) are regularly seen in the Genetics Clinic. These are the patients with congenital hydrocephalus; one case being isolated and the other two are part of a malformation syndrome. Only one of the three babies with congenital hydrocephalus had ventriculoperitoneal shunting.

Discussion

In most developed countries, invasive and non-invasive prenatal diagnostic testing are offered as part of routine prenatal care. In the local setting, the prenatal diagnosis performed are mostly non-invasive techniques which include ultrasonography for congenital anomalies and in a limited way, maternal serum screening.¹ Sonograms are performed with variable indications at any stage of pregnancy on women who are at risk for conceiving a fetus with congenital anomaly and who have poor obstetric history, family history of and previous birth of a child with congenital anomalies.

In this retrospective study, congenital anomalies were detected during routine ultrasound in 34.4% of the cases while 65.6% had specific indications for performing the ultrasound such as those mentioned above. This relatively high percentage of anomalies ascertained by routine

ultrasound of pregnant women is probably one of the reasons why ultrasound screening is offered routinely to all pregnant women in many other countries.² Advanced maternal age risk factor associated with fetal chromosomal abnormalities is also one of the more prominent reasons for doing routine ultrasound among pregnant women. In this study, almost 75% of the pregnant women referred to the clinic were between 20 to 35 years old and only a small percentage of the patients were above 35 years of age.

Most of the cases (84.5%) in this study had significant findings on congenital anomaly scan detected during the last trimester of pregnancy. In institutions with advanced technology, detection can be performed as early as the first trimester of gestation and makes use of nuchal translucency (NT) to detect not only fetal aneuploidy such as Down syndrome, but also a wide variety of fetal defects.³ At about 18-20 weeks of gestation, most fetal malformations can already be detected by antenatal ultrasound scanning. Theoretically, embryogenesis has already been completed by this time hence visualization of major organs is already feasible and with advancing fetal development, diagnostic accuracy increases. This was supported by our finding as regards the diagnostic accuracy of ultrasound done in the second trimester compared with the ones done during the third trimester which showed an 85.7% diagnostic accuracy for the former and 92.9% for the latter. However, it should be noted that the age of gestation is not the only factor that leads to an accurate diagnosis; the machine characteristics, the patient characteristics (fetal position, amniotic fluid volume) and the skills and experience of sonographers are the other factors that affect accuracy in prenatal diagnosis using ultrasonography. Nonetheless, congenital anomaly scans during the second trimester of pregnancy have allowed recognition of fetal anomalies despite differences in accuracy, reliability and sensitivity.^{3,4,5,6}

Our data shows that only 88.4% of those referred to the clinic for significant findings of congenital anomalies on ultrasound were delivered in PGH, while the rest delivered in other birthing centers. The diagnostic accuracy of ultrasonography among these cases delivered in PGH was over 90%. A follow-up postnatal evaluation of the cases which did not deliver in PGH would have allowed a more precise calculation of this diagnostic accuracy.

Findings of congenital anomalies or birth defects, whether isolated or part of a syndrome, cause substantial anxiety among couples. Depending on whether these are major or minor anomalies, differences as to their medical or surgical significance exist.⁷ In this report, no attempt to classify the malformations was done, but for easy reference, the anomalies were grouped according to anatomical involvement and frequency. Twin pregnancies, multiple fetal anomalies and hydrops fetalis were grouped separately.

Data from literature reports cardiac anomalies as the most common type of congenital anomalies detected on prenatal ultrasound. Interestingly, there were no cardiac malformations diagnosed among the cases referred to the Genetics Clinic. This may not be a true reflection of the absence of cardiac malformations but may be due to the inability to detect such malformations without doing a more detailed evaluation of the fetal heart using fetal echocardiography. Since no long term follow-up was done in most cases, frequency of this anomaly among the cases in this review upon postnatal evaluation is not known.

In this paper, isolated malformations of the central nervous system were by far the most frequently encountered anomalies. CNS malformations are next to fetal cardiac anomalies in frequency in previous report. Severe abnormalities of the CNS can be detected as early as the first trimester, but many are seen during the second and third trimesters of pregnancy. Similarly, cases with CNS abnormalities in this report were detected during these periods with 95% of cases identified during the last trimester.

CNS abnormalities contributed almost half of all cases and unlike many reports citing neural tube defects as the most common CNS malformation, congenital hydrocephalus accounted for most of the cases (77.4%). Postnatally, 56% of cases were isolated, the rest were found to have associated anomalies in the other organ systems; 12% of which were part of a malformation sequence. The mortality rate reported for congenital hydrocephalus in association with other anomalies was 27% higher than isolated ones while the survival rate is 35.6% lower. In this series, the fetal/ neonatal mortality rate was directly related to the presence of abnormalities outside the CNS.

Neural tube defects, particularly anencephaly and encephalocele, were the next most commonly detected CNS malformation in this five-year review of cases. Many are invariably lethal with about half of the cases dying in utero, and the remainder either die in the newborn period or live to undergo a number of surgeries or suffer various neurologic sequelae. In this series, the mortality rate of neural tube defects was 100% with more than half of the cases occurring as fetal death in utero.

Holoprosencephaly, a midline defect of the brain, is caused by the failure of the prosencephalon to differentiate into cerebral hemispheres and lateral ventricle during the 8th to 10th week of gestation leading to consequential malformations in the head and face. It may be an isolated malformation or part of a syndrome and the more severe type carries a very poor prognosis with only a few surviving the first 6 months of life. All 3 cases of holoprosencephaly in this study died shortly after birth.

Abdominal wall defects such as gastroschisis and omphalocoele are fairly common fetal anomalies. Despite the wide use of ultrasonography for over three decades now, there is paucity of data regarding the accuracy of detecting abdominal wall defects antenatally. However, there are reports of errors occurring in the first trimester of pregnancy attributed to normal embryologic events. At the early part of the first trimester, there is a transient stage of physiological umbilical herniation, during which the midgut herniates into the umbilical cord and extends beyond the abdominal wall.⁹ It is only at 11 weeks of gestation that the bowels become intra-abdominal in location. More accurate detection of these defects are expected after this period. Most of the abdominal wall defects seen in this study were ascertained in the last trimester of pregnancy and had a hundred percent diagnostic accuracy.

Congenital diaphragmatic hernia (CDH), another type of abdominal defect involves the diaphragm and can cause a very complicated neonatal course. Unlike gastroschisis and omphalocoele which carry a good prognosis when isolated, CDH can cause life-long problems of respiratory insufficiency and failure to thrive or early neonatal mortality, as is the case of all the CDH cases included in this study.

Hydrops fetalis is commonly diagnosed antenatally and regardless of the cause carries a high fetal mortality rate that ranges from 37 to 90% in previous studies. ^{10, 11} In this retrospective review, three of the cases of hydrops fetalis were stillbirths and of the 8 who were born alive the mortality rate was 100%. Unfortunately, most of these hydropic fetuses did not have complete work-up as regards the possible etiology.

Multiple gestation like twinning is one of the many complexities of pregnancy and poses grave risk to both mother and fetus. Congenital anomalies increase two- to three-fold and are primarily related to zygosity of the fetus with malformation rates being higher among monozygotic twins than in dizygotic ones.¹² The anomalies commonly seen are neural tube defects, hydrocephalus, congenital heart disease, esophageal atresia and genitourinary tract anomalies. Furthermore, intrauterine fetal death in one twin may occur in up to 50% of cases of twin pregnancies. In this study, multiple gestation was accurately diagnosed by prenatal ultrasound, with only one out of the seven cases showing no abnormalities. The rest of the cases had either both or one of the twins having an abnormality of the CNS with more than half of the cases, one twin dying in utero.

There were a few cases of renal anomalies detected in this study. The 3 cases of polycystic kidney disease had a 100% accuracy as well as mortality postnatally. This condition is characterized by nephromegaly accompanied with dilated collecting tubules and hepatic cysts. It follows an autosomal pattern of inheritance and in majority of cases is lethal especially if associated with oligohydramnios. Only a very few survive at birth; a high percentage diagnosed in utero are stillborn. In contrast, congenital hydronephrosis which was seen in two of the cases but confirmed in only one neonate, is compatible with life; many cases are physiologic with spontaneous resolution at birth and rarely need surgical intervention.

Because thanatophoric dysplasia (TD) and achondroplasia share sonographic features of shortened limbs on ultrasound, there were two cases of TD in this review which were incorrectly diagnosed as achondroplasia prenatally. It is important to differentiate the two genetic conditions because thanatophoric dysplasia is lethal while achondroplasia is compatible with life and the approach to obstetric and neonatal management may differ due to the natural history and prognosis.

Although the diagnostic accuracy of detecting these congenital anomalies prenatally is over 90%, the neonatal mortality and morbidity remain high and in-utero management of congenital anomalies are still not readily available.

Conclusion

Prenatal ultrasonography remains as a widely used non-invasive method for prenatal screening of high-risk pregnancies, offering diagnostic information at any age of pregnancy and has potential benefit to both the mother and the fetus if performed accurately. Over the last five years, prenatal ultrasound findings of congenital anomalies have increasingly become a major reason for referral to the Genetics Clinic. Malformations of the central nervous system are the most frequent congenital malformations seen on ultrasound followed by defects in the abdominal wall. The diagnostic accuracy of prenatal ultrasound in PGH is high and is directly proportional to the age of gestation when it is performed. Although these fetal anomalies can be diagnosed accurately in the majority of cases, neonatal mortality and morbidity remain high.

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