Original Research Paper Radiology

Antenatal ultrasonic evaluation of fetal congenital anomalies

Dr. Jagruti Kalola	M.D. (Radiodiagnosis), Assistant Professor, Department of Radiodiagnosis, P.D.U. Government Medical College and Civil Hospital, Rajkot, Gujarat, India.
Dr. AnjanaTrivedi	M.D. (Radiodiagnosis), Professor - HOD, Department of Radiodiagnosis, P.D.U. Government Medical College and Civil Hospital, Rajkot, Gujarat, India.
Dr. Manish Yadav	Resident (M.D. Radiodiagnosis), Department of Radiodiagnosis, P.D.U. Government Medical College and Civil Hospital, Rajkot, Gujarat, India.
Dr. Krushnadas Radadiya	Resident (M.D. Radiodiagnosis), Department of Radiodiagnosis, P.D.U. Government Medical College and Civil Hospital, Rajkot, Gujarat, India.

ABSTRACT

Introduction: The congenital malformation constitutes one of the major causes of perinatal mortality and morbidity. The incidence mentioned in various studies is 0.8 to 5%, approximately 1:150 live births

Objective: To find out incidence of fetal congenital anomalies by antenatal Ultrasonic evaluation

Methods: In the present study pregnant women were screened by routine obstetric ultrasound.

Results: In the present series 6125 pregnant women were scanned by obstetric ultrasound out of which 50 fetuses were found to have congenital malformations

Conclusion: Ultra sound is born to obstetrics, for its simple noninvasive procedure, easy acceptability to all. With the ultrasound imaging, it is now possible to make an intrauterine diagnosis of many fetal anomalies. Important things to an accurate antenatal diagnosis are careful scanning of the fetus and knowledge of the abnormalities that may be associated with a particular anomaly.

KEYWORDS Congenital malformation, ultra sound

Introduction

The most traumatic experience for women, her husband and their family is undoubtedly the unheralded birth of a deformed child, precipitating feeling of horror, desperation, inadequacy and guilt on the parents.

The congenital malformation constitutes one of the major causes of perinatal mortality and morbidity. The incidence mentioned in various studies is 0.8 to 5% approximately 1:150 live births[1]. In high risk pregnancies incidence is more. The commonest congenital anomalies diagnosed in most of the studies were those of central nervous system[2,3].

This study is the effort to detect congenital anomalies with the help of ultrasonography. The detection of nature of congenital anomalies can help in deciding the treatment protocol for planning the delivery as well as keeping all the necessary assistance ready for managing the newborn after delivery.

Aims and Objectives

THE STUDY WAS UNDERTAKEN-

1) To find out incidence of fetal congenital anomalies by antenatal Ultrasonic evaluation in civil hospital of Rajkot in India.
2) To define the systemic distribution of fetal congenital anomalies.
3) To define the fetal anomalies in relation to age of pregnant women, and gestational age of fetus.

Material and Methods

STUDY POPULATION

In the present study pregnant women were screened by routine obstetric ultrasound.

INCLUSION CRITERIA :

Pregnant women were randomly assigned to have screening sonogram for fetal congenital anomalies.

- Group a- in this group screening sonogram was done in 2nd trimester (between 12-28 weeks)
- Group b- in this group screening sonogram was done in 3rd trimester (28 weeks onwards)

EXCLUSION CRITERIA

- Pregnant women under 12 wks of gestational age
- Women with molar pregnancy

EQUIPMENT USED

Philips IU22 and my lab 50 real time scanner. In all cases trans-abdominal ultrasound examination was done using transducer of 3.5 MHz frequency.

In the present study **6125** pregnant women were scanned by obstetric ultrasound out of which **50** fetuses were found to have congenital malformation with total of 53 anomalies involving various systems among those 50 fetuses, which were confirmed by appropriate postnatal follow up. This study was done in Department of Radiodiagnosis in a PDU Medical college civil hospital in rajkot during the period of **November 2013 – November 2015.**

Present study has brought out the following results, which need to be discussed.

Results

This study was done in Department of Radiodiagnosis in civil hospital PDU medical college Rajkot in Gujarat during the period of **November 2013 – November 2015.**

INCIDENCE OF CONGENITAL ANOMALIES

In the present series **6125** pregnant women were scanned by obstetric ultrasound out of which 50 fetuses were found to have congenital malformations

DISTRIBUTION OF ANOMALIES IN VARIOUS SYSTEMS

Total No. of anomalous fetuses – 50

Total No. of anomalies – 50

Table 1 Distribution of anomalies in various systems

Systems involved	No. of anomalies	Percentage
CNS	23	42 %
GUT	13	24 %
GIT	7	14 %
CVS	4	8%
MSK	3	6 %
RS	1	2%
OTHERS	2	4 %
TOTAL	**53**	**100 %**

DISTRIBUTION OF ANOMALIES IN CENTRAL NERVOUS SYSTEM
Table 2

Anomalies	No of anomalies	Percentage	
		within CNS	Among all anomalies
Anencephaly	8	35%	15%
Hydrocephalus	5	21%	9%
Meningo/meningomyelocele with Spina bifida	4	17%	6%
Encephalocele	1	4.5%	2%
Dandy-Walker Malformation	2	9%	4%
Holoprosencephaly	1	4.5%	2%
Acrania	1	4.5%	2%
Arnold Chiari malformation	1	4.5%	2%
TOTAL	**23**	**100%**	**42%**
		100%	

DISTRIBUTION OF ANOMALIES IN GENITO URINARY SYSTEM
Table 3

Anomalies	No of anomalies	Percentage	
		within GUT	Among all anomalies
Multicystic dysplastic kidney disease	6	46%	11%
Polycystic kidney disease (echogenic kidney)	3	23%	5%
Posterior urethral valve (Bladder outlet obstruction)	2	15%	4%
Renal agenesis	1	8%	2%
Pelvi ureteric junction obstruction	1	8%	2%
Total	**13**	**100%**	**24%**

Discussion
Congenital anomalies or birth defects are defined as the abnormality that developed during intrauterine life and are present at birth4,5. Congenital anomalies are important as a cause of stillbirth & neonatal death. In view of this, it would be imperative to evaluate the fetus during the antenatal period.

Ultrasound is widely used as a screening tool for a diagnosis of congenital anomalies .

SONOLOGICAL FEATURES OF COMMON FETAL ANOMALIES

ANOMALIES OF CENTRAL NERVOUS SYSTEM

1) Anencephaly[6,7,8] Absence of vault of skull and brain is common anomaly of fetal central nervous system. (Fig.1)

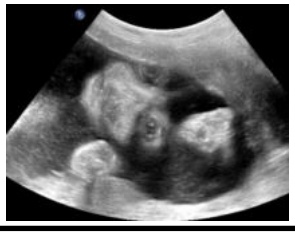

Fig 1- showing absence of skull vault of fetus with frog eye appearance

2) Hydrocephalus Hydrocephalus is defined as an increased amount of cerebrospinal fluid in ventriculosubarachnoid pathways of the brain.

Several techniques have been described to evaluate ventricular size.
1) Atrial size - above 10mm
2) Ventricle to hemispheric ratio.

Gestational Age LV/H Ratio (Normal Range)
20-24 weeks 32%
25-29 weeks 32%
30-35 weeks 31%
36-40 weeks 29%

3) Anterior horn measurement. - Combined anterior horns more than 20mm for Biparietal Diameter of under 6.5 cm (under 24 weeks)

3) HOLOPROSNCEPHALY
There are three types depending on the amount of division that occurs-

Alobar:
Single large ventricle with surrounding thin cortical mantle "boomerang or horseshoe" appearance.
Facial abnormalities are frequently associated with holoprosencephaly

Semi lobar Variety:
Rudimentary occipital horns, flax and interhemispheric fissure from, caudally, so formation of occipital lobes occurs.

Lobar variety:
Absence of the cavum septum pellucidum with fusion and squaring of the frontal horns. Midline cerebral structures may be normal.

4) Dandy Walker Complex 9
Dandy walker complex represents a group of developmental anomalies of posterior fossa that includes the classic Dandy – Walker malformation, Dandy Walker Variant and mega cistern magna.

Major components are-
1) Vermain agenesis or Hypoplasia
2) Posterior fossa cyst communicating with the fourth ventricle
3) Enlarged posterior fossa with displacement of the tentorium and lateral sinus.

Dandy Walker Variant partial/ complete absence of cerebellar vermis and small to normal sized but near –normal shaped cerebellar hemispheres. Sonographic continuity between the fourth ventricle and the cistern magna gave the appearance of the cleft.

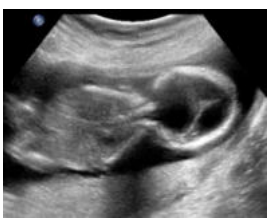

Fig 2 showing cystic dilatation of fourth ventricle with vermis hypoplasia

5) SPINA BIFIDA:
Evaluation of the fetal spine depends on visualization of the ossification centers within the fetal vertebrae. Each fetal vertebra has 3 ossification centre- one in the body and one at the base of

each transverse process. Three planes of imaging are commonly used coronal/parasagittal and transverse.

In longitudinal plan the spine has a 'rail –road track' appearance with gradual widening towards the fetal head and gradual tapering at the sacrum. Any disturbances of this relationship suggest a spinal abnormality.

6) ENCEPHALOCELE

- Herniation of intracranial structures through a defect in the cranium.
- They may contain only meninges and CSF (cranial meningocele) or brain tissue (encephalocele).

7) HYDRANCEPHALY :

Absence of a cerebral mantle of tissue and either and incomplete or absent falx.

The thalamus, basal ganglia and lower brain stem are intact.

Cerebrum is destroyed and replaced by fluid.

ANOMALIES OF GENITOURINARY SYSTEM

1) RENAL AGENESIS

ON USG

- Failure to demonstrate the kidneys.
- Failure to visualize the bladder on repeated scanning.
- In unilateral cases absence of 1 kidney with compensatory hypertrophy of the contra lateral kidney.

2) MULTICYSTIC DYPLASTIC KIDEY [10]

- Most common cause of congenital cystic renal dysplasia.
- Usually unilateral

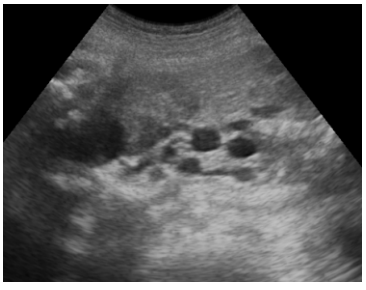

Fig 3 showing multiple non communicating cyst in rt kidney

On USG

The renal pelvis and ureter are usually atretic and not visible-occasionally, renal pelvis may be dilated

3) ECHOGENTIC KIDNEY:

There are wide differentials for echogenic kidneys.

Size	Differentials
Small	Obstructive dysplasia
Normal	Normal variant / Obstructive dysplasia
Enlarged	Autosomal recessive / dominant polycystic kidney disease
Enlarged with associated abnormalities	Trisomy 13, Meckel Gruber Syndrome, Perlmen Syndrome, Beckwith-Wiedmann Syndrome

4) PELVIURETERIC JUNCTION OBSTRUCTION

- Most common cause of fetal hydronephrosis.
- Unilateral in 70% of cases.
- When bilateral – asymmetric involvement
- On transverse scan anteroposterior diameter of renal pelvis should be measured.

<5mm = normal
5-10mm = normal – but require follow up.
>10mm = abnormal

Ratio between the maximum transverse pelvic diameter and the renal diameter if above 50% would suggest hydronephrosis.

5) POSTERIOR URETHERAL VALVES:

Affects male fetus.

Consisting of mucosal folds between the urethral wall and d i s t a l end of verumontanum.

Major sonographic signs :

- Dilation of fetal urinary bladder and proximal (prostate) urethra giving the classical Key Hole appearance.
- Ureteric dilation presenting as multiple serpigenous anechoic spaces.
- Hydronephrosis.

Conclusion

Ultra sound is born to obstetrics, for its simple noninvasive procedure, easy acceptability to all. The quantification of the true incidence and the definition of the systemic preponderance help in stratification of the affected fetuses for the sake of prognostication and tailoring of management policies.

REFERENCES

1) Anderson Nigel, Gosweel Owen, Duff Gerald. Prenatal Sonography For The Detection Of Fatel Anomalies : Results Of A Prospective Study & Comparison With Prior Series.
2) B Sarkar, S Bahaduria et al. antenatal detection of congenital anomalies. The Journal of Obst & Gynaec of India, 1991 : 41;37 – 40
3) Baaroncioni D, Scaglia C et al. Ultrasonography in pregnancy & fetal abnormalities : screening of diagnostic test Prenatal diagnostic test, 1995 : 15 (12) ; 1101 – 08
4) Chitty L S Ultrasound screening for fetal anomalies. Prenat Diagn, 1995:15(13); 1241-57
5) Hernadi L, Torocsik M et al. Examination of foetal developmental abnormalities
6) Cunnigham, William J, Walls. Ultrasound in evaluation of anecephaly, Radiology, January 1976:188:165; 167
7) Illokia LH, Azika ME, Ikourou Yoko 1, Bouramoueve. Anecephaly – cases seen in 3 years in Blazzavile. Journal of Obst. et. Biol Reprod (Paris, 1993 : 22(6); p621 – 624.
8) Hidalgo H, Bowie J, Rosenberg R Ram P, Ford K. Liipsit E. In utero sonographic diagnosis of fetal cerebral anomalies. AJR July 1982 : 139 : 143, 148
9) Judy, Estroff, Micheal Scott, Beryl – Bena Carraf. Dandy walker variant – prenatal sonographic features & clinical outcome.
10) Rosendhal H. Ultrasonography screening of fetal urinary tract malformations a prospective Eur J Obstetrics & Gynaecology Reprod Bio, 1990:36:37