Short Communication

Frequency of Different Structural Congenital Anomalies by Antenatal Ultrasound at Gestational Age of 11-24 Weeks

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Abstract

Objective: To determine the frequency of different structural congenital anomalies by antenatal ultrasound at gestation age of 11-24 weeks.
Study Design: Cross-sectional study
Place and Duration: Patients of Obstetrics and Gynaecology Department of Pakistan Atomic Energy Commission (PAEC) Hospital, Islamabad.
Methodology: 621 patients between gestational amenorrhea of 11-24 weeks attending the obstetric outdoor of PAEC hospital Islamabad who matched the selection criteria were included as cases. These were the patients who came through the outpatient department of PAEC General Hospital, Islamabad. Ultrasound was advised to every patient and done by a consultant radiologist. These females were then followed to see for the frequency of congenital anomalies at mentioned gestation.
Results: During the study period, a total of 621 patients between gestational amenorrhea of 11-24 weeks were selected. Patients mean age was 27.10 ± 5.13. Anomalies were found in 17 patients (2.7%). Hydrocephalus was found in 8 (1.28%), Anencephaly in 7 (1.12%) patients and spina bifida was found in 2 patients (0.32%).
Conclusion: Antenatal ultrasonography is a non–invasive highly sensitive, accurate and cost-effective imaging technique which gives good results. Screening of pregnant ladies by ultrasound, especially in 2nd trimester helps in avoiding mental agony and trauma to the family of carrying a handicapped child.
Keywords: Ultrasonography (USG), congenital anomalies, antenatal diagnosis, anencephaly, hydrocephalus, spina bifida.

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Introduction

Congenital anomaly is an unusual happening at birth that can occur because of genetic abnormalities, unfavorable intrauterine environment, infections and morphogenesis errors. Examples of such anomalies are cleft lip or palate or any other birth defect with serious medical or cosmetic importance.[1] Congenital anomalies affect 2-3% of all live births every year.[2] The average frequency of congenital malformations is 11.8 per 1000 births.[3]

Congenital anomalies can be divided into major and minor defects. Major defects can be lethal and may need medical and surgical interventions; therefore, early detection is essential and life saving[4].

It is reported that congenital malformations occur in 3% of all infants[5]. Congenital anomalies are second cause of infant morbidity after prematurity.

Antenatal ultrasound is very important in detecting structural anomalies. The sensitivity of prenatal

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ultrasound in the diagnosis of foetal anomalies is 96% and specificity is 79%.[6] Major foetal structural anomalies can be picked up by ultrasound at 11-14 weeks but detailed fetal anomaly scan should be done at 18-22 weeks. Cranial abnormalities are most common which can be detected in the first trimester[7]. Anencephaly is most commonly reported i.e. 44.9%[8]. Others are omphalocele (16.67%), encephalocele (15.58%), hydrops fetalis (4.3%) [10], spina bifida (2.59%), cystic hygroma and gastroschisis (0.073%)[9]. First-trimester ultrasound helps in early detection of a large number of foetal malformations.[11] Up to 70% of major anomalies can be diagnosed at 14 weeks of gestation and prognosis of such foetus is very poor[12]. Anomalies which have been detected during the second trimester by ultrasound are hydrocephalous (36.16%)[2], cardiac defects, polycystic kidney[7]. Detection rate is 95% in the second trimester.

Major congenital anomalies are horrific for the patients to know about. The gravity of the situation is enhanced when it is associated with other conditions like a patient having a major congenital anomalous baby at term with previous 2 or 3 caesarean sections when surgery becomes mandatory instead of termination of pregnancy.

The rationale of study is that major congenital anomalies are dreadful for patients to know about but early diagnosis helps in the decision of termination of pregnancy as it is based on religious beliefs and future planning for avoiding such deformed babies. Early detection and termination of major congenital anomalies will help parents to avoid emotional stress they can face during upbringing of mentally or physically handicapped child. So routine ultrasound is economical and helps in early diagnosis of congenital anomalies, thereby avoiding unnecessary state expenditure, mental stress and trauma to the family of having such handicap child.

Methodology

This analytic study was conducted for a period of six months i.e., from 1 Oct 2013 to 31 Mar 2014 at Pakistan Atomic Energy Commission Hospital, Islamabad, Pakistan. By keeping the confidence level 1-α as 95%, the absolute precision required (d) as 1.25%; anticipated population proportion (P) is 2.59%[2] (prevalence of common anomaly i.e. anencephaly was 28.5%) and putting all these values using software, calculated sample size is 621. So, 621 antenatal patients of the general population were included in the study. Non-probability consecutive sampling technique was applied. Our study included all antenatal women presented at gestational age 11-24 weeks assessed on LMP and all those excluded were women who presented after 24 weeks of gestation. All patients fulfilling inclusion criteria were recruited. Exclusion criteria were strictly followed to limit the confounding factor. Informed consent for termination of pregnancy was taken in patients with major foetal congenital anomalies. Fetal morbidity and mortality was explained to the patients if pregnancy prolongs. Every patient was advised antenatal ultrasound between 11-24 weeks of gestation. Ultrasound was done by consultant sonologist of our hospital. Reports of ultrasound were collected and recorded over pro forma designed for this purpose (which is attached at the end).

Data Collection and Statistical Analysis: The data was analysed using SPSS version 10 for windows. A descriptive analysis of quantitative variables was calculated. Variables (Age, parity and gestational age) were measured by mean and standard deviation. Frequency and percentages were calculated for all qualitative variables like different congenital anomalies (i.e. hydrocephalous, anencephaly, spina bifida). All the data was presented in the form of tables.

Effect modifiers like age, parity, previous history of congenital abnormality will be controlled by making cross matched stratified tables through Chi – square taking $P < 0.05$ as significant.

Results

During the study period, a total of 621 antenatal patients were taken and 17 patients were having structural congenital anomalies.

The subjects included in the study were patients who came through the outpatient department of PAEC Hospital, Islamabad. The patients were of 16-44 years of age and between gestational amenorrhea of 11-24 weeks. The mean gestational age was 18.04 ± 3.48. In the sample, Primigravida and multigravida all were included. The Mean age was found to be

27.10 ± 5.13 and mean parity was 1.68 ± 1.60, as shown in Table I.

Table I: Descriptive Analysis Of Patient Age, Gestational Age And Parity

	N	Minimum	Maximum	Mean	Std deviation
Age of patient	621	16	44	27.10	5.13
Gestational age	621	11	24	18.04	3.48
Parity	621	0	6	1.68	1.60

Fetal congenital anomalies were found in 17 patients, so prevalence was 2.7% as shown in Table II. Out of 17 patients, 8 were hydrocephalic fetus. The prevalence of hydrocephaly in our study is 1.28% as shown in Table III. So hydrocephaly is most common anomaly found in the study group.

Table II: Distribution of Congenital Anomalies

Congenital anomalies	Frequency	Percent
Yes	17	2.7
No	604	97.3
Total	621	100

Table III: Distribution of hydrocephalus

Hydrocephalus	Frequency	Percent
Yes	8	1.28
No	613	98.72
Total	621	100

The second commonest anomaly was anencephaly in our patients and this was found in 7 patients out of 621. So it is 1.12%. This is shown in Table IV. Spina bifida is least common in our study and this anomaly was found in only 2 patients so, the prevalence is 0.32%. This is shown in Table V.

Table IV: Distribution of Anencephaly

Anencephaly	Frequency	Percent
Yes	7	1.12
No	614	98.88
Total	621	100

Table V: Distribution Of Spina Bifida

Spina bifida	Frequency	Percent
Yes	2	0.32
No	619	99.68
Total	621	100

Discussion

In modern obstetric, the role of USG is very much enhanced and is being continuously evolving since its introduction. Modern obstetric practice without real time USG is difficult to imagine. In pregnancy and its complications, the role of USG is pronounced. Using this imaging technique, congenital anomalies can be detected in early gestation. Clinicians can detect pregnancy as early as 3 weeks after conception, confirm or revise gestational age with reasonable accuracy, diagnose multiple gestation in early pregnancy, confidently diagnose foetal death at any gestational age, assess foetal well-being, evaluate amniotic fluid volume, and diagnose a broad variety of foetal malformations to be used as a screening test and has irreplaceable role. Ultrasound has a relatively low cost and high sensitivity and specificity rates and is widely available. Use of ultrasound is much more as a diagnostic modality, rather than a therapeutic one. Ultrasound alone cannot reduce perinatal mortality, but the information obtained from an ultrasound can be used to guide the clinician to choose the appropriate therapy. Direct cost savings from screening ultrasound examinations may be closely related to the accuracy of these examinations in detecting foetal anomalies[20]. It will help in timely interventions, counseling of the couple regarding the outcome, whether it is surgically correctable, lethal or will result in handicapped child.

As ultrasound is very important in diagnosing congenital anomalies, dating scan in first trimester and anomaly scan in second trimester is recommended in every pregnancy. In total, 621 patients were included in the study at gestational amenorrhea of 11 - 24 weeks during 1 Oct 13 to 31 Mar 14. Patients were between 16 - 44 years of age with different parity. Mean age of patients and mean gestational age was 27.10 ± 5.13 and 18.04 ± 3.48 respectively. This is shown in Table I.

Worldwide surveys have shown that the birth prevalence of congenital anomalies varies greatly from country to country i.e. from 1.07% in Japan as high as 4.3% in Taiwan.[13] In this study, the prevalence of congenital anomalies is 2.7%, and a similar study conducted in Agha Khan Hospital

Karachi by Munim *et al.*[21] it is 2.8%, which is consistent with our study. The prevalence of hydrocephalus is 1.28% in our study as shown in Table III.

In a study conducted at Saudi Arabia by Murshid *et al.*[22] the prevalence of hydrocephalus is 1.6/1000 births, i.e. 0.16%. The risk factors for congenital hydrocephalus include the absence of antenatal care, maternal hypertension during pregnancy, Infections, Preeclampsia and hereditary component.[14] Most of these factors were interplaying in our patients, which is a reason for high prevalence.

The other anomalies were neural tube defects which include anencephaly and spina bifida. The second most common anomaly found was anencephaly, which is a lethal birth defect, therefore, termination of pregnancy at any gestation is accepted.[15, 17] Its incidence is 1.12%, as is shown in Table 4, whereas the incidence of anencephaly in Ireland, Scotland and Wales is 6-8/1000 births which is equivalent to 0.6-0.8% and incidence of neural tube defects in the United States is 0.1%.[16]

The purpose of study was not to find a correlation between congenital anomalies and maternal age however; most cases of anencephaly in our study were diagnosed in younger patients (n = 5). It appeared to be decreased with increasing maternal age, which correlates with age-specific prevalence of NTDs in UK.[19]

Results of other studies show that in the developed countries incidence of NTDs is very low. This is due to preconceptional counseling, antenatal screening and selective termination of affected pregnancies at earlier stage of gestation[19]. Their women in reproductive age group or planning for pregnancy, take prophylactic dose of folic acid (0.4mg daily) and those having previous abnormal outcome take therapeutic dose of folic acid (4mg daily) as recommended by U.S public health services. The known risk factors are detected and dealt appropriately during preconceptional counseling and prenatal care. Reasons for higher prevalence are lack of: preconceptional counseling, antenatal care, intake of folic acid and early diagnosis and termination of affected pregnancies.

Other neural tube defect was spina bifida. It is a defect of the central nervous system and patients born with major defects have lifelong disabilities including motor, sensory, orthopaedic and urologic problems. Spina bifida was found in 0.32% of cases as shown in Table V which is higher as compared to other studies from Iran, United States, Europe, and Japan.

In a study conducted in Children Hospital Iran, it is 1.6/1000 births i.e. 0.16%. In the United States 0.05% and in Europe and Japan it is 0.1%. These are very low as compared to our study. The incidence of spina bifida varies according to country, culture, and socioeconomic status. Because of its multifactorial aetiology its prevalence varies considerably in different parts of world.[18]

In a hospital-based prospective study, the overall incidence of congenital anomalies was 2.5% (63 of 2517) of live-born neonates. These findings are in accordance with the results of the European network of population-based registers for the epidemiological surveillance of congenital anomalies (EUROCAT) (2.4%)[23], Jones (2.3%)[24], and Jehangir *et al*.[25] in a tertiary care hospital, who reported an incidence of 2.9%. A lower incidence of congenital anomalies was reported in Kuwait by Madi *et al.*[26] who reported an incidence of 1.25%, in India by Swain *et al*.[27] who reported an incidence of 1.2%, and in the United Arab Emirates[28]. Furthermore, a lower incidence of congenital anomalies was reported in Egypt among live births and stillbirths: 1.16% in Alexandria and 1.58% in Cairo.[29]

Conclusion

Congenital anomalies can be decreased by preconceptional counseling, intake of folic acid supplements, good glycaemic control, avoidance of teratogenic/antiepileptic drugs. Early detection by anomaly scan and early termination can decrease the incidence of lethal congenital anomalies. Routine anomaly scan is done at 18-22 weeks but an anomaly scan performed at 11-13 weeks of gestation can detect gross congenital anomalies like Anencephaly at early gestation. So early detection and early termination can be offered which will reduce the physical/mental trauma to the patient.

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