Original Article

Prevalence of Congenital Anomalies Detected by the Second Trimester Ultrasound Scan in a Tertiary Care Hospital in Bangladesh

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Abstract:
Objectives: *The aim of the study is to determine the prevalence of congenital anomalies detected by the second trimester ultrasound examination.* ***Materials & Methods:*** *This is a cross sectional observational study. Total 6610 pregnant women, who underwent regular obstetrics ultrasound were recruited from January 2015 to December 2016. Data of antenatal ultrasounds were collected and analyzed statistically to determine the proportion and patterns of congenital anomalies in the second trimester of pregnancy.* ***Results:*** *154 cases of congenital anomalies were diagnosed. The prevalence of congenital anomalies was 23.29 per 1000 pregnancy. The mean maternal age was 27.04 years and mean gestational age at diagnosis was 22.3 weeks. About 15% of anomalies found in women above the age of 35 years. Central nervous system anomalies were found more common, followed by renal, musculoskeletal, gastrointestinal and respiratory. Cardiac defects and minor limb defects like polydactyly or syndactyly rarely found or more commonly missed.* ***Conclusion:*** *Ultrasound is a readily available, non-invasive, highly sensitive, cost-effective & safe means of determining gestational age, evaluating fetal health and anomalies. The second trimester targeted ultrasound scan at 18-22 weeks can be beneficial in early detection of anomalies and thus avoiding mental stress & trauma to parents & family of carrying a handicapped child. Antenatal ultrasound is an important screening test, yielding results that must be interpreted and integrated in a knowledgeable way.*

Key words: Congenital anomalies, Second trimester ultrasound, Targeted scan
Received: April 11, 2017; **Accepted:** April 28, 2017

Introduction:
Congenital anomalies are the structural, functional and metabolic disorders occur during intrauterine life and can be identified prenatally, at birth or sometimes may only be detected later in infancy.

The anomalies are the leading cause of neonatal morbidity & mortality. Defective embryogenesis or intrinsic abnormalities in the process of development result congenital anomalies. About 3,03,000 newborns die within 4 weeks of birth every year worldwide due to congenital anomalies [1].

Other than death, these anomalies can contribute chronic disability which may have severe impacts on individual, family, societies and overall healthcare system. The prevalence of birth defects is comparable all over the world; about 3% in the United States, 2.5% in India, 2.97% in Pakistan and 2% to 3% in the United Kingdom [2-5]. Though birth defects are global problem but their impacts are more severe in middle- and low-income countries than the high-income countries.

Congenital anomalies may present as isolated abnormalities or part of a syndrome. The most common, severe anomalies include congenital heart defects, cleft lip with or without cleft palate, Down syndrome & neural tube defects [6-8]. Laboratory test and imaging studies are available for detection of these anomalies. Ultrasound examination is one of the most important diagnostic tools which give a great amount of information about the anatomical structure as well as some physiological aspect of the status of the fetus. The current ACR (American College of Radiology)/ AIUM (American Institute of Ultrasound in Medicine) guidelines for the performance of the second & third trimester obstetrics examination describe the standard sonographic examination. The guidelines help to maximize detection of many fetal abnormalities. The level-I (Standard or routine) ultrasound scan is performed routinely on pregnant woman. This examination scans maternal uterus, ovaries, cervix and placenta, as well as a systematic review of fetal anatomy.

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In high risk cases, targeted or level-II scan is indicated for a detailed fetal sonogram. The level-II scan is specifically indicated when an anomaly is suspected because of maternal medical or family history, or any abnormality of the fetus is suspected on a routine scan. In general, the standard fetal anatomic survey refers to the second trimester scan performed between 16 and 22 weeks of gestation. When anatomic surveys are performed at 20 & 22 weeks gestational age, there is less need for repeat scan [9]. If a major structural defect is identified, termination of pregnancy is offered, as morbidity and mortality of this procedure increases with advancing gestation [10].

The diagnostic ability of ultrasound is well established by a number of studies [11,12]. Detection of fetal abnormalities depends on a number of factors including nature or type of the abnormality, sophistication of the equipment & experience of the operator. The prevalence of anomalies also depends upon the population being scanned. In Bangladesh 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 & family. The purpose of this study was to determine the prevalence of congenital anomalies detected by the second trimester ultrasound scan in our population.

Materials and Methods:
This is a cross sectional observational study, the study was conducted in the Radiology & Imaging Department of BIRDEM (Bangladesh Institute of Research and Rehabilitation in Diabetes, Endocrine and Metabolic Disorders) General Hospital-2 (Mohila & Shishu), Dhaka, Bangladesh. The BIRDEM General Hospital-2 is a tertiary care teaching hospital. About 8,000-10,000 routine ultrasounds take place every year out of which 3,000-3,600 are obstetrics.

In the Department of Radiology & Imaging at least two antenatal ultrasounds in each pregnancy are performed, one in first trimester of pregnancy at 10-12 weeks or earlier and another between 18-22 weeks. Third trimester ultrasound is requested when indicated. We performed about 6,610 ultrasounds in the 2nd trimester from January 2015 to December 2016. Trans-abdominal ultrasounds were performed on a HITACHI EUB7000HV Machine, using 3.75 MHz probe. Demographic detail, generalized medical history including drug intake, exposure to radiation or any viral infection, generalized disease like Diabetes Mellitus or Hypertension of study subjects were noted.

All the above mentioned variables including gestational age were entered in a database file & analyzed by SPSS version 12. The fetal anatomic survey shown in Table I was done for detection of anomalies.

Table-I: Fetal anatomic survey

Region	Structures examined
Head, Face & Neck	Lateral ventricles, midline falx, cavum septi pellucidi, cisterna magna, cerebellum
Chest	Four chamber view of the heart & outflow tracts Lungs Diaphragm
Abdomen	Stomach, kidneys, urinary bladder Umbilical cord-site of insertion, vessel number Anterior abdominal wall
Spine	Cervical, thoracic, lumbar & sacral
Extremities	Upper limbs Lower limbs

Results:
During the study period from January 2015 to December 2016, a total of 6,610 prenatal ultrasound were done. Out of these, 154 cases of congenital abnormalities were identified. Congenital abnormalities occurred among 2.32% of study population. The prevalence of congenital anomalies is 23.29 per 1000 pregnancy. The mean age of the women was 27.04 years and mean gestational age was 22.32 weeks. Among the study subject 15% were women above the age of 35 years.

Out of these 154 had different anomalies, majority from the central nervous system, followed by renal, musculoskeletal, gastrointestinal, respiratory and miscellaneous (like fetal hydrops, ascites, pleural effusion and cystic hygroma) etc. Polyhydramnios was found in 66% cases of neural tube defect, oligohydramnios was noted in cases of agenesis of kidneys, multicystic kidneys & bilateral PUJ (pelvi ureteric junction) obstruction cases.

We found 10 cases with multiple anomalies. Among 45 cases of twin pregnancy, 3 had stalked twin and 1 had anomalous monster fetus with twin-twin transfusion syndrome.

Table-II: Congenital anomalies involving the CNS (n=60)

CNS congenital anomalies	Number (%)
Anencephaly	25 (41.67%)
Hydrocephalus	12 (20.00%)
Encephalocele/ Meningoencephalocele	12 (20.00%)
Holoprosencephaly	8 (13.33%)
Caudal regression syndrome	3 (5.00%)

Majority of the congenital anomalies involve the central nervous system are Anencephaly, followed by Hydrocephalus and Encephalocele or Meningoencephalocele. Holoprosencephaly and caudal regression syndrome were also found. The distribution of different type of anomalies of the central nervous system (CNS) is shown in Table II.

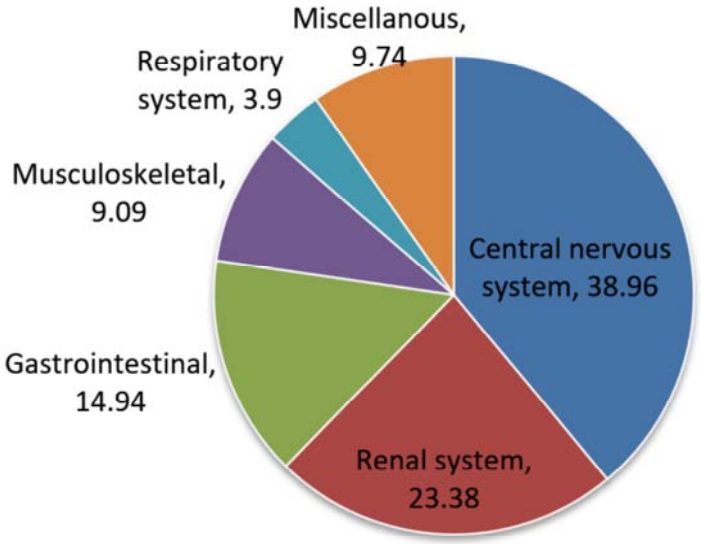

Figure-1: Percentage of congenital anomalies that involve different systems

Hydronephrosis was the most frequently found renal anomalies. Other anomalies involving the renal system were renal agenesis, multicystic or polycystic kidneys and posterior urethral valve. Table III shows the congenital anomalies involving the renal system.

Table-III: Congenital anomalies involving the renal system (n=36)

Renal system anomalies	Number (%)
Hydronephrosis (unilateral /bilateral)	22 (61.12%)
Renal agenesis	6 (16.67%)
Multicystic / polycystic kidneys	6 (16.67%)
Posterior urethral valve	2 (5.56%)

Among the musculoskeletal anomalies, we found achondroplasia, club foot, facial cleft, limb defect and osteogenesis imperfect. The distribution is shown in Table IV.

Table-IV: Congenital anomalies involving the musculoskeletal system (n=23)

Musculoskeletal anomalies	Number (%)
Achondroplasia	6 (26.09%)
Club foot	6 (26.09%)
Facial cleft	5 (21.39%)
Limb defect	4 (17.39%)
Osteogenesis imperfecta	2 (8.69%)

Hirschsprung's disease, anal canal malformation, duodenal atresia, omphalocele and gastroschisis were the gastrointestinal malformation. Among the respiratory anomalies, we found unilateral or bilateral enlarged echogenic lung, bronchial cyst, hypoplastic lung. Other than these anomalies, congenital diaphragmatic hernia, eventration, cystic hygroma were also found in our hospital. The distribution of these anomalies is shown in Table V.

Table-V: Congenital anomalies involving the gastrointestinal system, respiratory system and miscellaneous

Congenital anomalies	Number (%)
Gastrointestinal anomalies	**N=14**
Hirschsprung's disease/ anal canal malformation	4 (28.57%)
Duodenal atresia	4 (28.57%)
Omphalocele	3 (21.42%)
Gastroschisis	3 (21.42%)
Respiratory anomalies	**N=6**
Enlarged echogenic lung (unilateral/ bilateral)	2 (33.34%)
Bronchial cyst	2 (33.34%)
Hypoplastic lung	2 (33.34%)
Miscellaneous anomalies	**N=15**
Stalked twin	3 (20.00%)
Congenital diaphragmatic hernia	2 (13.34%)
Eventration	2 (13.34%)
Cystic hygroma	1 (6.67)
Vascular malformation (CNS)	1 (6.67)
Pelvic cyst	2 (13.34%)
Anomalous monster baby with twin-twin transfusion syndrome	1 (6.67%)
Ascites	1 (6.67%)
Pleural effusion	1 (6.67%)
Fetal hydrops	1 (6.67%)

Discussion:

Congenital anomalies is the complex interaction of genetic and environmental factors like maternal age, consanguinity of parents, viral infection, drugs, exposure to chemical toxins and radiation, and socioeconomic status. Therefore, the prevalence and type of congenital anomalies may vary with time, geographical location, race and ethnicity [13]. The prevalence of congenital abnormalities in our hospital is about 2.32% or 23.29 per 1000 second trimester pregnancy. The rate is similar to that of the other countries like India and the UK [3,6]. A large European data is also similar to our statistics [14]. The mean maternal age of our study is about 27 years which is also comparable to India [13].

Congenital anomaly is the one of the leading causes of infant mortality and morbidity worldwide. Even in the developed countries, in the USA congenital anomalies are responsible for about 20% of infant death [15]. In our study, we found majority (about 39%) of anomalies involving the central nervous system, which is also high worldwide [17,18,19]. Renal system anomalies found 2nd common anomalies in our study, followed by gastrointestinal, musculoskeletal, respiratory and other anomalies.

In this study, none of the cardiac defects or minor musculoskeletal defect was detected. The low detection rate was because the four chamber view was only included in those scans & no targeted imaging for fetal anomalies, facility of 3D/4D and Doppler scan was also limited.

Routine antenatal ultrasound screening as compared to selective (high risk) has been found economically justifiable [16]. Prenatal detection of anomaly as early as possible is important for pregnancy management & planning. Early detection of these anomalies can reduce the maternal morbidity and mortality. Moreover, early detection of fetal anomalies can provide treatment of the fetus in utero, especially for cardiac anomalies. Once an anomaly is detected, various management options are to be discussed with the patient in consultation with neonatologist, pediatric surgeon & neurosurgeon when necessary. But if the congenital anomaly is incompatible with life then pregnancy should be terminated.

Prenatal ultrasound is an attractive option for screening test of congenital abnormalities. Ultrasound scanning is the safest, least expensive and non-invasive test for diagnosis of fetal anomalies [19]. Though some centers have reported better pick-up rate around 11-14 weeks of pregnancy, a second trimester targeted scan at 18-20 weeks is recommended for detection of anomalies [20]. Some centers practice targeted scan at 18-20 weeks and again at 24 weeks to exclude anomalies [21].

Conclusion:

Ultrasound is a readily available, non-invasive, highly sensitive, cost-effective & safe means of determining gestational age, evaluating fetal health and anomalies. The second trimester targeted ultrasound scan at 18-22 weeks can be beneficial in early detection of anomalies and thus avoiding mental stress & trauma to parents & family of carrying a handicapped child. Antenatal ultrasound is an important screening test, yielding results that must be interpreted and integrated in a knowledgeable way.

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Citation of this article:
Begum SA, Nishat L, Mannan A, Rahman T, Rahman Y. Prevalence of Congenital Anomalies Detected by the Second Trimester Ultrasound Scan in a Tertiary Care Hospital in Bangladesh. Eastern Med Coll J. 2017; 2 (2): 5-9.