

STUDY OF CONGENITAL FETAL ANOMALIES WITH THE HELP OF SONOGRAPHIC EVALUATION

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Article Info: Received 01 October 2019; Accepted 29 October 2019

DOI: <https://doi.org/10.32553/ijmbs.v3i10.685>

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Conflict of interest: No conflict of interest.

Abstract:

Information regarding specific anatomic anomalies affords the physician the opportunity to offer the patient sophisticated prenatal procedures, such as fetal surgery or selective fetal reduction in multiple gestations. Likewise, prenatal knowledge about genetic, physiologic, and/or anatomic abnormalities enables the physician to tailor or manage the timing and mode of delivery for optimal maternal and fetal outcomes. Prenatal diagnosis also allows the neonatal and paediatric specialists to be adequately prepared for a potentially ill neonate at delivery. Recent progress in the fields of maternal fetal medicine, radiology, and genetics has resulted in great advances in prenatal diagnosis. Ultrasonography is the initial modality for evaluation of pregnant patient because of its widespread availability and reasonable cost, with other modalities used only if Ultrasonography results are non diagnostic. Hence based on above data the present study was planned Study of Congenital Fetal anomalies with the Help of Sonographic Evaluation.

The present study was planned in Department of Radio- Diagnosis, Katihar Medical College and Hospital, Katihar, Bihar, India. All pregnant females of second trimester who are referred from obstetrics and gynaecology department and thus sent to department of Radio diagnosis for antenatal Sonographic examination. A complete antenatal ultrasound examination of pregnant women included in the study will be done using gray scale & colour duplex examination.

Obstetric ultrasonography has become an important part of routine antenatal care. Routine anomaly screening improves perinatal outcome directly through termination of pregnancy for certain anomalies. Congenital fetal anomalies are one of the most threatening complications which are prevalent in the society associated with severe morbidity and mortality in the new born fetus or neonates. Ultrasound is the best possible non-invasive technique available to detect any congenital anomalies in pregnant women which will help to identify the severity of the disease, its outcome leading to pregnancy termination or gives an opportunity for fetal therapy or better neonatal care.

Keywords: Congenital Fetal anomalies, Sonography, etc.

Introduction

A birth defect, also known as a congenital disorder, is a condition present at birth regardless of its cause. Birth defects may result in disabilities that may be physical, intellectual, or developmental. The disabilities can range from mild to severe. Birth defects are divided into two main types: structural disorders in which problems are seen with the shape of a body part and functional disorders in which problems exist with how a body part works.[4] Functional disorders include metabolic and degenerative disorders. Some birth defects include both structural and functional disorders.[1]

Birth defects may result from genetic or chromosomal disorders, exposure to certain medications or chemicals, or certain infections during pregnancy. Risk factors include folate deficiency, drinking alcohol or smoking during pregnancy, poorly controlled diabetes, and a mother over the age of 35 years old. Many are believed to involve multiple factors. Birth defects may be visible at birth or diagnosed by screening tests. A number of defects can be detected before birth by different prenatal tests.[2]

Treatment varies depending on the defect in question. This may include therapy, medication, surgery, or assistive technology. Birth defects affected about 96 million people as of 2015. In the

United States, they occur in about 3% of newborns. They resulted in about 628,000 deaths in 2015, down from 751,000 in 1990. The types with the greatest numbers of deaths are congenital heart disease (303,000), followed by neural tube defects (65,000).[3]

A congenital physical anomaly is an abnormality of the structure of a body part. It may or may not be perceived as a problem condition. Many, if not most, people have one or more minor physical anomalies if examined carefully. Examples of minor anomalies can include curvature of the fifth finger (clinodactyly), a third nipple, tiny indentations of the skin near the ears (preauricular pits), shortness of the fourth metacarpal or metatarsal bones, or dimples over the lower spine (sacral dimples). Some minor anomalies may be clues to more significant internal abnormalities.

Birth defect is a widely used term for a congenital malformation, i.e. a congenital, physical anomaly that is recognizable at birth, and which is significant enough to be considered a problem. According to the Centers for Disease Control and Prevention(CDC), most birth defects are believed to be caused by a complex mix of factors including genetics, environment, and behaviors, though many birth defects have no known cause. An example of a birth defect is cleft palate, which occurs during the fourth through seventh weeks of gestation. Body tissue and special cells from each side of the head grow toward the center of the face. They join together to make the face. A cleft means a split or separation; the "roof" of the mouth is called the palate.[4]

A congenital malformation is a physical anomaly that is deleterious, i.e. a structural defect perceived as a problem. A typical combination of malformations affecting more than one body part is referred to as a malformation syndrome. Some conditions are due to abnormal tissue development: A malformation is associated with a disorder of tissue development. Malformations often occur in the first trimester. A dysplasia is a disorder at the organ level that is due to problems with tissue development.[5]

Conditions also can arise after tissue is formed: A deformation is a condition arising from mechanical stress to normal tissue. Deformations often occur in the second or third trimester, and can be due to oligohydramnios. A disruption involves breakdown of normal tissues.[5] When multiple effects occur in a

specified order, they are known as a sequence. When the order is not known, it is a syndrome.

A limb anomaly is called a dysmelia. These include all forms of limbs anomalies, such as amelia, ectrodactyly, phocomelia, polymelia, polydactyly, syndactyly, polysyndactyly, oligodactyly, brachydactyly, achondroplasia, congenital aplasia or hypoplasia, amniotic band syndrome, and cleidocranial dysostosis. Congenital heart defects include patent ductus arteriosus, atrial septal defect, ventricular septal defect, and tetralogy of Fallot.

Congenital anomalies of the nervous system include neural tube defects such as spina bifida, encephalocele, and anencephaly. Other congenital anomalies of the nervous system include the Arnold-Chiari malformation, the Dandy-Walker malformation, hydrocephalus, microencephaly, megalencephaly, lissencephaly, polymicrogyria, holoprosencephaly, and agenesis of the corpus callosum. Congenital anomalies of the gastrointestinal system include numerous forms of stenosis and atresia, and perforation, such as gastroschisis. Congenital anomalies of the kidney and urinary tract include renal parenchyma, kidneys, and urinary collecting system.[6] Defects can be bilateral or unilateral, and different defects often coexist in an individual child.

The mother's consumption of alcohol during pregnancy can cause a continuum of various permanent birth defects: craniofacial abnormalities, brain damage, intellectual disability, heart disease, kidney abnormality, skeletal anomalies, ocular abnormalities. The prevalence of children affected is estimated at least 1% in U.S. as well in Canada. Very few studies have investigated the links between paternal alcohol use and offspring health. However, recent animal research has shown a correlation between paternal alcohol exposure and decreased offspring birth weight. Behavioral and cognitive disorders, including difficulties with learning and memory, hyperactivity, and lowered stress tolerance have been linked to paternal alcohol ingestion. The compromised stress management skills of animals whose male parent was exposed to alcohol are similar to the exaggerated responses to stress that children with fetal alcohol syndrome display because of maternal alcohol use. These birth defects and behavioural disorders were found in cases of both long- and short-term paternal alcohol ingestion. In the same animal study, paternal alcohol exposure

was correlated with a significant difference in organ size and the increased risk of the offspring displaying ventricular septal defects at birth.[7]

Substances whose toxicity can cause congenital disorders are called teratogens, and include certain pharmaceutical and recreational drugs in pregnancy, as well as many environmental toxins in pregnancy. A review published in 2010 identified six main teratogenic mechanisms associated with medication use: folate antagonism, neural crest cell disruption, endocrine disruption, oxidative stress, vascular disruption, and specific receptor- or enzyme-mediated teratogenesis.[8]

An estimated 10% of all birth defects are caused by prenatal exposure to a teratogenic agent. These exposures include medication or drug exposures, maternal infections and diseases, and environmental and occupational exposures. Paternal smoking use has also been linked to an increased risk of birth defects and childhood cancer for the offspring, where the paternal germline undergoes oxidative damage due to cigarette use. Teratogen-caused birth defects are potentially preventable. Nearly 50% of pregnant women have been exposed to at least one medication during gestation. During pregnancy, a woman can also be exposed to teratogens from the contaminated clothing or toxins within the seminal fluid of a partner. An additional study found that of 200 individuals referred for genetic counseling for a teratogenic exposure, 52% were exposed to more than one potential teratogen.[9]

A low socioeconomic status in a deprived neighborhood may include exposure to “environmental stressors and risk factors”. Socioeconomic inequalities are commonly measured by the Cartairs-Morris score, Index of Multiple Deprivation, Townsend deprivation index, and the Jarman score. The Jarman score, for example, considers “unemployment, overcrowding, single parents, under-fives, elderly living alone, ethnicity, low social class and residential mobility”. In Vos’ meta-analysis these indices are used to view the effect of low SES neighbourhoods on maternal health. In the meta-analysis, data from individual studies were collected from 1985 up until 2008. Vos concludes that a correlation exists between prenatal adversities and deprived neighbourhoods. Other studies have shown that low SES is closely associated with the development of the fetus in utero and growth retardation. Studies also suggest that children

born in low SES families are “likely to be born prematurely, at low birth weight, or with asphyxia, a birth defect, a disability, fetal alcohol syndrome, or AIDS”. [63] Bradley and Corwyn also suggest that congenital disorders arise from the mother’s lack of nutrition, a poor lifestyle, maternal substance abuse and “living in a neighborhood that contains hazards affecting fetal development (toxic waste dumps)”. In a meta-analysis that viewed how inequalities influenced maternal health, it was suggested that deprived neighborhoods often promoted behaviors such as smoking, drug and alcohol use. After controlling for socioeconomic factors and ethnicity, several individual studies demonstrated an association with outcomes such as perinatal mortality and preterm birth.[10]

Prenatal diagnosis has revolutionized prenatal care from the perspective of both the patient and the physician. For the patient, prenatal diagnosis provides genetic, anatomic, and physiologic information about the fetus or fetuses that can be used to make informed and individualized decisions regarding the pregnancy. For the physician, prenatal diagnosis provides vital information that can be utilized for better antepartum management. Information regarding specific anatomic anomalies affords the physician the opportunity to offer the patient sophisticated prenatal procedures, such as fetal surgery or selective fetal reduction in multiple gestations. Likewise, prenatal knowledge about genetic, physiologic, and/or anatomic abnormalities enables the physician to tailor or manage the timing and mode of delivery for optimal maternal and fetal outcomes. Prenatal diagnosis also allows the neonatal and paediatric specialists to be adequately prepared for a potentially ill neonate at delivery. Recent progress in the fields of maternal fetal medicine, radiology, and genetics has resulted in great advances in prenatal diagnosis. Ultrasonography is the initial modality for evaluation of pregnant patient because of its widespread availability and reasonable cost, with other modalities used only if Ultrasonography results are nondiagnostic. Hence based on above data the present study was planned Study of Congenital Fetal anomalies with the Help of Sonographic Evaluation.

Methodology:

The present study was planned in Department of Radio- Diagnosis, Katihar Medical College and Hospital, Katihar, Bihar, India. All pregnant females of

second trimester who are referred from obstetrics and gynaecology department and thus sent to department of Radio diagnosis for antenatal sonographic examination. A complete antenatal ultrasound examination of pregnant women included in the study will be done using gray scale & colour duplex examination.

A complete second trimester antenatal ultrasound examination of pregnant women was done using gray scale & color duplex examination on PHILIPS HD7 machine with a transducer of frequency 3.5 to 5 MHZ. The information about the gestational age, location of placenta, fetal biometry & fetal anomalies was collected. The scans were performed as a standard level one ultrasonography. In cases of uncertain abnormal findings, the women were reviewed by a level two scan with repeated scans. This data was compared with the findings at delivery / termination of pregnancy & appropriate statistical analysis was performed.

All the patients were informed consents. The aim and the objective of the present study were conveyed to them. Approval of the institutional ethical committee was taken prior to conduct of this study.

Following was the inclusion and exclusion criteria for the present study.

Inclusion Criteria: All pregnant women coming for antenatal sonographic examination during the second trimester at Department of Radio diagnosis.

Exclusion Criteria: Females with Multiple gestations.

Results & Discussion:

The recent development of high-resolution ultrasound equipment has markedly improved the diagnostic accuracy of ultrasound. In particular, the introduction of high-frequency vaginal probes has enabled early diagnosis of certain fetal abnormalities from the 12th to 14th week of pregnancy. Such early testing is of special importance for women with a history of pregnancies associated with birth defects.

The study would determine the sensitivity and specificity of ultrasound modality in evaluating congenital fetal anomalies. Many modalities are available to detect congenital anomalies at an early stage like laboratory & imaging studies, out of which sonography has emerged as the investigation of choice. Ultrasound is non-invasive and safe and hence can be used repeatedly. It is quick, inexpensive, and sensitive causing no discomfort to

the patient at any time of gestation. Fetal anomaly scan is usually carried out at second trimester of pregnancy.

Table 1: Age of Pregnant Females

Age	No. of Cases	Number of anomalies detected by ultrasound
Below 20 years	30	0
21 – 25 years	225	8
26 – 30 years	128	5
31 – 40 years	117	1
Total Cases	500	14

Table 2: Type of Anomalies

Organ system	Number of anomalies detected by ultrasound
CNS	4
Genitourinary	3
Gastrointestinal	2
Cardiovascular	2
Musculoskeletal	1
Craniofacial	1
Others	1
Total Cases	14

Women who delay child bearing are at an increased risk of having an adverse outcome of pregnancy. [11] Increasing maternal age is independently linked with definite adverse pregnancy as well as fetal abnormalities and multiple gestation. [12] Bobrowski R et al have statistically shown that mothers between 25 to 30 years of age stand at a higher risk of producing malformed babies. [13] Sugunabai [14] reported a higher incidence of malformation in babies born to mothers aged over 35 years, whereas Datta et al [15] documented statistically insignificant association of increased maternal age and congenital anomalies.

A study conducted by Trish Chudleigh [16] from Rosie Ultrasound Department, Cambridge University Hospitals NHS Foundation Trust stated that Ultrasound screening for fetal abnormalities has been available to pregnant women in England for over two decades. The Department of Health commissioned the Fetal Anomaly Screening Programme to develop and extend the second trimester anomaly scan to ensure an effective and accessible service for all pregnant women in England.

Muhammad Nafees, Muhammad Hamid Akram, Makki Muhammad Afridi and Aqsa Javed have conducted an ultrasonographic study on 200 patients

out of which 134 had different congenital anomalies. [17] The most common congenital anomalies

detected were from central nervous system with relatively more prevalent in cousin marriages.



Figure 1: Sonographic imaging demonstrates atrio-ventricular septal defect



Figure 2: Image demonstrates protrusion over occipital region of fetal head

According to Sarah A. Waller, Theodore J. Dubinsky and Manjiri Dighe [18] Ultrasonography provides patients with an excellent means of screening for anomalies, and the use of soft markers has individualized each patient's decision to pursue diagnostic testing.

A study conducted by Grandjean H et al, on the purpose of the Eurofetus study was to evaluate the accuracy of the antenatal detection of malformations by routine ultrasonography in unselected populations concluded that systemic ultrasonographic screening during pregnancy can now detect a large population of fetal malformations. [19]

A report published in 'The society of Obstetricians and Gynaecologists of Canada' by Yvonne Cargill and Lucie Morin was done to review the benefits and requirements for a complete second trimester ultrasound and the documentation needed. The outcome of the report was that a complete second trimester ultrasound provides information about the number of fetuses, the gestational age, the location of the placenta, and fetal and maternal anatomy. [20]

The benefits of scanning in early pregnancy are therefore divided into several levels: first, earlier diagnosis of normal and abnormal intrauterine pregnancy and the detection of ectopic pregnancy;

second, more accurate dating of early pregnancies on the basis of the measurement of the gestational sac and the crown-rump length; third, measuring the nuchal translucency (NT) at 12 to 14 gestational week as a marker for chromosomal abnormalities (mainly Down syndrome) and certain organ anomalies (mainly in the cardiovascular system); fourth and perhaps most importantly, the ability to detect structural anomalies during the first and early second trimester of pregnancy. [21]

Early diagnosis of presence or absence of congenital anomalies is beneficial to mother, both physically and psychologically. A negative sonogram is certainly reassuring particularly for the couples with an increased risk of fetal anomalies. This reassurance was particularly seen in parents with previous congenital anomalies, where normal early scan reduced the anxiety levels to a great extent. On the other hand, anticipation of a positive diagnosis may be valuable in itself. Earlier detection of fetal structural malformations would allow for earlier antenatal referral to a tertiary care facility and coordination of care among appropriate subspecialists. [22] Parents are mentally prepared for these anomalies and a better co-ordination with specific sub-specialties can be assured to the baby.

Conclusion:

Obstetric ultrasonography has become an important part of routine antenatal care. Routine anomaly screening improves perinatal outcome directly through termination of pregnancy for certain anomalies. Congenital fetal anomalies are one of the most threatening complications which are prevalent in the society associated with severe morbidity and mortality in the new born fetus or neonates. Ultrasound is the best possible non-invasive technique available to detect any congenital anomalies in pregnant women which will help to identify the severity of the disease, its outcome leading to pregnancy termination or gives an opportunity for fetal therapy or better neonatal care.

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