Original Article

CONGENITAL ANOMALIES: PREVALENCE OF CONGENITAL ABNORMALITIES IN 2ND TRIMESTER OF PREGNANCY IN MADINA TEACHING HOSPITAL, FAISALABAD ON GRAY SCALE ULTRASOUND

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ABSTRACT

Objectives:
To evaluate the antenatal prevalence of major congenital anomalies and malformation patterns in our hospital population

Study Design:
Cross-sectional observational.

Settings:
Radiology Department of Madina Teaching Hospital Faisalabad.

Duration:
12 months from January 2009, to December 2009

Sample size:
2890.

Material and Methods:
The patients, who under went regular obstetrics ultrasound from, were recruited. Data of Antenatal ultrasounds was statistically analyzed on structured data collection form to determine the prevalence of congenital anomaly in 2nd trimester.

Results:
We diagnosed 86 cases of fetal anomalies. The antenatal prevalence of congenital anomalies was 29.75 per 1000 and 2.97%. The mean maternal age and mean gestational age at diagnosis was 26.5 years and 24 weeks respectively. Out of the total (N=86), 15.6% occurred in women above the age of 35 years. Central nervous system and Musculo-skeloton were commonly diagnosed, fallowed genitourinary, renal and miscellaneous (hydrops fetalis, pleural effusion) etc. However, facial and heart defects rarely found or more commonly missed.

Conclusion:
The prevalence of major congenital anomalies in our population appears to be similar to international figures AS 2.97%. The study showed the preponderance of neural tube defects. In contrast about a quarter of musculo-skeleton and genitourinary, however no facial defects and cardiac defect were detected because of high patient turn out, have no 3D/4D and Doppler ultrasound. Based on these findings we recommend that the obstetricians should advised, regular ultrasound at least at 18-23 weeks by consultant radiologist at low risk patients.

Keywords: Congenital anomaly, 2nd trimester ultrasound

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INTRODUCTION

A congenital anomaly is an abnormality of structure, function or body metabolism that is present at birth and results in physical or mental disability, or is fatal. Each year, eight million children are born worldwide with congenital anomalies, of which 3.3 million die

before the age of five; 3.2 million of the survivors may be mentally and/or physically disabled.[1] The prevalence of birth defects is comparable all over the world; about 3% in the United States, [2] 2.5% in India, [3] and 2% to 3% in the United Kingdom. [4] the most prevalent conditions include congenital heart defects, [5] orofacial clefts, Down syndrome, [6] and neural tube defects. [7]. There are a number of laboratory and imaging studies available for detection of these anomalies. Out of these ultrasound is the one which gives a great amount of information about the structure and to some extent physiological aspects of the state of fetus. Some anomalies like anencephaly can be picked as early as 12 weeks when skull ossification is complete.[8] The overall detection time varied from early to late pregnancy depending upon when the patient reports to hospital for antenatal checkup.

Second trimester ultrasound scan has become an essential part of antenatal care. In cases where a major structural defect is identified, termination of pregnancy is offered.[9] The morbidity and mortality of this procedure increases with advancing gestation. Therefore early detection of such abnormalities will result in the reduction of such complications.

The diagnostic ability of ultrasound is well established by a number of studies.[10-11] Detection of fetal abnormalities depends on a number of factors including the nature or type of abnormality, sophistication of equipment and experience of operator. The Prevalence of abnormalities also depends upon the population being scanned. In Pakistan where the social support system is virtually non-existent, bringing up a child with mental or physical handicap is a major burden for the parents and family. Primary prevention with Folic acid for this purpose has a limited role. In cases where primary prevention does not seem possible, prenatal diagnosis by ultrasound scan provides the next best alternative. The purpose of this study was to describe the prevalence of congenital abnormalities seen in low risk population in periphery of Madina Teaching Hospital, Faisalabad, Pakistan.

MATERIAL AND METHODS

This is a cross sectional observational study conducted in the department of Diagnostic Radiology at the MTH & UMDC, Faisalabad and Pakistan. The Madina Teaching Hospital is a tertiary care teaching hospital in the private sector equipped with the latest diagnostic and therapeutic facilities. About 10000-12000 routine ultrasounds take place every year out of which 3000-3200 are obstetrics. In the department of radiology, two antenatal ultrasound in pregnancy are performed, one at 11-14 weeks and the other between 18-22 weeks. A third trimester ultrasound is requested when indicated. We performed about 2890 ultrasounds on low risk pregnant women in 2nd trimester between January–2009 December 2009. Consultant Radiologist performed all the Transabdominal ultrasounds after obtaining a verbal consent, on a Toshiba Femio Machine, using 3.75 MHz probe. After enquired about any history of drug intake, exposure to any viral infections and history of any generalized disease like Diabetes mellitus or hypertension and demographic detail of study subjects was noted. In addition, types of birth defect, sex and age of mother was also noted. All the above-mentioned variables along with the detail anatomical survey at time of scan (Table 1) and demographic variables including gestational age were entered in a database file and analyzed by SPSS version 10.

Table 1. Anatomical survey at the time of scan.

Head and brain (lateral ventricle, septum pellucidum and cerebellum etc)
Heart, four chamber view and its position
Stomach bubble and its position
Umbilical cord insertion and anterior abdominal wall
Extremities including the position of hands and feet and number of digits
Spine
Bladder and kidneys.

RESULTS

During the study period from January 2009-December 2009, a total of 2890 prenatal ultrasounds were reported. Out of these 86 cases of congenital abnormalities were identified and they served as the study population. Congenital abnormalities occurred among 2.97% of all low risk population. The mean age and gestational age of the women in this study was 26.5 years (SD ± 5.3) and 24 weeks (SD ± 6.8) Among the study subjects 15.6% were women above the age of 35 years. Out of these 86 had different anomalies majority were from central nervous system, followed by Musculo-skeleton, miscellaneous (like cystic hygromas, IUGR's,

hydrops-fetalis, isolated pleural effusions and ascites), genitourinary & renal, and gastrointestinal. We had 47 cases of neural tube defects, 16 of musculo-skeletal, 08 of genitourinary, 12 of miscellaneous, 03 of gastro-intestinal, none from cardiovascular system and facial defects. We had 4 cases from central nervous system which showed multiple anomalies. Polyhydramnios was seen in 57% cases of neural tube defects and 61% cases of musculoskeletal anomalies. Oligohydramnios was noted in cases of agenesis of kidneys and polycystic kidney disease. We had 20 cases with twins and out of these one had one fetus affected, other normal.

The Spectrum of Abnormalities is shown in Table 2.

Total anomalies: 86 (2.97%)

Systems	Anomalies	Number (%)
Central Nervous system: 47(54.65%)	**a.** Hydrocephalus	17(36.10)
	b. Anencephaly	08(17.02)
	c. Encephaloceles	04(08.50)
	d. Microcephaly	02(04.25)
	e. Meningomyoceles	05(10.63)
	f. Dandy-Walker	02(04.25)
	g. Spina Bifida	05(10.63)
	h. Agenesis of corpus collasum	02(04.25)
	g. Arnald chiari syndrome	02(04.25)
	i. Sacrococcygeal teratoma	03(06.38)
Genitourinary: 8(9.30%)	**a.** Polycystic Kidneys	03(37.50)
	b. Posterior urethral valve	04(50.00)
	c. Pelvic cystic mass	01(12.50)
Miscellaneous: 12(13.95)	**a.** Hydrops Fetalis	07(58.33)
	b. Cystic Hygromas	02(16.66)
	c. Down syndrome	02(16.66)
	d. Plural Effusion	01(08.30)
Gastrointestinal: 3(6.38%)	**a**. Gastro schisis	01(33.33)
	b. Bud chiari syndrome	01(33.33)
	c. Duodenal atresia	01(33.33)
Musculoskeletal: 16(38.60%)	**a**. Skeleton dysplasia	11(68.75)
	b. Thanotophoric dwarfism	01(06.25)
	c. Osteogenesis Imperfecta	04(25.00)
	d. Thanatophoric dwarfism	01(06.25)

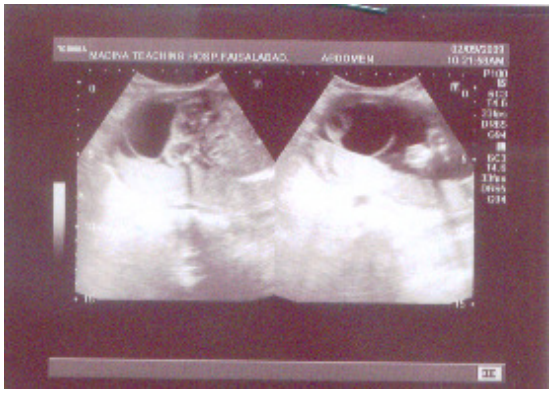
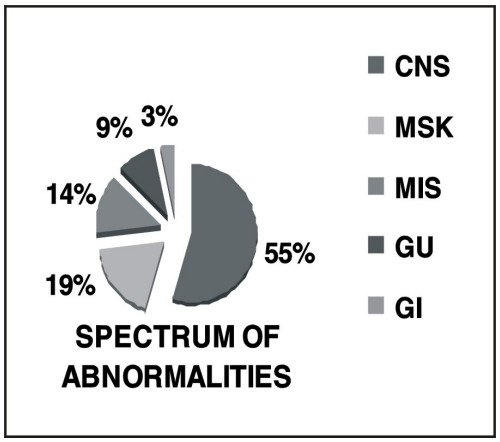


Fig. 1. Ultrasound. 26 year old woman at 16 3/7 weeks gestational age underwent sonographic examination. Sonographic imaging (using 3.75 MHz transducer) demonstrates a cystic area around the neck-cystic hygroma.



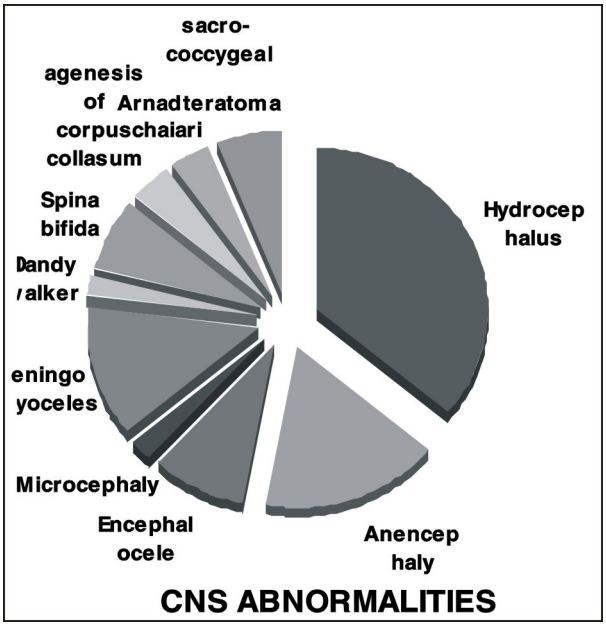


Fig. 2. Ultrasound. 26 year old woman at 28 2/7 weeks gestational age underwent sonographic examination. Sonographic imaging (using 3.75 MHz transducer) demonstrates shoetening of long bones especially proximal, serial monitoring USG scan reveals Skeleton Dysplasia.

DISCUSSION

Congenital malformations affect approximately 2-3% of all live births every year.[18] Ultrasound antenatal detection of congenital anomalies has become a new goal of obstetric management.

A congenital anomaly consists of a departure from normal anatomic architecture of an organ or system. Anomalies may result from an intrinsically abnormal promordium or

anlage of an organ or from a normal promordium that is affected during development by extrinsic forces.

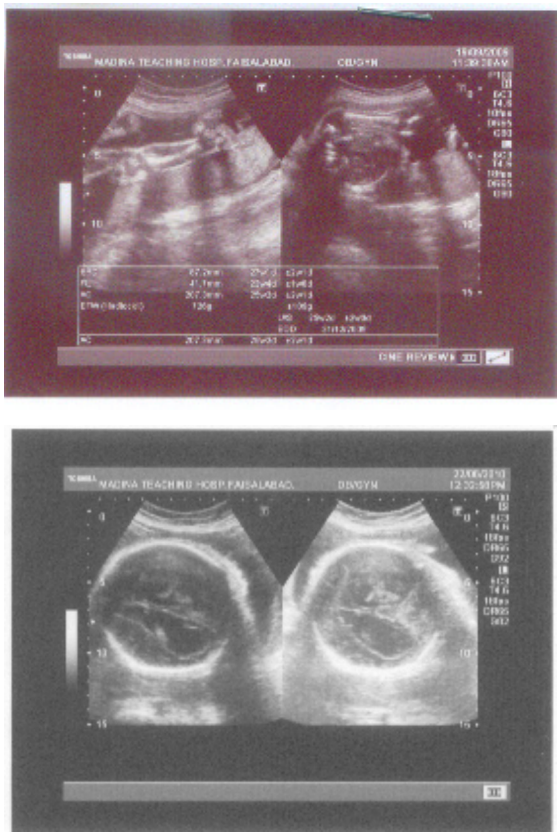


Fig. 3. Ultrasound. 26 year old woman at 16 3/7 weeks gestational age underwent sonographic examination. Sonographic imaging (using 3.75 MHz transducer) demonstrates dilated lateral ventricle, 3rd ventricle also dilated not visualized in this image-hydrocephalus.

Different anomalies may be classified as malformations, deformations and disruptions. Co-existent group of anomalies is described as polytopic field defect, sequence, syndrome and association.

Other classification may be major and minor anomalies. Major anomaly is one with a medical, surgical or cosmetic importance and with impact on morbidity and mortality. Minor anomaly is one that does not have a serious surgical, medical or cosmetic significance and does not affect normal life expectancy or lifestyle.[19]

The results we concluded in Madina Teaching Hospital show the preponderant presence of central nervous system anomalies which has been the case in a local study[20] as well as internationally.[21,31,32] Spectrum of the anomalies is shown in Table 2.

Routine antenatal ultrasound screening as compared to selective (high risk) has been found economically justifiable also.[22]. It also helps us for careful antenatal surveillance and judicious timing of delivery.[23, 24] This all has increased the responsibilities of doctors from just delivering the baby to a state where he or she has to cater from diagnosis to timing of delivery to future planning of pregnancies.[25] Some centers have reported better pickup rate around 11-14 weeks of pregnancy and recommended a second trimester anomaly scan in routine antenatal care to increase the prenatal detection of fetal defects.[26] Others have suggested sonogram at 16-18 weeks followed by serial scans to exclude or confirm an anomaly.[27] In spite of all efforts pickup rate for cardiac anomalies remains poor.[28] There is significant variation in pickup rates of anomalies in different regions of world including Europe depending upon operator's experience, equipment and different policies for scanning.[29] The current study evaluates various aspects of ultrasound screening at a teaching hospital of UMDC in Faisalabad.

The role of ultrasound in the detection of fetal anomalies is dependent on the prevalence of anomalies in a study population, the expertise of the examiner, the gestational age at scanning, the definition of anomaly-major and minor, and the postnatal ascertainment of anomalies. The skill and experience of the sonographers is a critical factor in the detection of fetal anomalies.[13] The ultrasound scan failed to detect any facial defects in our study. Low prediction rate of 17.5% has been reported by some of the earlier studies.[30] Similarly none of cardiac defects were diagnosed on the scan. The low detection rate was because the four chamber view was only included in the scan in their study population, and no targeted imaging for fetal anomalies (TIFFA), facility of 3D/4D and Doppler scan. Based on our results we conclude that the antenatal ultrasound scan at 18-23 weeks can be beneficial in detention of early anomalies, helps a great deal in avoiding un-necessary state expenditure, mental agony and trauma to family of carrying a handi-capped child.

CONCLUSION

Antenatal ultrasound is a non–invasive highly sensitive, accurate and cost effective imaging technique which gives good results in experienced hands. Meticulous screening for pregnant ladies by ultrasound, especially in 2nd trimester and follows up of anomaly cases if required.

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