Fetal Abnormalities in the Fetal Anomaly Clinic at Siriraj Hospital

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Abstract : **In order to reduce perinatal morbidity and mortality rates, a Fetal Anomaly Clinic has been set up at Siriraj Hospital. The aims are to identify abnormal fetuses and manage them in an appropriate way before delivery. From 1st May, 2000 to 30th April, 2002, 142 pregnancies with fetal abnormalities out of 10,386 pregnant women examined were found at the Fetal Anomaly Clinic, Siriraj Hospital. There were 32 fetuses (22.5%) with CNS abnormalities and neural tube defects with a mean gestational age at diagnosis of 18.6 weeks, 12 fetuses (8.5%) with gastrointestinal abnormalities with a mean gestational age at diagnosis of 17.1 weeks, 12 cases (8.5%) with cardiovascular and pulmonary abnormalities with a mean gestational age at diagnosis of 24.8 weeks, 10 cases (7%) with skeletal abnormalities with mean gestational age at diagnosis of 26.7 weeks, 15 cases (10.6%) with chromosome abnormalities with a mean gestational age at diagnosis of 21.7 weeks, 18 cases (12.6%) of KUB abnormalities with a mean gestational age at diagnosis of 25.6 weeks, and 43 cases (30.3%) of other abnormalities with a mean gestational age at diagnosis of 24.5 weeks. Counsellings was given before making a decision and all abnormalities were confirmed by autopsy, amniocentesis, paediatric neonatologists or paediatric surgeons.**

เรื่องย่อ : **ความผิดปกติของทารกในคลินิกทารกที่มีความผิดปกติที่โรงพยาบาลศิริราช**

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เพื่อที่จะลดอัตราความพิการและอัตราตายของทารกแรกคลอด คลินิกทารกที่มีความผิดปกติได้ถูกจัดตั้งขึ้นมา โดยมีวัตถุประสงค์เพื่อตรวจหาทารกที่มีความผิดปกติในระหว่างการตั้งครรภ์ และให้การดูแลรักษาทารกก่อนที่จะคลอดออกมา พบความผิดปกติของทารกจำนวน 142 รายจากสตรีตั้งครรภ์จำนวน 10,386 ราย

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ที่มารับการตรวจอัลตราซาวด์ที่คลินิกทารกที่มีความผิดปกติระหว่างวันที่ 1 พฤษภาคม 2543 ถึง 30 เมษายน 2545 โดย 32 ราย (22.5%) เป็นทารกที่มีความผิดปกติทางด้านระบบประสาทและไขสันหลังและให้การวินิจฉัยได้ในช่วงอายุครรภ์เฉลี่ย 18.6 สัปดาห์, 12 ราย (8.5%) เป็นทารกที่มีความผิดปกติของระบบทางเดินอาหารและให้การวินิจฉัยได้ในช่วงอายุครรภ์เฉลี่ย 17.1 สัปดาห์, 12 ราย (8.5%) เป็นทารกที่มีความผิดปกติของระบบหลอดเลือดหัวใจและทางเดินหายใจและให้การวินิจฉัยได้ในช่วงอายุครรภ์เฉลี่ย 24.8 สัปดาห์, 10 ราย (7%) เป็นทารกที่มีความผิดปกติของระบบกระดูกและให้การวินิจฉัยได้ในช่วงอายุครรภ์เฉลี่ย 26.7 สัปดาห์, 15 ราย (10.6%) เป็นทารกที่มีความผิดปกติของโครโมโซมและให้การวินิจฉัยได้ในช่วงอายุครรภ์เฉลี่ย 21.7 สัปดาห์, 18 ราย (12.6%) เป็นทารกที่มีความผิดปกติของระบบทางเดินปัสสาวะและให้การวินิจฉัยได้ในช่วงอายุครรภ์เฉลี่ย 25.6 สัปดาห์, 43 ราย (30.3%) เป็นทารกที่มีความผิดปกติของระบบอื่น ๆ และให้การวินิจฉัยได้ในช่วงอายุครรภ์เฉลี่ย 24.5 สัปดาห์ ทุกรายจะได้รับคำปรึกษาแนะนำก่อนที่จะตัดสินใจอื่นๆ และความผิดปกติทุกอย่างจะถูกยืนยันโดยการตรวจทางพยาธิวิทยา, การเจาะตรวจน้ำคร่ำโดยกุมารแพทย์หรือกุมารศัลยแพทย์

INTRODUCTION

The incidence of fetal abnormalities is about 2% of newborns.[1-3] They are the commonest single identificable cause of death of a baby in pregnancy and early childhood[3] and often result in considerable disability in survivors. This results in high perinatal mortality and morbiditiy rates which could be reduced by a screening programme to detect those at risk of abnormality and to enable accurate diagnosis and management. Ultrasound is a non-invasive tool which is used by many centers around the world. In the Department of Obstetrics and Gynaecology, Siriraj Hospital, an Anomaly Clinic has been set up and running for 2 years. The objective of this clinic is to detect fetuses who are at risk of an anomaly and to provide information that will assist the best possible management before delivery. This is expected to reduce perinatal mortality and morbidity rates and to provide a basis for a standard screening programme for pregnant women in the future.

MATERIALS AND METHODS

This was a cross-sectional prospective study, conducted at the Maternal-Fetal Medicine Unit, Department of Obstetrics and Gynaecology, Siriraj Hospital, Mahidol University, Bangkok, Thailand. A total of 10,386 pregnant women were scanned during the period from 1st May, 2000 to 30th April, 2002. Fetal abnormalities were found in 142 pregnant women in this group.

Pregnancies were dated using menstrual dates or an early scan during the first trimester. Ultrasound scans were performed, using a 3.5 MHz curvilinear transducer on a Toshiba Acocee or Aloka SSD-1700 machine. Anomaly scans were normally performed between 18-20 weeks gestation in all pregnant women who came to scan at this gestation. Scans, before or after 18-20 weeks, were also performed in women in whom it was indicated, including those with abnormal vaginal bleeding, advanced maternal age, previous fetal anomaly, suspected IUGR and maternal diabetes mellitus. Fetal abnormalities were recorded according to the type of abnormality. Parental counselling was performed after the ultrasound scan. After counselling, patients chose to terminate or continue the pregnancy. Serial ultrasound were performed in order to detect the progress of the abnormality if the parents decided to continue the pregnancy. Paediatric neonatologists were also consulted in order to plan the delivery.

RESULTS

One hundred and fourty two fetal abnormalities were found in 10,386 pregnant women at a mean gestational age of 26.6 weeks as shown in table 1. The abnormalities were classified as abnormalites

of the central nervous system (CNS) including neural tube defects, gastrointestinal abnormalities, cardiovascular and pulmonary abnormalities, skeletal abnormalities, chromosome abnormalities, KUB abnormalities, and others as shown in tables 2-8 respectively. The mean gestational age at diagnosis is also presented for each type of abnormality.

Table 1. Types of abnormalities

Types	Number of cases
CNS and neural tubes defect	32
KUB abnormalities	18
Hydrops fetalis	18
Chromosome abnormalities	15
Gastrointestinal abnormalities	12
Cystic hygroma	11
Skeletal abnormalities	10
Cardiovascular abnormalities	8
Pulmonary abnormalities	4
Amniotic band syndrome	2
Umbilical cord abnormalities	1
Other syndromes and abnormalities	11
Total	142

Mean gestational age at diagnosis was 26.6 weeks.

Table 2. Details of CNS abnormalities and neural tube defects

Types	Number of cases
Hydranencephaly	7
Anencephaly	6
Ventriculomegaly	5
Holoprosencephaly	3
Dandy walker malformation	2
Schizencephaly	2
Agenesis of corpus callosum	2
Porencephaly	1
Encephalocoele	1
Microcephaly	1
Posterior fossa cyst	1
Choroid plexus cyst	1
Total	32

Mean gestational age at diagnosis was18.6 weeks. The diagnoses were confirmed by autopsy in 22 cases (68.7%).

Table 3. Details of gastrointestinal abnormalities

Types	Number of cases
Gastroschisis	7
Omphalocoèle	2
Duodenal atresia	1
Intraabdominal mass	1
Bowel dilated	1
Total	12

Mean gestational age at diagnosis was 17.1 weeks. All cases were confirmed and corrected by paediatric surgeons.

Table 4. Details of cardiovascular and pulmonary abnormalities

Types	Number of cases
Golf ball	4
Second degree A-V block	1
Complete A-V block	1
Cardiac tumour	1
Hypoplastic left heart	1
Congenital adenomatoid malformation	1
Pleural effusion	3
Total	12

Mean gestational age at diagnosis was 24.8 weeks. All diagnoses, except golf ball and pleural effusion, were confirmed by paediatric cardiologists after delivery.

Table 5. Details of skeletal abnormalities

Types	Number of cases
Thanatophoric dysplasia	5
Achondroplasia	2
Micromelia	2
Short stature	1
Total	10

Mean gestational age at diagnosis was 26.7 weeks. Four cases of thanatophoric dysplasia and 2 cases of achondroplasia were confirmed by autopsy. The other cases had clinical short stature.

Table 6. Details of chromosome abnormalities

Types	Number of cases
Trisomy 18	6
Trisomy 21	4
Trisomy 13	3
Turner's syndrome	2
Total	15

Mean gestational age at diagnosis was 21.7 weeks and mean maternal age was 37.6 years. All cases were confirmed by amniocentesis, and termination of pregnancy were performed after counselling.

Table 7. Details of KUB abnormalities

Types	Number of cases
Renal agenesis	4
Hydronephrosis	4
UPJ obstruction	2
Multicystic disease of the kidney	2
Bilateral polycystic kidney	2
Multiple dysplastic right kidney	1
Bilateral renal agenesis	1
Bladder outlet obstruction	1
Renal pyelectasis	1
Total	18

Mean gestational age at diagnosis was 25.6 weeks. All cases of renal agenesis decided to terminate pregnancies after counselling. The other cases were confirmed by ultrasound performed by a paediatric neonatologist.

DISCUSSION

Screening for fetal abnormalities has been performed during antenatal care, especially in the second trimester of pregnancy. Improvements in ultrasound resolution have led to publications of case reports and series that have identified fetal anomalies in the first trimester. However, the sensitivity of the first trimester scan has been found to be low.[4] It is important to remember that many abnormalities can be clearly identified between 18-20 weeks and the features of other congenital anomalies may change with advancing gestation. From this study, anomalies of the central nervous system, neural tube defects, and gastrointestinal tract were most readily identified between 17-18 weeks.

There has been a study[5] which reported that screening a low-risk population using a 4-chamber view at 20 weeks of gestation might only identify 5-10% of major cardiac defects. Heart defects are the most common group of congenital abnormalities found at birth, but are poorly detected prenatally. The majority of babies with major heart defects are born to women outside the high risk group.[6]

Table 8. Details of other abnormalities

Types	Number of cases
Cystic hygroma	11
Hydrops fetalis	
Rhesus	1
Hemoglobin Bart's	3
Unknown	14
Amniotic band syndrome	2
Umbilical cord cyst	1
Cleft lip	1
Club foots	1
Clench hands	1
Other syndromes	
Cantrell's pentalogy	1
Ectodactyly-ectodermal dysplasia	1
Treacher Collins syndrome	1
Fraser syndrome	1
Kleebattschadel	1
Pfiffer's syndrome	1
Limb body wall disruption	1
Arnold-Chiari malformation	1
Total	43

The mean gestational age at diagnosis was 24.5 weeks. All syndromes were confirmed by autopsy. The pregnancy was terminated in the fetus with Hb Bart's hydrops fetalis. The single lesion cystic hygroma disappeared at follow-up. The other cases were managed by paediatric neonatologists and paediatric surgeons.

The study showed that, although a significant proportion of defects can be detected at this early gestation, a scan is not capable of screening some anomalies, and a 20-week scan should still be performed.[7] The combination of first and second trimester anomaly scans detected 81% of structural congenital abnormalities.

The severe skeletal dysplasias have an overall incidence of about 0.2/1,000. A short femur length alerts the clinician to the measurement of other long bones which will help diagnose major skeletal abnormalities.[8] From our study, fetal skeletal abnormalities were detected after 20 weeks which might be the result of our referral system which delayed the diagnosis of many fetal skeletal abnormalities. The detection rate for severe skeletal dysplasias is about 84%,[8] although for musculoskeletal abnormalities in general, detection is about 45%. Thanatophoric dysplasia was the most common finding which was clearly seen on ultrasound with definite features. However, most cases were referred from other local hospitals which resulted in the high number of presenting cases, compared to the other forms of skeletal dysplasia.

Gastroschisis and omphalocoele were the most common abdominal wall defects seen. From our study, the mean gestational age at detection of the defects was 17.1 weeks. The diagnosis of abdominal wall defects cannot be confirmed before 11 weeks and 5 days of gestation because the physiological hernia of the mid gut has not yet resolved. However, the defects can be easily seen by ultrasound and an abnormal scan in early gestation leads the parents to have serial ultrasound.

From our study, ultrasound could also detect soft markers of fetuses who had chromosome abnormalities. Fifteen cases were detected using many soft markers including ventriculomegaly, choroid plexus cysts and pyelectasis. All chromosome abnormalities were confirmed by amniocentesis. The mean maternal age was above 35 years in this group which supports the higher incidence of chromosome abnormalities at an advanced maternal age. However, ultrasound can detect features of chromosome abnormalities, especially Down's syndrome in only 5%.[9-11] Many reports have shown that between 43 and 100% of fetuses affected by Down's syndrome can be detected, depending on the combination of fetal nuchal translucency and maternal age used to identify a high risk group.[9-14] The wide variation in these results shows that the method of implementation of screening is important to achieve good results.

Many syndromes have multiple severe abnormalities which can be identified by ultrasound during the second trimester. Therefore, when multiple defects are seen, some syndromes can be defined. Further post-ultrasound scan counselling needs accurate informations. Invasive procedures, including amniocentesis, chorionic villous sampling, fetal blood sampling may be indicated to make a diagnosis. The risks of these techniques need to be discussed.[15]

Finally, it is important to examine the abnormal fetus after delivery. Other features are recognised in about 20% of cases and the diagnosis is changed in about 5%.[16,17] Post-mortem examination should be carried out in order to obtain accurate diagnosis and this will help parents decide in a subsequent pregnancy.[18]

CONCLUSION

The Anomaly Clinic in Department of Obstetrics and Gynaecology at Siriraj Hospital has just been set up. The main objective of this clinic is to perform an ultrasound scan on pregnant women at 18-20 weeks in order to confirm normality and provide the prospective parents with reassurance. The identification of fetal abnormalities and those associated with long term handicap should lead to providing unbiased information which parents can make choices about this and subsequent pregnancies.

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