ULTRASOUND DIAGNOSIS OF FETAL ANOMALIES

By

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SUMMARY

Among the 1737 consecutive antenatal subjects undergoing routine sonographic evaluation during the 2nd or 3rd trimester there were 27 fetal anomalies (1.55%). Of them 22 (81.48%) were diagnosed antenatally by ultrasound. A false positive diagnosis was made on 3 occasions.

Commonest fetal anomaly located was neural tube defect (29.63%), either anencephaly (4) or spina bifida (4). The next common was congenital hydrocele (14.82%) Other significant anomalies diagnosed included hydrocephalus (2), microcephaly (2) cleft lip (2) and urinary tract abnormalities which included multicystic dysplastic kidney (1) and obstructive uropathy (1). Other interesting anomalies diagnosed were omphalocele (1), duodenal atresia, and achondroplasia. There were 2 subjects with fetal hydrops, immune and non-immune types one each.

We emphasise that a routine sonography, preferably around 20 weeks of gestation, should be advocated for diagnosis of fetal growth, well-being and fetal anomalies. If one anomaly is located a careful study could identify that other multiple anomalies present in the same fetus.

Introduction

Congenital anomalies or birth defects are defined as abnormalities that develop during intrauterine life and are present at birth. A major anomaly usually is defined as incompatible with life, such as anencephaly or requiring major surgery for correction such as cleft lip or congenital heart disease. Minor anomalies are defects that may not require medical or surgical intervention. Introduction of real-time ultrasonography in obstetric practice has led to prenatal detection of many fetal structural anomalies. This has the beneficial effect that those with lethal anomalies could opt for termination of pregnancy, and those with correctable anomalies could decide on intrauterine or extrauterine correction of the abnormality. Classification of anomalies detected before delivery by ultrasound has proven to be useful in designing management and in evaluation of outcome results.

In our obstetric practice we consider routine sonographic evaluation of all pregnant subjects is mandatory, and all subjects undergo atleast one sonographic examination, preferably in the midtrimester. As regards diagnosis of fetal anomalies we find that sonography is extremely valuable in confirming or excluding a defect, which has

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been clinically suspected or in subjects with high risk for fetal anomalies.

Material and Methods

Beginning in July, 1985, over a period of 3 years 1737 consecutive pregnant subjects were scanned either in the 2nd trimester or beyond. This routine scanning aimed at determination of (i) gestational age, (ii) gestational weight, (iii) fetal position, (iv) fetal well-being, (v) fetal anomalies, (vi) fetal gender, (vii) quantity of amniotic fluid, (viii) placental position, grading and abnormalities, (ix) co-existing pelvic mass, and (x) uterine anomalies.

Fetal growth and maturity were assessed by employing the multiple growth parameters wherein five basic measurements, namely, BPD, OFD, antero-posterior and transverse abdominal diameters, and femur length, and two calculations, namely, head circumference (HC) and abdominal circumference (AC), were used. Four ratios could also be checked by utilizing these measurements, namely, HC/AC, FL/BPD, FL/AC and Cephalic Index (CI).

The normal intracranial anatomy included the fetal brain, the ventricular system and the arterial system. Cerebral cortex, lateral ventricles and corpus callosum are located from above downwards; more inferiorly the double echogenic lines of cavum septi pellucidi, the third ventricle sandwitched between the thalamic bodies, the middle cerebral arteries, and the insula could be imaged. Angling the transducer posteriorly, the connection of the thalamus to the brain stem was visualized, and by increasing the angle the bilobular cerebellum, the vermis in the middle and the cisterna magna (posterior and inferior to cerebellum) were located. The normal ratio of the lateral ventricle to hemispheric width of 50% around 20 weeks and 30% at 28 weeks and beyond was recognised. The strong echogenicity of the choroid plexus could be seen in the ventricular system. In front of the cerebellar lobes the 4th ventricle and the pons could be imaged. The medulla was located at a lower level below the pons, and these structures were more clearly identified by the sagittal scan of the posterior fossa.

Increased visualization of the face could be achieved using the coronal plane, and mandible, maxilla and nares could be visualized. The distance between the orbits of the eye can be measured, and by locating the lens the movements of the eye could be evaluated.

The fetal spine was imaged in both the longitudinal and transverse planes. In the transverse plane the 3 ossification centers (2 lateral and 1 anterior) could be seen arranged in a triangle, and each segment of the spine from the calvareum to the sacrum may be examined and the integrity of the circular neural arch confirmed. In the longitudinal plane only 2 of the 3 ossification centers could be seen, and there will appear two rows of bright echogenic dots that diverge slightly in the cervical region and converge slightly in the lumbar region. In this plane the skin of the fetal back could be visualized.

The long bones of the fetal extremities were easily identified, and the fetal hand and foot were carefully examined. The clavicle and the ribs were also imaged without any difficulty.

In the chest the four-chamber view of the heart was sort for. Normally, the right and left ventricular chambers could be identified by their characteristic relations to the fetal thorax, and then the position of the tricuspid and bicuspid valves, the interventricular septum, the foramen ovale and the two atrial chambers were imaged. The surrounding lung tissue filled the remainder of the chest and has the same echogenicity

of the liver. After imaging the four-chamber view the next step was identification of the outflow tracts which include the aortic valve, aortic arch, carotid artery, and the descending aorta. The pulmonary arteries and the ductus arteriosus could be seen draping over the circular aorta in the transverse plane by rotating the transducer through 90°.

In the fetal abdomen the liver occupied most of the upper abdomen. The umbilical vein entered the liver and terminated in the portal sinus, which became the beginning of the left portal vein. The junction of the umbilical vein and the left portal vein appeared as an echolucent hockey-stick like structure. The ductus venosus appeared as a slitlike echolucency coursing between the junction of the umbilical vein and portal sinus and inferior vena cava. The gallbladder appeared as a sonolucent elongated cystic structure on the right side of the abdomen. The stomach, when filled with amniotic fluid, could be readily identified as a sonolucent cystic bubble in the upper left portion of the abdomen. Through this acoustic window, the pancreas and spleen may be visualised. Loops of large and small bowel were also visible as thin incomplete echogenic fluid filled rings. Low level echoes might represent the meconium. The fetal diaphragm was clearly located which separated the thoracic contents from the abdominal structures.

The fetal kidneys were easily identified, with the borders having a relatively echodense, ring. The echolucent renal pyramids and the renal pelvis were identified with little more difficulty. The adrenals were seen as oval-shaped echolucent structures lying immediately cephalad to the kidney in the longitudinal plane. The fetal bladder was recognized as a sonolucent cystic bubble within the fetal pelvis.

The fetal gender was determined by locating the external genitalia. In the male fetus, it was possible to visualize the penis, the scrotum and the testicles inside the scrotum. In the female fetus the labia appeared as two crescent-shaped structures of soft tissue density separated by a thin sonolucent line, which represented amniotic fluid between the labia.

Diagnosis of Fetal Anomalies

Among the 1737 obstetric patients undergoing ultrasound study in the 2nd or 3rd trimester, fetal structural anomaly was detected in 22 subjects (1.27%), and in another 5 subjects the anomalies were missed at sonography (18.52%), and this gives a total incidence of fetal anomaly of 27 numbers, i.e., 1.55%.

Neural tube defects constituted the commonest form of anomaly recognised in this series, i.e., 8 of the 27 anomalies (29.63%), and they included anencephaly (4), spina bifida (2), and menigomyelocele (2), The next common anomaly that was located was congenital hydrocele in 4 subjects (14.82%), followed by 7.41% incidence of hydrocephaly, microcephaly, cleft lip and urinary tract abnormalities. The least common type of anomalies seen included duodenal astressia, examphalos, ventricular septal defect of the heart, achondroplasia and talipus.

Neural tube defects

Anencephaly was the congenital anomaly which could be diagnosed with sufficient precision in all the 4 subjects. The diagnosis was confirmed by the absence of cerebral hemispheres and overlying skull and scalp. Majority of these subjects had polyhydramnios, and all were first seen in the third trimester. Pregnancy was terminated by amniotomy or by extra-amniotic

placement of ethacridine lactate. Equal number of fetuses with anencephaly belonged to either sex.

Spina bifida referred to a defect in the spine resulting from failure of the two halves of the vertebral arch to fuse. These lesions were usually encountered in the lumbosacral and cervical regions. When meninges protrude through this defect, the lesion will be designated meningocele, if neural tissue also protrude the same will be meningomyelocele. Sonographically spina bifida was identified as a splaying of the posterior ossification centers of the spine giving the vertebral segment a U-shaped or V-shaped appearance. The posterior ossification centers were more widely spaced than those in vertebral segements above and below the lesion. The 'fish-hook' spine in which the lumbosacral area of the fetal spine has a posterior abnormal curvature was typically present in one subject. A protruding sac representing the meningomyelocele was located in yet another subject. The 'lemon' sign and 'banana cerebellum' were not demonstrated in this series, but one subject had associated hydrocephalus.

Among the 2 subjects with spina bifida one was missed and among the two subjects with menigomyelocele one was missed at ultrasonography. Of the 4 subjects, the defects were seen in lumbosacral region in 2 subjects and cervical region in the other 2 subjects. Wherever the defect was diagnosed at sonography the pregnancy was terminated. One associated with hydrocephalus, a near term pregnancy was terminated by amniotomy, and the other subject in the mid-trimester was terminated by ethacridine instillation.

Hydrocephalus

This abnormality was diagnosed by locating the huge dilatation of the cerebral centricles by CSF. Both cases diagnosed in the third trimester, one breech presentation with spina bifida, and the other with cephalic presentation, were subjected to destructive operations to achieve safe delivery.

Microcephaly

There were 2 subjects with microcephaly. In this anomaly the head was small, and the diagnosis was based on all parameters remaining within normal limits except a biometric reduction in the skull measurements. Both pregnancies were seen near term, and they were delivered normally. Abdominal perimeter and femur length measurements and comparison of head perimeter with the accurate gestational age along with the HC : AC and HC : FL ratios could help in the diagnosis.

Fetal Hydrops

There were two subjects presenting with fetal hydrops, a state of excessive fluid accumulation into both the extravascular compartment and the body cavities, leading to the development of anasarca and ascites, and pleural effusion. Both were presenting in the third trimester with live fetuses, one was an immune hydrops in a Rh negative mother, and the other was a non-immune hydrops. Both pregnancies were terminated and resulted in neonatal death. The mother with non-immune hydrops subsequently had a normal child. Fluid accumulation was more severe in the non-immune hydrops, and both were diagnosed at routine sonographic evaluation.

Duodenal atresia

The double bubble sign, characteristic of duodenal atresia, resulting from dilation of the stomach and the proximal aspect of the duodenum, was identified in one subject

undergoing routine sonographic evaluation. Since the patient presented near term, after spontaneous delivery neonatal therapy was aimed at. However, the baby died in the neonatal period at the time of surgery.

Omphalocele

The sonographic finding in this ventral abdominal wall defect included extruded viscera (mainly the fetal liver) covered by a membranous sac and the insertion of the umbilical cord at the apex of the sac. This defect was detected in a subject with 36 weeks pregnancy, the diagnosis was confirmed at caesarean delivery, and the infant died in the neonatal period. This defect was also diagnosed during routine sonographic evaluation.

Multicystic dysplastic kidney

This subject presented with polyhydramnios in the midtrimester. The left kidney was the seat of multiple non-communicating cystic structures of variable size, with absent renal pelvis and inability to identify normal renal parenchyma. Because the defect was unilateral the fetal bladder contained urine. This condition was confused with jejunal atresia and pregnancy was terminated. Jejunal obstruction is associated with triple or quadruple 'bubble sign'. However, intestinal obstructions are not evident until the third trimester because the amount of amniotic fluid swallowed is minimal prior to 28 weeks. The diagnosis in this particular case was confirmed at autopsy.

Bladder outlet obstruction

This condition was encountered in one subject presenting with huge distension of fetal abdomen. The child was delivered spontaneously, and urethral catheterisation and dilatation relieved the problem.

Congenital hydrocele

There were 4 subjects with congenital hydrocele, and the condition was bilateral in 2 subjects. No active treatment was advocated, and spontaneous resolution was anticipated.

Achondroplasia

This condition was diagnosed in a subject with a previous history of achondroplasia. The diagnosis was suspected by 28th week of gestation and confirmed by 31st week. The typical finding was the limb shortening with no other deformity. The diagnosis was arrived at by measuring all the long bones, and comparing the measurements with that of cranial and abdominal perimeter and the gestational age.

Cleft lip

Among the two subjects presenting with this problem one was clearly diagnosed antenatally by sonography. The non-fusion between the median nasal and maxillary processes with hypertrophied tissue at the edge of the cleft was quite evident.

In addition one subject with congenital heart disease (ventricular septal defect), and another fetus with tallipes were missed at routine antenatal sonography. A false positive diagnosis was made at 3 occasions (hydrocephalus, congenital heart disease and microcephaly), and in all these situations the babies were born healthy.

Conclusion

Routine antenatal sonographic evaluation identifies a good number of fetal malformation. In this study atleast 50% of the fetal anomalies were diagnosed just by a routine sonography, and the other 50% of subjects had either a risk factor such as polyhydramnios or a previous history. We feel that a systematic ultrasound scanning around 20 weeks should be routinely advocated for early detection of fetal anomalies.