ORIGINAL ARTICLE

Early Ultrasonic Detection of Neural Tube Defects

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ASTRACT

Objective: *Evaluation of early neural tube defects by ultrasound examination.*

Study Design: *Observational study.*

Material and Methods: *Public and private hospitals of Lahore, from October 2011 to January 2014. (Amna Inayat Medical College, Avicenna Medical College, Shaikh Zayed Hospital, Fatima Memorial Hospital, National Hospital, Defence Medical Group). Ultrasound done in public and private centers of Lahore, Pakistan. Routine anomaly scan (18 – 23 weeks gestation) done for 2034 pregnant women to assess the effectiveness of diagnostic ultrasound in the diagnosis of neural tube defects. During the anomaly scan the fetus with neurological defects was recorded separately and followed up.*

Results: *Out of the 2034 pregnant women screened, 27 fetus (.01%) were diagnosed with neural tube defects. Out of these, 4 fetus had anencephaly. Two of the women opted to terminate the pregnancy immediately and the other two mothers opted to continue to term and they delivered stillborns. There were 3 women whose fetus were diagnosed with acrania. Encephalocele was seen in 5 patients. Three had frontal encephaloceles and 2 had posterior encephaloceles. Fourteen cases of spina bifida with associated congenital anomalies were detected. The most common associated anomaly was hydrocephalus (seen in 9 of the fetus), lumbar meningocele (seen in 8 of fetus). Scoliosis (seen in 6 fetus). One patient was diagnosed with hydrocephalus and after birth it was determined to be agenesis of corpus callosum.*

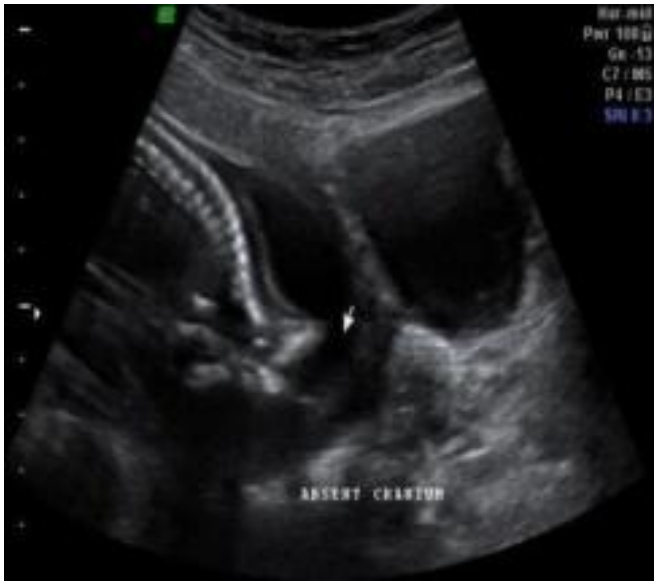
Conclusion: *Ultrasound is a beneficial investigation to make early diagnosis in neural tube defects. Anomaly scan is a routine investigation assisting the neurosurgeon in treatment and prognosis for the fetus. Such cases with neural tube defects should be assessed early and delivered in tertiary care hospitals so that early treatment may be initiated for better prognosis.*

Key Words: *Neural tube defects, anomaly ultrasound scan, spina bifida, encephalocele.*

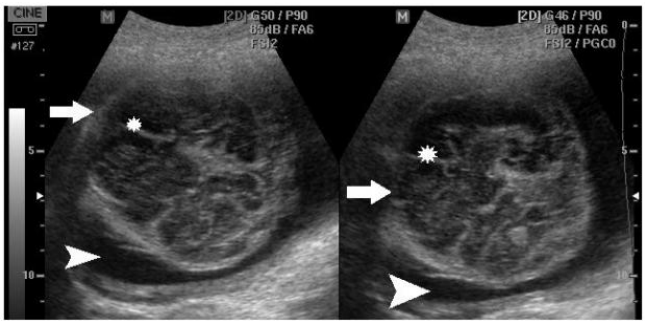
INTRODUCTION

Neural tube defects are improper development of the spinal cord and brain during intrauterine life. Central nervous system (CNS) malformations are the most frequent type of congenital anomaly second to congenital heart disease.[3] Ultrasound can accurately diagnose upto 90% of the abnormalities before 23 weeks of gestation. An accurate fetal diagnosis depends upon a precise description of the sonographic appearance of the CNS and careful evaluation for associated malformations, which are often present.[1] The abnormalities seen during intrauterine life are anencephaly, acrania, encephalocele, spina bifida, agenesis of corpus callosum and the rare anomaly of caudal regression syndrome. The most common defects detected during intrauterine life are spina bifida (abnormal development of the spine and spinal cord), encephalocele, and anencephaly (abnormal development of the brain). Anencephaly is incompatible with life but spinal abnormality may be treated with surgery immediately after birth and in certain advanced surgical centers during intrauterine life.[9] The diagnosis will determine the type and the timing of the surgery required. Anencephaly is defined as little or no tissue above the orbits and absent calvarium: parts of the occipital bone and mid brain may be present. This condition is incompatible

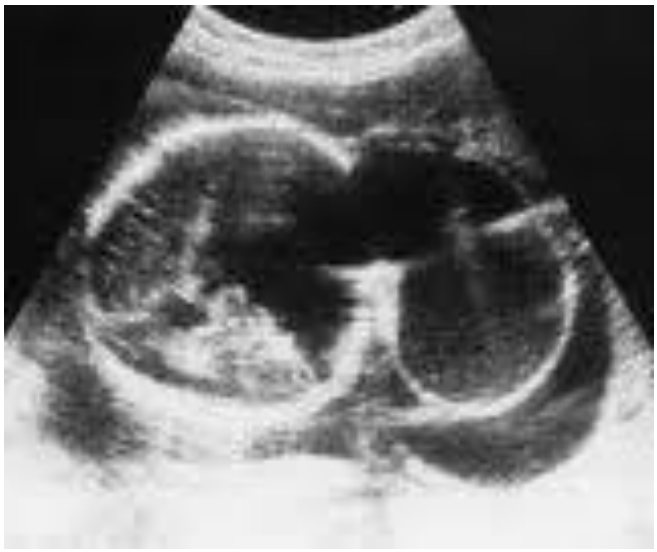
with life and early detection with early termination is accepted. Alpha feto protein is typically the highest in this abnormality. Almost 100% of cases can be determined during intrauterine life. Fetal acrania (exencephaly) is a congenital abnormality characterized by the complete or partial absence of skull bones surrounding the fetal brain. The brain may be anatomically developed but physiologically not functioning thus incompatibility with life. It can be diagnosed from 11 weeks onwards and is usually not associated with any other anomalies but polyhydramnios may be present. Fetus will usually show normal facial development and normal cardiac activity. Two major differentials for acrania is severe osteogenesis imperfecta and congenital hypophosphatasia which result in poor mineralization of the calvarium.[8] But in both cases, a thin, deformed fetal calvaria is noted. Encephalocele is a condition where the meninges and part of the brain protrude through a gap in the cranium which is usually midline in nature. It is estimated that only half survive to birth.[10] It is seen more commonly in females than males. The alpha-fetoprotein levels are not typically elevated with this defect because the defect is covered by skin. Once an encephalocele is diagnosed, a thorough examination of the baby is recommended to look for other anomalies. The encephaloceles at the back of the skull are more likely to be associated with neurologic problems and they usually have a 55% survival rate. Occasionally, an encephalocele may go undetected because of its size and location, but typically it is an obvious malformation. On ultrasound, the size of the defect, the amount of herniated brain tissue, the location, and the presence of other anomalies is important when determining the type and time of treatment. The absence of herniated brain tissue and no other associated anomalies are favorable prognostic features. Associated ultrasonographic features may include hydrocephalus and microcephaly. Spina bifida is one of the most common CNS abnormalities and 75% can be determined prenataly.[11] The two common types are spina bifida occulta (15%) and open spina bifida (85%). Majority of the cases the defect is located posteriorly where 90% are in the lumbosacral region. In open spina bifida, the most common presentation is a cystic tumor in the lumbosacral region, containing cerebrospinal fluid (CSF) and sometimes neural content covered by meninges. When the herniation sac contains only CSF, the anomaly is called meningocele; if neural tissue is present, the anomaly is called meningomyelocele. At least 80% of these case will have a raised maternal alpha fetoprotein level.[11] Spina bifida



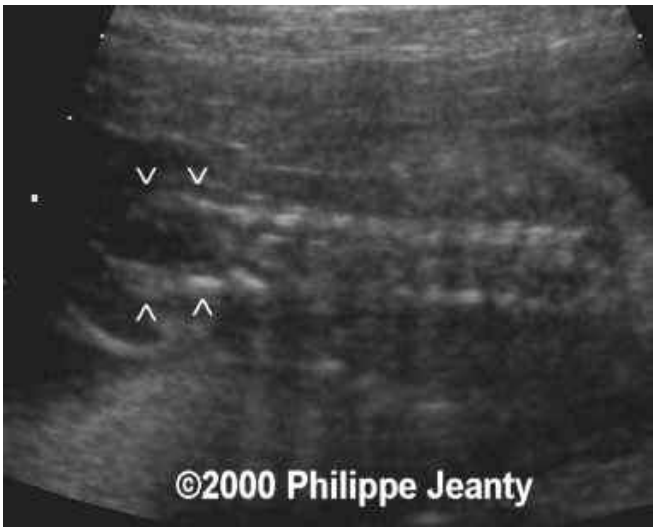
Ultrasound Image of Anencephaly



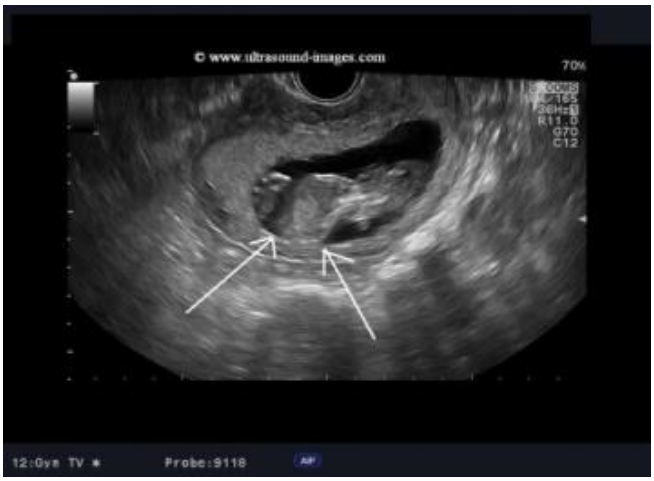
Ultrasound of Acrania



Ultrasound Image of Encephalocele



Ultrasound Image of Spina Bifida



Ultrasound Image of Caudal Regression Syndrome

occulta is more difficult to determined prenatally and usually not associated with cranial findings and will have a normal maternal alpha feto-protein level. Common associated anomalies which are ultrasonically visible are hydrocephalus, congenital talipes equinovarus and developmental dysplasia of hip, lemon and banana signs in cranium. A sonogram of the fetal brain at 14 weeks of gestation cannot detect agenesis of the corpus callosum since this structure does not become sonographically apparent until 18 to 20 weeks of gestation and does not acquire its final form until 28 to 30 weeks. But, any fetal hydrocephalus should be reassessed after birth and underlying cause should be determined.[12] Caudal regression syndrome, a rare spinal anomaly (only 300 cases reported so far), seen in 1:100000 pregnancies occurs mostly in children of diabetic mothers and is also associated with various other genitourinary, anal, vertebral, and limb anomalies.[13] The presentation and imaging appearance vary with the degree of deformity, ranging from minimal to severe regression of the coccyx, sacrum, and lumbar spine. Progressive absence of bone structures occurs in a caudal to cranial direction.

MATERIALS AND METHODS

This observational study was conducted in various public and private centers in Lahore, Pakistan from October 2011 to January 2014. Majority of patients were undergoing routine anomaly scans during the 18 to 23 weeks of gestation. Many patients had come to the hospital for the first scan and previous records were not available. All patients were explained about the reason to undergo an anomaly scan and were also briefed about the normal / abnormal features in their child. Those patients whose fetus had anomaly were counseled and explained to deliver the child in a tertiary care hospital. A total of 2034 patients were assessed during routine anomaly scan and 27 patients were diagnosed with neural tube defects. A record was kept of all fetus with congenital anomalies and the exact diagnosis on the ultrasound scan. The purpose was to assess the accuracy of ultrasound with postnatal findings. Some of the patients were not traceable but a few would return with the result of either surgical intervention or further investigations to confirm the diagnosis.

RESULTS

Out of the 2034 patients who walked in for a routine anomaly scan, 27 patients had fetus with neural tube defects. That accounts for .01% of the study. These patients were counseled and advised to deliver in tertiary care hospitals. Four fetus were diagnosed with anencephaly. Two of the mothers who were carrying anecephalic fetus opted to terminate the pregnancy immediately whereas the other two decided to continue pregnancy to full term and delivered stillborns. The detection rate for anencephaly was 100%. Three fetus were diagnosed with acrania. One of the fetus had polyhydramnios and acrania was detected during her 18 week scan. The patient carrying this fetus decided to terminate the pregnancy immediately but the other two carried till term. Their decision was based on religious reasons that since the fetus had a normal heartbeat; they wanted to continue pregnancy to term.

Table 1:

	Types	Number of Fetus Diagnosed on Ultrasound	Related Features
Anencephaly		4	
Encephalocele	Frontal	3	
	Occipital	2	
Acrania		3	Polyhydramnios in 1 fetus
Spina bifida	Lumbar	14	Hydrocephalus—9 fetus Lumbar meningoceles—8 fetus Scoliosis—6 fetus
Hydrocephalus		1	Later determined to be agenesis of corpus callosum
Total		27	

The detection rate for acrania was also 100%. Five fetus were diagnosed with encephalocele. Three had frontal encephalocele and 2 had posterior encephalocele. Upon delivery the diagnosis was again reconfirmed. Only one of the fetus survived because the defect was smaller in size, contained no herniated brain tissue, and no other associated anomalies. During the duration of the study, two fetus were delivered with encephalocele which were not diagnosed antenatally. This was due to their smaller size and posterior location. Frontal encephaloceles which were larger in size were more frequently detected whereas the posterior located and smaller encephaloceles were more occasionally missed. Thus the detection rate for encephalocele was 71%. Spina bifida was one of the more common anomalies detected and 14 fetus were diagnosed with this condition. It was usually associated with other anomalies thus the significantly easier detection. The majority of the fetus had at least one or more associated anomalies the most common being hydrocephalus. Only one of these patients was diagnosed at 30 weeks gestation with spina bifida, severe hydrocephalus, lumbar meningocele, scoliosis. That was her first scan during the pregnancy and her first visit to any hospital. She was immediately counseled and advised to remain an admitted in the hospital. But unfortunately she decided not to the remain in the hospital, and no proper follow up record is available. The detection rate during this study for spina bifida aperta was 80%. Occult spina bifida was not diagnosed in any of the fetus and was detected after birth.

DISCUSSION

Neural tube defects can be detected early during antenatal ultrasound thus allowing the parents to either terminate the pregnancy early or get early treatment for surgically amenable conditions. Some centers abroad also feature intrauterine surgery for repair of spina bifida.[9] Spina bifida is a serious congenital anomaly. The neonatal morbidity and mortality rate is estimated as 25%.[11] The majority of the children without treatment die in the first few months of life. Survival rate of those treated in the immediate neonatal period approaches 40% at seven years. Twenty five percent of these children are almost totally paralyzed, 25% require intense rehabilitation and 25% do not have a significant lower extremity dysfunction. Seventeen percent will have normal continence in long term follow up. The presence of severe hydrocephalus is considered a poor prognostic sign.[4] Early detection and a thorough fetal ultrasound may spare a child the severe consequences of spina bifida. Every pregnancy requires a thorough ultrasound exam especially during 18 – 23 weeks of gestation. In Pakistan, where many patients don't have the facility for proper ultrasound scanning, neural tube defects may be overlooked due to failure on part of the operator or poor resolution machine. So as a patient is scanned on multiple occasions during the period of gestation, it's always a good idea to rescan a new patient for anomalies even if they have crossed 23 weeks. Most of the patients in this study were being assessed in tertiary care hospitals, but there was a significant majority who were having their first ultrasound exam after 23 weeks.

CONCLUSION

A combination of early ultrasound and neural tube defect screening are an effective method of diagnosing any anomaly, thus allowing the type and timing for early treatment. The earlier the diagnosis with early treatment, the better the prognosis. It also allows parents to terminate a pregnancy which will inevitably

result in a stillborn, thus saving the mother the hardship of having to carry the fetus for the difficult duration of the pregnancy. Thus giving her a chance to conceive again as soon as possible.

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