



## Ultrasound Study of Congenital Foetal Anomalies

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### Abstract

**Objective:** The purpose of our study was to determine how well we were detecting fetal anomaly in our region and to compare most common and either preventable or not.

### Aims And Objective

- To find out the different types of anomalies,
- To find out the most common anomaly.
- To find out whether the anomaly is preventable or not

**Subjects And Methods:** Over a 2-year period, we compared the reports of sonographic studies done in 300 pregnancies at first trimester gestation with all pregnancy outcomes established by medical records and, whether born alive or dead. Fetal anomaly was defined as any structural anomaly except for those specifically excluded. We compared our results, expressed as percentage of anomalies found.

**Results:** At antenatal examination, we found 40 anomalies in 300 babies who had prenatal sonography at first trimester. Our success rate for fetal anomaly detection ranged from 31 cases with neural tube defect, 2 cases with idiopathic cause, 2 cases with hydrops, 1 case with skeletal dysplasia, 1 case with respiratory abnormalities noted. Which conformed after delivery.

**Conclusion:** Antenatal ultrasound successfully diagnosed foetal abnormalities, and out of them neural tube defects more common, which can preventable by taking folic acid supplementation.

**Keywords:** anomalies, antenatal, ultrasound, firsttrimester, neuraltube defects, hydrops.

### INTRODUCTION

Antenatal ultrasonography is very useful for evaluating foetal anatomy. Ultrasonography (US) is usually performed in the first trimester (up to 13 weeks 6 days gestation) for dating, determination of the number of fetuses, and assessment for early pregnancy complications.

The present study was undertaken to detect such anomalies. Because most anomalies are sporadic and occur in otherwise low-risk women, a

properly performed foetal anatomic survey is an essential part of obstetric screening. Indeed, the concept of a screening foetal anatomic survey during the second trimester has been widely adopted around the world. Antenatal ultrasonography can provide reassurance of a normal pregnancy in the vast majority of cases, and a systematic foetal survey can detect the majority of foetal malformations. Detection of any abnormality on the basic examination necessitates

a comprehensive and targeted anatomic survey to seek further information on the nature of the underlying abnormalities.

### MATERIALS AND METHODOLOGY

Antenatal ultrasound done in 300 patients between first trimester at Department of Radiodiagnosis, RMMCH, Chidambaram. This study is conducted between November 2013 - June 2015. The cases were selected randomly. The study was conducted in pregnant women living in and around Chidambaram.

All examinations were performed with Philips Envisor series using 5 MHz convex sector transducer and Wipro GE Logic 400MD scanner. Gestational age was assessed by menstrual history in conjunction with sonographic estimate of BPD, HC, AC, FL according to estimated normograms for these parameters.

If the menstrual history was unreliable or discrepancy between menstrual dates and sonographic gestational age was >2 weeks sonographic gestational age was accepted and serial ultrasound examination is suggested to assess the interval growth pattern.

### COMPONENTS OF FOETAL ANATOMIC SURVEY

#### HEAD AND NECK

Calvaria: Shape

Brain - documentation of thalami, lateral ventricles, cerebellum, vermis, cisterna magna, and cavum septum pellucidum includes the following views:

- Transthalamic
- Transventricular
- Transcerebellar

Face / neck

Face: lips, anterior maxilla, nose, orbits/globes, and profile view for mandible

Neck: nuchal fold

Spine: longitudinal and transverse views

#### Thorax

Heart

Four-chamber view Views of outflow tracts

Lungs

#### Abdomen

Stomach: presence, situs, and size

Gut

Anterior abdominal wall/cord insertion site

#### Genitourinary tract

Kidneys, Urinary bladder

± Genitalia

#### Extremities

Upper extremities, including both hands

Lower extremities, including both feet

Vessels: number of vessels in umbilical cord

#### Measurements

Biparietal diameter

Head circumference

Abdominal circumference

Femur

Lateral ventricular atrium cisterna magna

± Humerus

#### Others

Placenta

Amniotic fluid

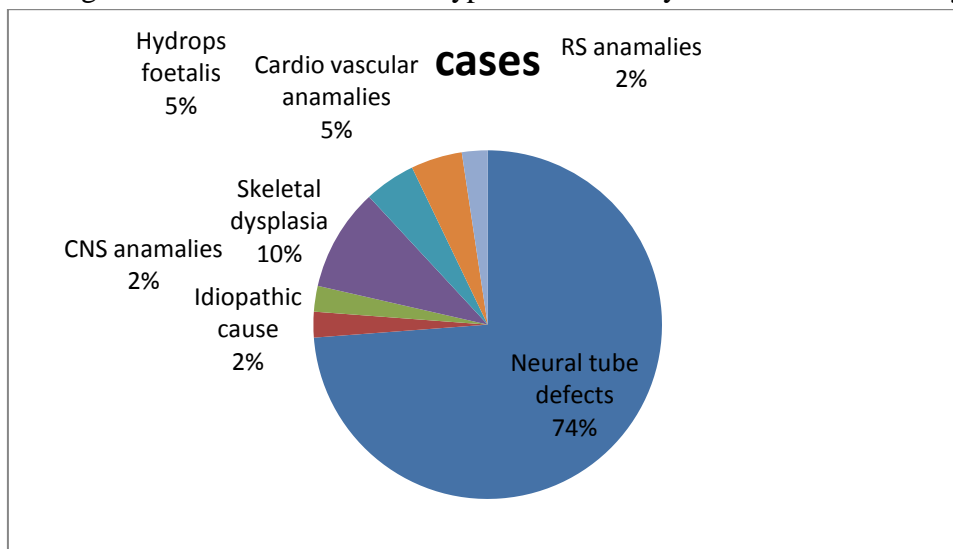
Cervix

### OBSERVATION

In this study neural tube defects are the most common anomaly associated with polyhydramnios. Anomalies are more common in multigravida patients.

Sl.No.	Type of Anomaly	No. of Cases
1	Neural Tube Defects	31
2	Idiopathic Cause	1
3	CNS Anomalies	2
4	Skeletal Dysplasia	1
5	Hydrops Foetalis	2
6.	Cardio vascular anomalies	2
6	RS anomaly	1
Total		40

**Figure -1** The Bar Diagram Shows The Different Types Of Anomaly Associated With Polyhydramnios

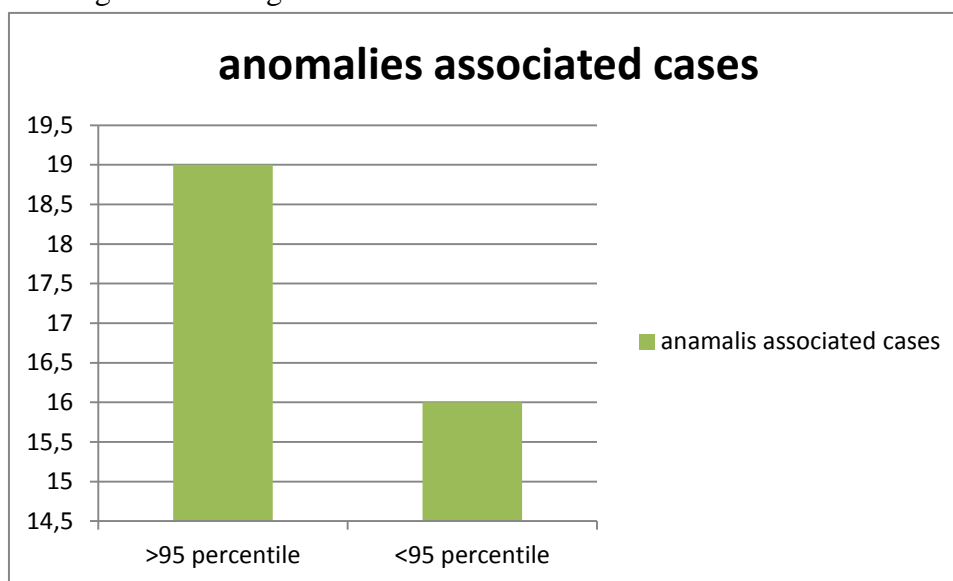


**Table – 2** The Table Showing The Value Of Amniotic Fluid Index

	No.of Cases
Amniotic Fluid Index >95 <sup>th</sup> percentile	19
Amniotic Fluid Index <95 <sup>th</sup> percentile	6

If the amniotic fluid index is more than 95<sup>th</sup> percentile. There is more probability for the presence of anomaly.

**Figure – 2** The Bar Diagram Showing The Value Of Amniotic Fluid Index



**Table – 3** The Table Showing The Value Of Obstetric Status Of The Patient

Obstetric Status of the Patient	No. of Cases
Multigravida	22
Primi gravida	18

Anomalies are more common in multigravida patients.

Figure – 3 The Bar Diagram Showing The Value Of Obstetric Status Of The Patient



Image no.1 Axial Brain Scan Showing Anencephaly.



Coronal section of brain showing gross hydrocephalous,

Image no.3



Axial section shows diaphragmatic inversion.

Image no.4



Image no.2 showing post delivery foetus



echogenic lung Imagno. Sagital section shows 5.



Axial section showing omphalocele, Image no.8,9



Sagital section of spine shows skyphosis. Image no.6



Coronal section showing short -limb,absent tibia and fibula. Image no.7



Axial section of abdomen showing omphalocele, Image no.10





Post delivery foetud confirming the findings, Image no.11



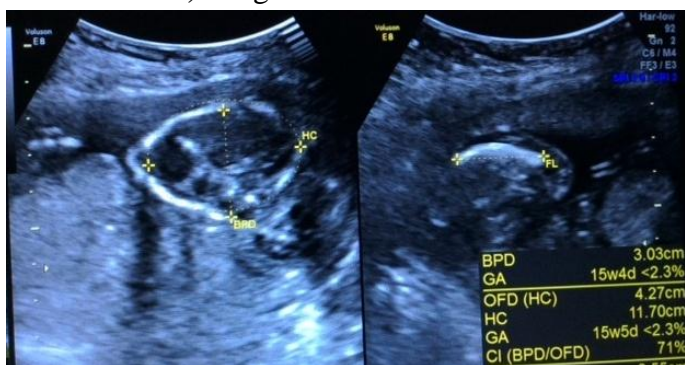
Axial section shows menigomylocele, Image no 14,15



Sagittal section shows megacystis. Image no12



Coronal section shows microcephaly and curved femur, Image no13



**RESULTS**

Antenatal ultrasound successfully diagnosed foetal abnormalities, and out of them neural tube defects more common, which can preventable by taking folic acid supplementation.

**DISCUSSION**

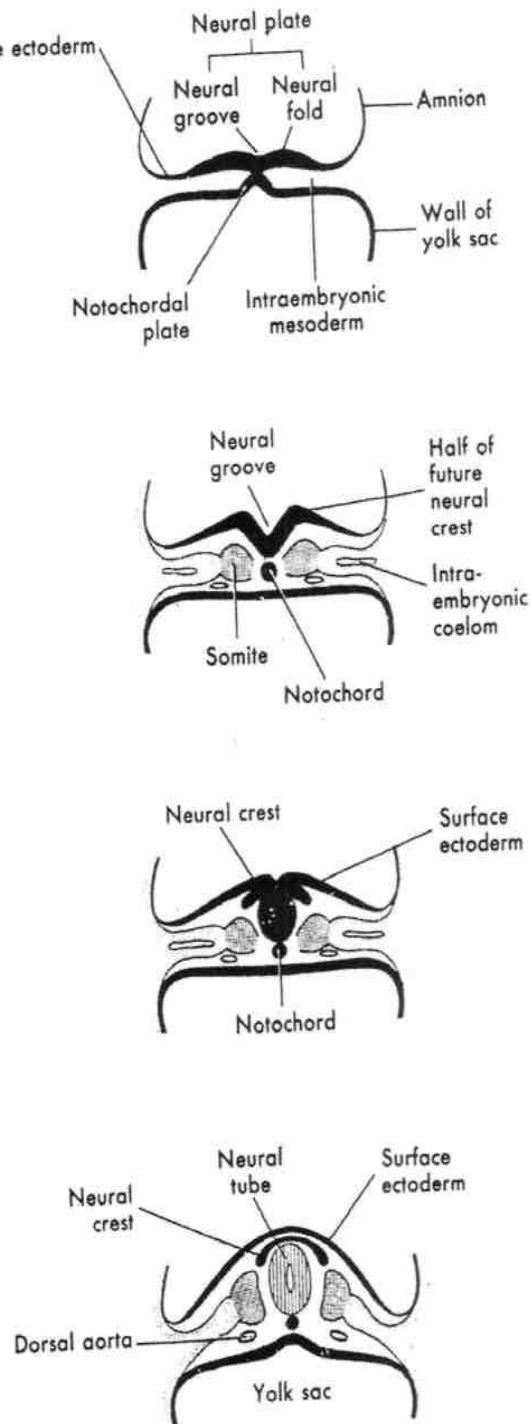
In this study neural tube defects are the most common anomaly (12) associated with polyhydramnios.

**NEUROEMBRYOLOGY**

At its very early stages the human embryo is basically a simple bilaminar disk. At about 2 weeks of embryonic life the neural plate appears on the dorsal aspect of the disk as an area of focal ectodermal proliferation. At approximately 18 days of gestation the neural plate invaginates, forming the neural groove. The lateral portions of this groove then thickened and proliferate, forming paired elevations called the neural folds.

The edges of these folds bend medially towards each other, eventually making contact and closing

over the top of the neural groove to form the neural tube. The proximal two thirds of the neural tube thickens to form brain; the caudal one third represents spinal cord. The neural tube lumen will become the brain ventricular system and the central canal of the spinal cord.



Transverse section of the embryo showing the neural plate and early development of the neural groove

## NEURAL TUBE CLOSURE

Closure of the neural tube begins in the hindbrain region and proceeds in a 'zipperlike' fashion towards both ends of the embryo. Ciliated epithelial cells lining the neural tube begin to secrete a watery liquid that distends the brain cavity, while the flaring cephalic end of the CNS constricts to form the primary brain vesicles. Failure of neural tube apposition in concert with fluid pressure that is inadequate to enlarge and form the ventricles properly is thought to result in the Chiari II malformation.

Neural tube defects (NTDs) are a heterogeneous group of malformations resulting from failure of normal neural tube closure between the third and fourth week of embryological development. Anencephaly, encephalocele, and spina bifida are the three most common forms of NTDs. Less common types of NTDs include iniencephaly, amniotic bands and other types of spinal abnormalities.<sup>30-35</sup>

Anencephaly and spina bifida are most common with nearly equal prevalence of 1 per 1,000. The prevalence is much higher during early pregnancy, especially among pregnancy losses.

Anencephaly is characterized by the absence of cranial vault and telencephalon. Necrotic remnants of the brain stem and rhombencephalic structures are covered by a vascular membrane. Associated malformations are very common. It includes spina bifida, cleft lip, cleft palate, and club foot.

The forebrain is replaced by an angiomatous mass with multiple cavities containing cerebrospinal fluid. This highly vascularized neural and mesenchymal tissues is known as AREA SUBSTANTIA CEREBRO VASCULOSA.

Spina bifida is another common anomaly involving the neural tube closure. It is subdivided into spina bifida occulta and spina bifida aperta.

Spina bifida occulta is characterized by vertebral schisis covered by normal soft tissues. Large defects are associated with pigmented and dimpled lesions overlying the skin and subcutaneous tissues.

Spina bifida aperta is a full thickness defect of the skin, underlying soft tissues and vertebral arches, exposing the neural canal. The defect may be covered by thin meningeal membrane (Meningocele).

In the presence of neural tissue inside the sac, the lesion is defined as myelo meningocele. Lumbar and lumbo sacral areas are more frequently affected.

Approximately 3% of all spontaneous abortions show evidence of an NTD. The vast majority of NTDs is sporadic and is believed to be multifactorial in origin.

It has been observed that lower socioeconomic classes have a predisposition for NTDs, leading to the theory that nutritional deficiency may be a causative factor. In 1976, Smithells et al implicated vitamin deficiencies among pregnancies with NTDs, observing decreased levels of folate, ascorbate, methionine and riboflavin in lower socioeconomic groups. This was confirmed by others.<sup>35,39</sup>

In 1991, a landmark prospective, randomized, double blind study was published that showed women who had previous pregnancies with isolated NTDs had a 72% reduction of a recurrence of the NTD when supplemented with 4mg per day of folate at least four weeks before conception through the twelfth week of gestation. In September 1992, the U.S. Public Health Service recommended that all women attempting pregnancy take the recommended daily allowance (0.4mg) of folate for 1 month before conception and for at least 3 months after conception.

Several teratogens have been implicated in the etiology of NTDs. Two anticonvulsant medications, carbamazepine and valproic acid, have been demonstrated to cause these defects, which some believe is secondary to lower levels of serum folate when on these medications. This risk has been estimated as approximately 1%. Other agents that have been implicated include zinc deficiency, hyperthermia, aminopterin, clomiphene citrate, and insulin-dependent diabetes mellitus.

The link between environmental and genetic factors has been the object of many studies, and different genes involved in the folate metabolism pathway have been investigated, including the most commonly studied thermolabile mutation (C6771) in the MTHFR gene.

The recurrence risk for NTDs varies according to different risk factors. Families who have had a child with NTD have a tenfold increase in recurrence risk in the range of 1% of 2%.

Early detection of fetal anomalies has advantages compared with detection at 18 weeks gestation. It allows time for the instigation of therapy, where appropriate. The couple can protect their privacy, since at this stage the pregnancy is not obvious; if they choose to terminate the pregnancy, the procedure can be performed on an outpatient basis. This is also associated with a more favorable cost-benefit ratio. The reassurance of normality in early pregnancy may be of particular advantage to those women who are at high risk of having an affected fetus.

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