DOWNLOAD PDF ULTRASOUND DIAGNOSIS OF FETAL ANOMALIES

Chapter 1 : Prenatal Diagnosis

The recent development of high-resolution ultrasound equipment has markedly improved the diagnostic accuracy of ultrasound. In particular, the introduction of high-frequency vaginal probes has enabled early diagnosis of certain fetal abnormalities from the 12th to 14th week of pregnancy.

Meta-analysis of negative likelihood ratios by nuchal translucency measurement to detect fetal cardiac anomalies. Findings Meta-analysis of the included 11 studies showed a positive likelihood ratio of 5. Evidence summary The reported sensitivity and likelihood ratios of nuchal translucency measurement to detect cardiac anomalies ranged widely by centre and condition, and generally the technique seems to have poor diagnostic value. Use of maternal serum alpha-fetoprotein to detect structural anomalies The diagnostic value and clinical effectiveness of biochemical markers including maternal serum alpha-fetoprotein to detect neural tube defects was reviewed in this section. Alpha-fetoprotein to detect neural tube defects Description of included studies Two studies were identified. The other was a caseâ€"control study in the USA comparing the ability of routine ultrasound and maternal serum alpha-fetoprotein levels to detect neural tube defects. Findings The first study, which investigated maternal serum alpha-fetoprotein as a screening test , was conducted between and in the USA and involved 27 women. The prevalence of neural tube defects was reported as 1. Sensitivity , specificity and positive and negative likelihood ratios were reported as In the caseâ€"control study , an integrated database of consecutive pregnancies between and was used. Evidence summary There were only two studies dealing with the diagnostic value and effectiveness of maternal serum alpha-fetoprotein level as a screening test. Results from a single study indicate maternal serum alpha-fetoprotein level to have good diagnostic value in predicting and ruling out structural anomalies, but evidence from another study shows it to have less value as a screening test than routine ultrasound. There is no evidence assessing the diagnostic value and effectiveness of combining maternal serum alpha-fetoprotein and routine ultrasound. As the topic was very wide, it was decided to limit the review to studies where antenatal ultrasound was used for any purpose and direct data were obtained from pregnant women. Studies and reviews about prenatal screening and diagnosis were excluded. A series of six questions was prepared, targetting: Studies were tabulated according to the question asked and data entered accordingly. Thirty-four pregnant women with isolated CPC detected during a mid-trimester scan who had already been counselled by their physicians regarding the findings at a university-based hospital in the USA were enrolled for the study. Interviews lasting approximately 15 minutes were conducted by a trained research assistant or nurse clinician at 24 weeks of pregnancy, and no information was given about CPCs by the research team. The interview included both open-ended and more specific questions, and all were audiotaped and transcribed verbatim. Common themes were identified, and several categories of responses identified for each theme. Initial validation was undertaken by an independent qualitative study consultant not involved in the research. Participants were all parents attending a fetal medicine unit in the UK with an antenatal diagnosis of surgical anomaly principally abdominal wall defects and gastrointestinal and thoracic anomalies. Women unable to read English and those booked to give birth somewhere else were excluded. Participants were asked to complete STAI after ultrasound at the fetal centre. Then each couple had a detailed consultation with the paediatric consultant and the clinical nurse specialist. Before leaving, the subjects were given a second STAI and asked to complete and return within 1 week. A control group comprising pregnant women with a normal ultrasound scan and uncomplicated pregnancy was recruited and asked to complete STAI as the other group. Non-parametric tests were used for comparison, and data are quoted as medians and interquartile ranges IQRs. There was wide variation among the selected studies in terms of questions addressed, methods used, and when and where they were conducted. The studies were not graded in terms of research quality or removed because of poor quality, although many had problems of design and reporting. This was done because, in spite of poor quality, these studies gave useful information. The main findings of the review are discussed below. Antenatal ultrasound is very attractive to pregnant women and their partners as it provides early visual confirmation of pregnancy, direct contact with their baby and reassurance about fetal wellbeing. At the same time, these features may augment the potential for feelings

of anxiety, shock and disappointment when the scan shows a problem. Recent trends in the use of ultrasound have led to more findings of uncertain clinical importance, and this is likely to have important psychological and social consequences for women. Although it was reported in earlier studies that some women feared that ultrasound might harm their babies, there is a paucity of evidence about it from the later studies. Reports of a reduction in anxiety after ultrasound examination are likely to reflect increased anxiety before the scan rather than a real benefit. No reliable evidence is available for any positive health behaviour e. None of the trials comparing ultrasound use with no ultrasound use has looked at its social and psychological impact on parents and babies. In general, participants in the second study were college educated mean years of education The mean maternal age was Mean gestational age at CPC detection was Women with positive serum screening results were less likely to describe CPC as benign compared with women with a normal serum screen OR 0. Seventy-seven percent of women reported seeking additional information about CPCs beyond that given by their provider at the original scan, with the most common source being the internet. But only half of the women with a reassuring serum screen and none with an abnormal serum screen described their reaction as temporary. Sixty-eight percent of women revealed that they continued experiencing negative emotions even after receiving the diagnostic tests results, but neither increased maternal age nor visualisation of CPC on ultrasound were associated with persistence of the initial negative response. The later emotional responses included anxiety Fifty-six pregnant women subjects 26, control 30 completed the questionnaire in the third study. Maternal age was significantly lower in subjects median No correlation was found between the score and maternal age or social class, or between maternal and paternal scores. After grouping the subjects into fetal diagnostic groups, a significant decrease in anxiety levels was found for those with anterior abdominal defects but not with cystic adenomatoid malformation. No correlation was found between the scores and maternal age. The study showed that there was a high anxiety state in both prospective mothers and fathers of fetuses diagnosed with congenital malformations on ultrasound which is over and above that associated with pregnancy. Counselling by specialist staff reduced levels of parental anxiety significantly. Evidence summary Results from a well-conducted structured review show that visual confirmation of fetal wellbeing is the primary reason why women seek ultrasound during pregnancy. There is a lack of evidence regarding its other benefits and harms. Evidence from a qualitative study indicates that detection of an isolated choroid plexus cyst on antenatal ultrasound leads to negative emotions and anxiety in the majority of women, who then seek additional information from other sources. Detection of surgically treatable congenital anomalies on antenatal ultrasound led to increased anxiety levels in the parents but counselling by specialist staff helped to alleviate it significantly. Health economics evidence In the NICE clinical guideline on diabetes in pregnancy an economic model was developed to compare the cost-effectiveness of screening for congenital cardiac malformations using a four chamber ultrasound scan versus the four chamber plus outflow tracts view. This was considered to be important because women with diabetes are at increased risk of having a baby with a cardiac malformation. It was felt that this model was also relevant for the antenatal care guideline and therefore it was adapted for the antenatal care population. The results are summarised here; futher details are provided in Appendix E. Prenatal ultrasound scanning for fetal anomalies is now undertaken at around 20 weeks rather than 18 weeks. However, the screening window should be between 18 weeks 0 days and 20 weeks 6 days. Screening later than 20 weeks 6 days may delay the diagnosis of an abnormality to a point where termination of an affected pregnancy becomes problematic and may involve additional procedures such as feticide. However, it should be remembered that where women are very overweight, performing the ultrasound scan can be very difficult and time-consuming. There is also a potential for an increase in repetitive strain injury RSI -related problems if sonographers are expected to complete all anomaly scans by 20 weeks. There are likely to be further benefits of this method for detecting congenital cardiac malformations over and above that of TGA detection the main focus of the model. It is noted that some of the reviewed literature is from the s and s when scanning equipment was less well developed. The literature on scanning for fetal heart anomalies is more recent, however. It is also important to note that detection rates very much depend on the expertise of the person scanning as well as gestation and standard of equipment. Detection rates have improved in certain areas but this is due to further training as well as to advances in technology. The prevalence of fetal anomalies and

their detection rates can be evaluated either individually or after categorising them into four groups based on the RCOG criteria â€" lethal anomalies, anomalies with possible survival and long-term morbidity, anomalies amenable to intrauterine therapy, and anomalies with possible short-term or immediate morbidity Table 9. Ultrasound cannot reassure women that their baby is normal, as many anomalies are missed. Ultrasound may not offer improved outcomes despite antenatal diagnosis , but may offer reproductive choices and the opportunity to plan intrauterine therapy or managed delivery. Evidence from a single study shows that a first-trimester scan with nuchal translucency measurement is equally effective as the second-trimester scan in detecting fetal malformation overall. However, this may not be true for individual conditions, for example spina bifida is more likely to be detected by the second-trimester scan, while anencephaly and anterior abdominal wall defects may be detected in the earlier scans. There is insufficient evidence that routine ultrasound between 10 and 24 weeks improves long-term outcomes after birth. There is no evidence to support the use of selective rather than routine ultrasound scanning for fetal anomalies, gestational age determination and the diagnosis of multiple pregnancies. Findings from an HTA review suggest a second-trimester scan is the most cost-effective strategy for screening for fetal anomalies. However, there is also evidence that each different method of screening has its advantages and disadvantages, and these often seem to balance out. No one screening method stands out as being much more cost-effective than any other. However, there is some evidence that better training leads to improved performance of fetal cardiac screening and some limited evidence that antenatal diagnosis of TGA leads to better outcome for the babies. Diagnostic accuracy of the nuchal test: Different cut-off points across centres and for different cardiac defects affected sensitivity and false positive rates, which are important considerations for women undergoing this test. Diagnostic accuracy of AFP AFP has lower diagnostic value than routine ultrasound in screening for neural tube defects. There is no evidence for effect on outcomes. However, the introduction of screening using AFP has led to a reduction in the number of affected babies born at term with neural tube defects. At the first contact with a healthcare professional, women should be given information about the purpose and implications of the anomaly scan to enable them to make an informed choice as to whether or not to have the scan. The purpose of the scan is to identify fetal anomalies and allow: If an anomaly is detected during the anomaly scan pregnant women should be informed of the findings to enable them to make an informed choice as to whether they wish to continue with the pregnancy or have a termination of pregnancy. Fetal echocardiography involving the four chamber view of the fetal heart and outflow tracts is recommended as part of the routine anomaly scan. Routine screening for cardiac anomalies using nuchal translucency is not recommended. When routine ultrasound screening is performed to detect neural tube defects, alpha-fetoprotein testing is not required. Research recommendation on screening for fetal anomalies Research should be undertaken to elucidate the relationship between increased nuchal translucency and cardiac defects.

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Chapter 2 : Ultrasound diagnosis of fetal abnormalities â€" Northwestern Scholars

Screening for fetal structural anomalies. As to the time in pregnancy at which ultrasound screening should be performed, it should be first noted that most structural anomalies are increasingly detected with advancing gestation. 11 In early pregnancy, it is possible to recognise with confidence certain types of fetal malformations, like anencephaly, which can be reliably diagnosed at

Prediction of ectopic pregnancy in women with a pregnancy of unknown location. Ultrasound Obstet Gynecol ; Symptomatic patients with early viable intrauterine pregnancy: Obstet Gynecol ; Single dose methotrexate for the treatment of ectopic pregnancy. The use of transvaginal ultrasound in the diagnosis of ectopic pregnancy. Am J Obstet Gynecol ; Practice Committee of Society of Reproductive Medicine. Medical Treatment of Ectopic Pregnancy: Overview of guidelines of off-label use of methotrexate in ectopic pregnancy: Predictors of methotrexate treatment in women with tubal ectopic pregnancy. N Engl J Med ; Time to forget the last menstrual period. Ultrasound Obstet Gynecol ; 9: Br J Obstet Gynaecol ; Underestimation of gestational age crown-rump length dating curves. Conceptual age and ultrasound measurements of gestational sac and crown-rump length in vitro fertilization pregnancies. Fertil Steril ; Pregnancy dating by fetal crown-rump length: Early and simple determination of chorionic and amniotic type in multifetal pregnancy in the first fourteen weeks of pregnancy by high frequency transvaginal ultrasound. The lambda sign at 10â€"14 weeks gestation as a predictor of chorionicity in twin pregnancies. Ultrasound Obstet Gynecol ; 7: Prenatal detection of chorionicity of triplet pregnancy by ultrasonographic examination of the ipsilon zone. Screening for fetal trisomies by maternal age and fetal nuchal translucency at 10 to 14 weeks gestation. Br J Obstet Gynaecol ; First trimester screening for fetal aneuploidy: Biochemistry and nuchal translucency. Ultrasound Obstet Gynecol ; Training of potential examiners. Ultrasound Obstet Gynecol ;8: First-trimester screening for trisomies 21 and N Engl J Med ; Cell-free DNA analysis for non-invasive examination of trisomy. What is the role of the 11 â€" 14 week ultrasound in women with negative cell-free DNA screening for aneuploidy? Diagnosis of fetal abnormalities at the 10â€"14 week scan. Rossi AC and Prefumo F.: Accuracy of ultrasonography at weeks of gestation for detection of fetal structural anomalies: Obstet Gynecol ; Syngelaki A, Chelemen T, Daklis et al. Challenges in the diagnosis of fetal non-chromosomal abnormalities at weeks. Improved detection rate of structural abnormalities in the first trimester using an extended examination protocol. Ultrasound Obstet Gyneco l; Abnormalities of the heart and great vessels in chromosomally normal fetuses with increased nuchal translucency thickness at 11â€"13 weeks of gestation. Nuchal translucency and the risk of congenital heart disease. Transvaginal ultrasonography at early pregnancy cannot be used alone for targeted organ ultrasonographic examination in a high risk population. Qualified and trained sonographers in the US can perform early fetal anatomy scans between 11 and 14 weeks. Management of twin pregnancies consisting of a complete hydatidiform mole and normal fetus.

Chapter 3 : First Trimester Ultrasound Diagnosis of Fetal Abnormalities

This well-referenced teaching atlas is a comprehensive and practical overview of fetal ultrasound technology, providing up-to-date diagnosis and examination guidelines for the most clinically important anomalies and diseases.

Fetus sucking thumb- 3D ultrasound images This 34 week fetus is seen sucking its thumb in these 3D ultrasound images. Again it is surface rendering that brings out the anomaly. These ultrasound images show crenation of the margins of the lenses which appear highly prominent in the fetal eyes. These ultrasound appearances are diagnostic of fetal cataract. Fetal cataract can be diagnosed as early as 15 weeks of gestation by transvaginal ultrasound. Amutha Csp, MD, India. Such extensive and multiple cystic lesions are diagnostic of severe, multiple cystic hygroma lesions see ultrasound images above. The 3-D ultrasound images of this fetus show involvement of the soft tissues and muscles of the neck, producing protrusion of the tongue and associated macroglossia. This ultrasound imaging study of fetal multiple cystic hygromas of the fetus are courtesy of Dr. The ultrasound machine used here is the Toshiba Aplio system. Cystic hygromas are classified based on their location into: Limbs, bones, chest wall and scrotum etc. Our case above is primarily cervico-mediastinal with extensive involvement of the axilla and base of the tongue also and is extremely rare in occurence. The prognosis too is rather bleak in such cases. This ultrasound appearance seen best in a sagittal section of the fetal face can be appreciated in both 2-D and 3-D Ultrasound images shown above. Note the overhanging upper lip in these ultrasound images of micrognathia. Micrognathia is diagnosed usually in the third trimester because the lower jaw undergoes maximum growth in this time period of gestation. Associated with micrognathia, there is also an element of difficulty or improper Fetal swallowing resulting in polyhydramnios, which is also evident in these ultrasound images. In addition the third ultrasound image also shows evidence of clubfoot or talipes equinovarus. Micrognathia is the result of an abnormality in the development of the first branchial arch during early fetal life.

Chapter 4 : CENTRAL NERVOUS SYSTEM - DIAGNOSIS OF FETAL ABNORMALITIES - THE

Ultrasound can identify the majority of major structural fetal abnormalities. Prenatal diagnosis can lead to improved outcomes by ensuring that delivery occurs in a hospital with the necessary personnel to manage newborns who may require surgery or other specialized care.

The prenatal diagnosis of congenital heart disease: A practical approach for the fetal sonographers. J Clin Ultrasound The Four-Chamber View The structures that can be visualized in the four-chamber view are as follows: Schematic illustration of the four-chamber view. The ultrasonographer should obtain a number of views of the four chambers to gain adequate visualization of all these structures Fig. These echograms illustrate the need to obtain a number of four-chamber views of the heart. Echogram of four-chamber view showing equal-sized ventricles of normal thickness, a moderator band in the right ventricle arrow , a normal-looking interventricular septum, and a normal septum primum. The septum secundum is not apparent. A second four-chamber view in same patient clearly shows septum primum and secundum, but the moderator band is not clear. Although both ventricles should be similar in thickness and size, the right ventricle is normally characterized by the appearance of the moderator band, which consists of thickened trabeculae extending from the septum to the parietal wall of the right ventricle see Fig. Absence of the moderator band from its normal location implies corrected or L-transposition of the great arteries. In this abnormality, the right ventricle functions as the left chamber, assuming the role of pumping blood into the aorta. Thus, in contrast to D-transposition, L-transposition allows blood flow to the lungs and body to be maintained normally. Hyperechogenicity or thickness of the ventricular walls or ventricular septum, or both, is consistent with cardiomyopathy Fig. A Cardiomyopathy in four-chamber view with very thick ventricular walls. Four-chamber view of the heart showing hyperechogenicity at the base of the interventricular septum arrow , consistent with cardiomyopathy. In dextroversion, the apex of the heart points to the right side of the chest. The main differential diagnoses of this condition are cystic adenomatoid malformation of the lungs, diaphragmatic hernia, and polysplenia. In levoversion the apex of the heart points to left side of the chest excessively; the main differential diagnosis of this condition includes left pulmonary hypoplasia and a right-sided pulmonary mass. Defects in the IVS constitute the most common form of congenital heart disease and can occur in various locations within the septum. Ultrasonic visualization of small ventricular septal defects, regardless of location, may not be possible in the four-chamber plane. For example, membranous or perimembranous defects located adjacent to the aortic valve are situated below the anterior portion of the IVS and are not visualized in the four-chamber plane Fig. Such defects usually are depicted in the parasagittal plane Fig. Schematic illustration of membranous septal defect adjacent to the aortic valve and below the anterior aspect of the interventricular septum IVS. This defect is not visible in the four-chamber view of the heart. Schematic illustration of parasagittal view, showing left ventricle LV and aortic outflow tract Ao. A portion of the right ventricle RV is noted anteriorly, opposite the left atrium LA. The interventricular septum can be readily assessed in this plane. Ultrasound appearance of the parasagittal view. The septal insertion of the tricuspid valve should be visualized slightly below that of the mitral valve see Fig. If the insertion is lower, Ebstein anomaly should be considered. Interestingly, hyperechogenicity limited to the area of the mitral valve is usually inconsequential; that is, cardiomyopathy is not a consideration. The foramen ovale is the opening between both atrial septi, allowing blood to flow from the right atrium to the left. In endocardial cushion defect, a single common atrioventricular valve is noted superior to a large ventricular septal defect. Diagnostic Utility of the Four-Chamber View Because the four-chamber view depicts so many cardiac structures and is also the plane used to evaluate cardiac arrhythmia, it is included in all obstetric studies. Subsequently, Sharland and Allan 21 showed that after routinely including the four-chamber view in obstetric studies, 53 fetuses were referred for possible CHD over a period of 32 months; previously, from to , only 8 fetuses had been referred. This is understandable because the four-chamber view does not allow for assessment of the pulmonary and aortic arteries-outflow tracts that are evaluated best in the parasagittal, short axis, and great vessel views. Parasagital View The parasagittal view encompasses the left ventricle. This plane is significant

for two main reasons: The aorta at the level of the aortic valve can be readily assessed. Membranous IVS defects located adjacent to the aortic outflow tract and below the plane of the four chamber view stand a better chance of being visualized in this view. Short Axis View The short axis view Fig. It is used to evaluate the size of the aorta and pulmonary arteries at the valvular level. Echogram of short axis view, depicting the tricuspid valve TRIC and the aortic valve to its right. The pulmonary artery continues into the ductus arteriosus DA. The right branch of the pulmonary artery RT PA is seen as it diverts to pass below the aorta. Great Vessel View The great vessel view depicts the relatively parallel relationship of the superior vena cava, aorta, and pulmonary arteries after the latter two cross each other Fig. Clear visualization of this plane indicates that the crossing of the aorta and pulmonary artery is normal and virtually rules out transposition of the great vessels. Aortic or pulmonary stenosis also can be recognized in this view. Schematic drawing of the crossing of the pulmonary artery PA and the aorta Ao. Note how the branch of the pulmonary artery passes below the aorta see Fig. Echogram of GVV showing aortic stenosis with very small aortic outflow A outflow and a normal-sized pulmonary artery. Instead, the aortic outflow tract is large and the aorta is overriding. The great vessel view also allows a comparative measurement of the two arteries a short distance beyond the valvular level; the measurements should be obtained from the inner aspects of these arteries. Normally the ratio of pulmonary artery: Coarctation of the aorta most frequently occurs at the isthmus, the area of the arch at the insertion site of the ductus arteriosus. The isthmus is normally narrow in the fetus but dilates in the newborn after the ductus closes. Because narrowing in the isthmus occurs normally, in utero diagnosis of coarctation of the aorta is difficult. Echogram of normal-looking aortic arch. The normal constriction at the isthmus is not apparent in this view. Pulmonary Cystic Adenomatoid Malformation Three types of pulmonary cystic adenomatoid malformation have been characterized The cysts are large 2 cm. The prognosis depends on the size of the tumor and the degree of fetal hydrops Fig. If the fetus survives, resection of the affected portion of the lung is usually indicated. Longitudinal echogram of fetus with a large right type I cystic adenomatoid malformation. The cysts are large and up to 1. Note the marked displacement of heart to the left side of the chest. The fetus died of hydrops at 33 weeks. Note the brightly hyperechogenic mass involving the left lung and displacing the heart to the right side of the chest, since the left ventricle is contiguous with the mass. This echogram depicts a cystic adenomatoid malformation type III. The cysts are less than 1 cm in diameter, but other anomalies of the renal or gastrointestinal tract may be present. The lesion is microcystic, brightly echogenic, and large, and its presence is associated with a poor prognosis see Fig. A pulmonary cystic adenomatoid malformation can result in a right or left shift of the mediastinum see Fig. The differential diagnosis includes pulmonary sequestration and diaphragmatic hernia. Lung weight at autopsy is 2 SD below the mean. Etiology of Pulmonary Hypoplasia Etiology.

Chapter 5 : Diagnostic Ultrasound in the First Trimester of Pregnancy | GLOWM

Incorporating the graphic strength of an atlas with the educational utility of a textbook, ULTRASOUND DIAGNOSIS OF FETAL ANOMALIES is essential for helping specialists to reliably identify prenatal irregularities and disease for the best results.

Inhibin A Ultrasonography This is a non-invasive procedure that is harmless to both the fetus and the mother. High frequency sound waves are utilized to produce visible images from the pattern of the echos made by different tissues and organs, including the baby in the amniotic cavity. The developing embryo can first be visualized at about 6 weeks gestation. Recognition of the major internal organs and extremities to determine if any are abnormal can best be accomplished between 16 to 20 weeks gestation. Although an ultrasound examination can be quite useful to determine the size and position of the fetus, the size and position of the placenta, the amount of amniotic fluid, and the appearance of fetal anatomy, there are limitations to this procedure. Subtle abnormalities may not be detected until later in pregnancy, or may not be detected at all. A good example of this is Down syndrome trisomy 21 where the morphologic abnormalities are often not marked, but only subtle, such as increased nuchal translucency the subcutaneous space between skin surface and underlying cervical spine. Enough amniotic fluid is present for this to be accomplished starting about 14 weeks gestation. For prenatal diagnosis, most amniocenteses are performed between 14 and 20 weeks gestation. However, an ultrasound examination always proceeds amniocentesis in order to determine gestational age, the position of the fetus and placenta, and determine if enough amniotic fluid is present. In the third trimester of pregnancy, the amniotic fluid can be analyzed for determination of fetal lung maturity. This is important when the fetus is below 35 to 36 weeks gestation, because the lungs may not be mature enough to sustain life following birth. This is because the lungs are not producing enough surfactant. After birth, the infant could develop respiratory distress syndrome from hyaline membrane disease. The amniotic fluid can be analyzed by looking for an appropriate number of lamellar bodies. Other tests for fetal lung maturity include: These tests have poor positive predictive value for respiratory distress, so the decision to do amniocentesis can be made by consideration of issues around gestational age and urgency of delivery. Risks with amniocentesis are uncommon, but include fetal loss and maternal Rh sensitization. The increased risk for fetal mortality following amniocentesis is about 0. Rh negative mothers can be treated with RhoGam. Contamination of fluid from amniocentesis by maternal cells is highly unlikely. If oligohydramnios is present, then amniotic fluid is difficulk to obtain. It is sometimes possible to instill saline into the amniotic cavity and then remove fluid for analysis. Chorionic Villus Sampling CVS In this procedure, a catheter is passed via the vagina through the cervix and into the uterus to the developing placenta under ultrasound guidance. An alternative approach is transabdominal. The introduction of the catheter allows sampling of cells from the placental chorionic villi. These cells can then be analyzed by a variety of techniques. The most common test employed on cells obtained by CVS is chromosome analysis to determine the karyotype of the fetus. The cells can also be grown in culture for biochemical or molecular biologic analysis. CVS can be safely performed between 9. CVS has the disadvantage of being an invasive procedure, and it has a small but significant rate of morbidity for the fetus; this loss rate is about 0. Rarely, CVS can be associated with limb defects in the fetus. The possibility of maternal Rh sensitization is present. There is also the possibility that maternal blood cells in the developing placenta will be sampled instead of fetal cells and confound chromosome analysis. Maternal blood sampling for fetal DNA This technique makes use of the phenomenon of fetal blood cells gaining access to maternal circulation through the placental villi. Ordinarily, only a very small number of fetal cells or cell free DNA enter the maternal circulation in this fashion not enough to produce a positive Kleihauer-Betke test for fetal-maternal hemorrhage. The sequencing of maternal plasma cell-free DNA cfDNA testing can detect fetal autosomal aneuploidy, but without the risks that invasive procedures inherently have. Fluorescence in-situ hybridization FISH is another technique that can be applied to identify particular chromosomes of the fetal cells recovered from maternal blood and diagnose aneuploid conditions such as the trisomies and monosomy X. The problem with this technique is that it is difficult to get large amounts of fetal DNA. There may not be

enough to reliably determine anomalies of the fetal karyotype or assay for other abnormalities. Ordinarily, only a small amount of AFP gains access to the amniotic fluid and crosses the placenta to maternal blood. However, when there is a fetal defect in the body wall, such as a neural tube defect from failure of part of the embryologic neural tube to close, then there is a means for escape of more AFP into the amniotic fluid. Neural tube defects include anencephaly failure of closure at the cranial end of the neural tube and spina bifida failure of closure at the caudal end of the neural tube. The incidence of such defects is less than 1 per in the United States. Also, if there is an omphalocele or gastroschisis both are defects in the fetal abdominal wall , the MSAFP will be higher. In order for the MSAFP test to have the greatest utility, the gestational age must be known with certainty. Also, the race of the mother and presence of gestational diabetes are important to know, because the MSAFP can be affected by these factors. The greater the MoM, the more likely a defect is present. The MSAFP has the greatest sensitivity between 16 and 18 weeks gestation, but can still be useful between 15 and 22 weeks gestation. The most common cause for an elevated MSAFP is a wrong estimation of the gestational age of the fetus. Using a combination of MSAFP screening and ultrasonography, almost all cases of anencephaly can be found, and most cases of spina bifida. Neural tube defects can be distinguished from other fetal defects such as abdominal wall defects by use of the acetylcholinesterase test performed on amniotic fluid obtained by amniocentesis--if the acetylcholinesterase is elevated along with MSAFP then a neural tube defect is likely. If the acetylcholinesterase is not detectable, then some other fetal defect is suggested. Prevention of many neural tube defects can be accomplished by supplementation of the maternal diet with just 4 mg of folic acid per day, but this vitamin supplement must be taken a month before conception and through the first trimester. Maternal serum beta-HCG This test is most commonly used as a test for pregnancy. Beginning about a week following conception and implantation of the developing embryo into the uterus, the trophoblast will produce enough detectable beta-HCG the beta subunit of human chorionic gonadotropin to diagnose pregnancy. Thus, by the time the first menstrual period is missed, the beta-HCG will virtually always be elevated enough in maternal urine to provide a positive pregnancy test. The beta-HCG can also be quantified in serum from maternal blood, and this can be useful early in pregnancy when threatened abortion or ectopic pregnancy is suspected, because the amount of beta-HCG will be lower than expected. Later in pregnancy, in the middle to late second trimester, the beta-HCG can be used in conjunction with the MSAFP to screen for chromosomal abnormalities, and Down syndrome in particular. Very high levels of HCG suggest trophoblastic disease molar pregnancy. The absence of a fetus on ultrasonography along with an elevated HCG suggests a hydatidiform mole. The HCG level can be used to follow up treatment for molar pregnancy to make sure that no trophoblastic disease, such as a choriocarcinoma, persists. Maternal serum unconjugated estriol The amount of unconjugated estriol in maternal serum is dependent upon a viable fetus, a properly functioning placenta, and maternal well-being. The substrate for estriol begins as dehydroepiandrosterone DHEA produced in the fetus. This is further metabolized in the placenta to estriol. The estriol crosses to the maternal circulation and is excreted by the maternal kidney in urine or by the maternal liver in the bile. The measurement of serial estriol levels in the third trimester will give an indication of general well-being of the fetus. If the estriol level drops, then the fetus is threatened and delivery may be necessary emergently. Estriol tends to be lower when Down syndrome is present or when there is adrenal hypoplasia with anencephaly. Inhibin-A Dimeric inhibin-A is secreted by the placenta and by the maternal ovarian corpus luteum. Dimeric inhibin-A can be measured in maternal serum. An increased level of inhibin-A is associated with an increased risk for trisomy A high inhibin-A may also be associated with risk for preterm delivery. In addition, low PAPP-A levels in the first trimester may predict an adverse pregnancy outcome, including a small for gestational age SGA baby or stillbirth. The "quadruple screen" adds inhibin-A. Approach to diagnosis of trisomy 21 can be based upon timing. Second-trimester quadruple screening with measurement of AFP, beta-HCG, unconjugated estriol, and inhibin A has a high diagnostic yield. Both can be combined to provide high rates of detection of trisomy

Chapter 6 : Radiology | Ultrasound Diagnosis of Fetal Anomalies

A comprehensive and practical overview of fetal ultrasound technology, providing up-to-date diagnosis and examination guidelines for the most clinically important anomalies and diseases.

The corpus callosum is a bundle of fibers that connects the two cerebral hemispheres. It develops at weeks of gestation. Agenesis of the corpus callosum may be either complete or partial usually affecting the posterior part. Agenesis of the corpus callosum is found in about 5 per 1, births. Agenesis of the corpus callosum may be due to maldevelopment or secondary to a destructive lesion. It is commonly associated with chromosomal abnormalities usually trisomies 18, 13 and 8 and more than genetic syndromes. Agenesis of the corpus callosum is demonstrated in the mid-coronal and mid-sagittal views, which may require vaginal sonography. This depends on the underlying cause. The condition is classified into a Dandy-Walker malformation complete or partial agenesis of the cerebellar vermis and enlarged posterior fossa , b Dandy-Walker variant partial agenesis of the cerebellar vermis without enlargement of the posterior fossa , and c mega-cisterna magna normal vermis and fourth ventricle. Dandy-Walker malformation is found in about 1 per 30, births. Ultrasonographically, the contents of the posterior fossa are visualized through a transverse suboccipito-bregmatic section of the fetal head. Enlarged cisterna magna is diagnosed if the vertical distance from the vermis to the inner border of the skull is more than 10 mm. Download a K clip on Dandy-Walker malformation Prenatal diagnosis of isolated partial agenesis of the vermis is difficult and a false diagnosis can be made prior to 18 weeks gestation, when the formation of the vermis is incomplete and anytime in gestation, if the angle of insonation is too steep. Experience with apparently isolated partial agenesis of the vermis or enlarged cisterna magna is limited and the prognosis for these conditions is uncertain. Microcephaly means small head and brain. Microcephaly is found in about 1 per 1, births. This may result from chromosomal and genetic abnormalities, fetal hypoxia, congenital infection, and exposure to radiation or other teratogens, such maternal anticoagulation with warfarin. It is commonly found in the presence of other brain abnormalities, such as cephalocele or holoprosencephaly. The diagnosis is made by the demonstration of brain abnormalities, such as holoprosencephaly. In cases with apparently isolated microcephaly it is necessary to demonstrate progressive decrease in the head to abdomen circumference ratio to below the 1st centile with advancing gestation. Such diagnosis may not be apparent before the third trimester. In microcephaly there is a typical disproportion between the size of the skull and the face. The brain is small, with the cerebral hemispheres affected to a greater extent than the midbrain and posterior fossa. Megalencephaly means large head and brain. Megalencephaly is a very rare abnormality. This is usually familiar with no adverse consequence. However, it may also be the consequence of genetic syndromes, such as Beckwith-Wiedemann syndrome, achondroplasia, neurofibromatosis, and tuberous sclerosis. Unilateral megalencephaly is a sporadic condition. The diagnosis is made by the demonstration of a head to abdomen circumference ratio above the 99th centile without evidence of hydrocephalus or intracranial masses. Unilateral megalencephaly is characterized by macrocrania, a shift in the midline echo, borderline enlargement of the lateral ventricle and atypical gyri of the affected hemisphere. Isolated megalencephaly is usually an asymptomatic condition. Unilateral megalencephaly is associated with severe mental retardation and untreatable seizures. In porencephaly there are cystic cavities within the brain that usually communicate with the ventricular system, the subarachnoid space or both. Schizencephaly is associated with clefts in the fetal brain connecting the lateral ventricles with the subarachnoid space. Destructive cerebral lesions are found in about 1 per 10, births. Hydranencephaly is a sporadic abnormality that may result from widespread vascular occlusion in the distribution of the internal carotid arteries, prolonged severe hydrocephalus, or an overwhelming infection such as toxoplasmosis or cytomegalovirus. Porencephaly may be caused by infarction of the cerebral arteries or hemorrhage into the brain parenchyma. Schizencephaly may be a primary disorder of brain development or it may be due to bilateral occlusion of the middle cerebral arteries. Download a K clip on schizencephaly Diagnosis: Complete absence of echoes from the anterior and middle fossa distinguishes hydranencephaly from severe hydrocephalus in which a thin rim of remaining cortex and the midline echo can always be identified. In porencephaly there is one or more cystic

area in the cerebral cortex, which usually communicates with the ventricle; the differential diagnosis is from intracranial cysts arachnoid, glioependymal that are usually found either within the scissures or in the midline and compress the brain. In schizencephaly there are bilateral clefts extending from the lateral ventricles to the subarachnoid space, and is usually associated with absence of the cavum septum pellucidum. Hydranencephaly is usually incompatible with survival beyond early infancy. The prognosis in porencephaly is related to the size and location of the lesion and although there is increased risk of impaired neurodevelopment in some cases development is normal. Schizencephaly is associated with severe neurodevelopmental delay and seizures. Arachnoid cysts are fluid-filled cyst contained within the arachnoid space Prevalence: Arachnoid cysts are extremely rare Etiology: Unknown; infectious process has been hypothesized but it is unlikely that this may explain the congenital cysts Diagnosis: Arachnoid cysts appear on antenatal ultrasound as sonolucent lesions with a thin regular outline that do not contain blood flow, do not communicate with the lateral ventricles and anyhow are not associated with loss of brain tissue. They occur most frequently in the area of the cerebral fissure and in the midline. Large cyst may cause significant mass effect and the distinction from porencephaly may be difficult. Interhemispheric cysts associated with agenesis of the corpus callosum most likely are not arachnoid cysts, but rather glioependymal cysts. Large cysts may cause intracranial hypertension and require neurosurgical treatment. Spontaneous remission has been described both in the postnatal as well as in the antenatal period. Glioependymal cyst, that should be suspected in those cases with associated agenesis of the corpus callosum probably reflect a greater degree of derangement in the development of the brain and this may be reflected in a worse outcome. These cysts, which are usually bilateral, are in the choroid plexuses of the lateral cerebral ventricles. The choroid plexus is easily visualized from 10 weeks of gestation when it occupies almost the entire hemisphere. Thereafter and until 26 weeks, there is a rapid decrease in both the size of the choroid plexus and of the lateral cerebral ventricle in relation to the hemisphere. Choroid plexus cysts contain cerebrospinal fluid and cellular debris. Single or multiple cystic areas greater than 2 mm in diameter in one or both choroid plexuses. They are usually of no pathological significance, but they are associated with an increased risk for trisomy 18 and possibly trisomy
In the absence of other markers of trisomy 18 the maternal age-related risk is increased by a factor of 1. This is a midline aneurismal dilation of the vein of Galen due to an arteriovenous malformation with major hemodynamic disturbances. Vein of Galen aneurysm is a very rare abnormality. Vein of Galen aneurysm is a sporadic abnormality. The diagnosis is made by the demonstration of a supratentorial midline translucent elongated cyst. Color Doppler demonstrates active arteriovenous flow within the cyst. There may be associated evidence of high-output heart failure. Download a 1 MB clip of a vein of Galen aneurysm Prognosis: In later life hydrocephalus and intracranial hemorrhage may develop. Good results can be achieved by catheterization and embolization of the malformation.

Chapter 7 : A Gallery of High-Resolution, Ultrasound, Color Doppler & 3D Images - Fetal face and neck

Read "Ultrasound Diagnosis of Fetal Anomalies" by Michael Entezami with Rakuten Kobo. Recent advances in ultrasound technology have dramatically advanced prenatal care, and its use is now standard.

Chapter 8 : Prenatal diagnosis of congenital anomalies

Fetal brain MRI at â‰¥32 weeks for diagnosis of abnormalities that are not detectable by ultrasound, such as grey matter heterotopias, late sulcation and migration anomalies. Follow up: Follow-up should be standard.

Chapter 9 : The Fetal Medicine Foundation

First Trimester Ultrasound Diagnosis of Fetal Abnormalities is an authoritative, systematic guide to the role of first trimester ultrasound in pregnancy risk assessment and the early detection of fetal malformations.