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Role of Antenatal Screening for Congenital Anomalies by Grey Scale Ultrasound to Reduce the Perinatal Mortality and Morbidity

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Conclusion: Congenital anomalies are one of the causes for perinatal mortality and morbidity that can be reduced by different measures including avoidance of risk factors and by Antenatal screening for congenital anomalies on grey scale USG. The risk factors can be reduced by the public as well as obstetricians awareness. Ultrasound is simple, noninvasive and sensitive enough for anomaly detection and may have a vital role in reducing perinatal mortality and morbidity by screening and early detection of anomalies. Therefore, screening of all pregnant women by second-trimester ultrasound especially by experienced radiologist should be done.

Keywords: anomaly scan, antenatal ultrasound, fetal anomalies, perinatal mortality and morbidity.

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I. Introduction

WHO defines any gross structural or functional disorder recognized at the time of birth as Congenital anomaly (1). According to one study, congenital anomalies affect 1 in 33 infants. Worldwide 3.2 million of babies born with congenital abnormalities may suffer from mental or physical disability every year (2). In developing countries like Pakistan having a lack of social support system, parents may have huge financial, psychological and social burden with bringing up a disabled child.

Congenital anomalies which are not diagnosed antenatal present as emergencies in pediatric surgical departments and is included in the list of significant causes for perinatal mortality and morbidity.(1)

Birth defect is a major cause of morbidity and mortality in infants worldwide; One study shows that birth defects were the cause of approximately 260,000 deaths in the year 2004 alone, 7% of these were neonatal deaths (3). The proportion of congenital defects related to perinatal deaths are increasing might be due to the reduction of other causes of mortality because of improvement of perinatal and neonatal care. (1) Congenital anomalies are the cause for 6-9% of perinatal deaths in Pakistan that makes a significant proportion which can be reduced by a prenatal diagnosis of gross fetal defects. (4)

The prevalence is variable among populations of the different area. Different studies show the prevalence of congenital anomalies as 2.5% in India and 3% in the United States and the United Kingdom (5, 6). Cardiac and orofacial anomalies, Down syndrome and neural tube defect are the most common anomalies reported worldwide (7, 8). Different risk factors have been recognized to contribute to these defects. These risk factors include Maternal age, maternal illness, radiation exposure, poor nutritional status with Folic acid deficiency, drug intake cousin marriage and smoking (9, 10).

Different measures can be utilized to reduce the congenital defects related to mortality and morbidity. Avoiding risk factors can reduce the incidence of fetal defects. Therefore, the knowledge about the risk factors in a specific region or population may help in reducing

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the risk. On the other hand antenatal anomaly scan for the screening of congenital fetal defects may assist in deciding the termination of pregnancy or therapeutic intervention including perinatal surgical planning in case of major anomalies (11).

Although there are different laboratory tests or radiological modalities are available for detection of fetal anomalies, but Grey scale ultrasound has well-established role for diagnostic abilities as it gives a large amount of structural information (11, 12). A fetal defect can be detected at any stage of pregnancy, but all depends upon when a patient visits Sonologist for anomaly scan. The most sensitive time for anomaly detection is the second trimester around 18 to 24 weeks (11, 13). Detection of anomalies at this stage carries significance for decision as termination of pregnancy in cases where major anomalies are detected (14).Overall success for detection on grey scale USG depends upon gestational age, available system, the skill and experience of radiologists (11).

II. Materials and Methods

We conducted this prospective study from January 2016 to December 2017at Radiology department of MINAR cancer hospital Multan in co-ordination with children hospital complex Multan, Pakistan. It is a tertiary care hospital covering most of the area of southern Punjab consisting of approximately more than 50% of the total country population. We enrolled 3098 patients for an obstetrical scan during the study period. The details of these patients including their age, gestational age, and parity, history of any medical disease, socioeconomic status, and folic acid intake were recorded in predesigned Performa. DATA recorded was analyzed statistically by applying SPSS-24.

III. Results

Out of 3098 patients, we detected 78 patients having at least one gross fetal defect. We categorized the anomalies detected on grey scale ultrasound according to body system involved including Central nervous system (CNS), cardiovascular system (CVS), gastrointestinal system (GIT), genitourinary system (GU) and musculoskeletal system MSK. Congenital fetal defects have a prevalence of 2.38 % of the study population. The mean age of patients was 25.89 years SD± 5.24 and mean gestational age was 26.5 weeks SD ±6.43. Only 5.4% of women were above age of 30 years. None was above 35 years of age. The congenital fetal defects involving (CNS) were highest contributing 48.6 % followed by (MSK) and (GU) 13.5 % and 8.1 % respectively. Involvement of (CVS), (GIT) and respiratory system (RS) was lowest with a prevalence of 2.7% of each. Oligohydramnios was found associated with polycystic kidney diseases, and poly hydramnios was common in patients with neural tube defects and gastrointestinal obstruction. Twelve of total patients (15% of total anomalies) were detected with major anomalies. Termination of pregnancy was suggested for them following the recommended indications as per literature (15, 16). Nine of these were having CNS anomalies including severe hydrocephalus with cortical thinning and myelomeningocele, cystic hygroma and anencephaly. Three of these were having Thonatophoric dysplasia, a type of dwarfism with associated anomalies of a narrow chest and underdeveloped lungs and is lethal with poor outcome (17).

Only 5% of women belonged to high socioeconomic status while rest of the 95 % belonged to the low and middle socioeconomic group with a p-value of 0.003. None of the women with detected fetal defects was taking folic acid. 81.1 % of women had Cousin Marriage with a significant p-value of 0.0016.

IV. Discussion

Approximately 2-3 % of live births are affected by congenital anomalies that contribute significantly to perinatal mortality and morbidity (18). It also affects the quality of life of survivors that may cause a social and financial burden on parents especially in developing countries like Pakistan.

Anomalies are classified as major or minor depending upon the fetal outcome and further management. Anomalies that need medical and surgical intervention are categorized as major, and these may have an impact on perinatal mortality and morbidity. An anomaly that does not require any medical or surgical intervention and is compatible with life expectancy is categorized as minor (11).

The incidence of antenatal detection of congenital anomalies is 2.38 % in our study group that corresponds with the figures given in the literature. The most commonly detected anomalies in our case are those of CNS constituting 48.6%. The literature review also shows the CNS anomalies as commonest followed by MSK and GU. The mean age of mothers and gestation was 25.89 years SD ±5.2 and 26.5 weeks SD ±6.4 respectively. The number of women presented in the second trimester was 36 (66.6%) and presented in the third trimester was 28 (33.3%). No anomaly was detected in the first trimester. Different studies show different gestational ages for anomaly detection. For example, a study by Fatema shows respondents between 34 to 36 weeks and a study by the Padma shows anomaly detection between 29 to 32 weeks of gestations (19, 20). The most sensitive and significant time for anomaly detection is second-trimester USG. The morbidity and mortality related to anomalies increase with increasing gestational age. Therefore to avoid such complications anomalies should be detected earlier (11).

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Literature shows cousin marriage which is highly practiced in our population is one of the main risk factors for congenital anomalies with a p-value of 0.0018 (21, 22) and was seen in 60 (81.1%) women of our study group with a p-value of 0.0016. Increased risk of anomalies in cousin marriage has been explained as the homozygous expression of a recessive gene inherited both parents (1). Folic acid deficiency is one of the recognized risk factor for congenital anomalies. Many studies have shown its association with neural tube defects. Folic acid intake reduces the risk of neural tube defect (23). In our study, none of the women was taking folic acid. Correlation of smoking with congenital anomalies has been shown by some studies (23), but none of the mothers in our study group had a history of smoking. Advanced mother age >35 years is a high risk factor for chromosomal anomalies (24), but in our study, none of the mothers was above 35 years of age as we did not conduct a chromosomal analysis. Both the maternal age and maternal smoking are not significant risk factors in our population.

V. Conclusion

Congenital anomalies are one of the causes for perinatal mortality and morbidity that can be reduced on the one hand by avoiding risk factors and on another hand by Antenatal detection of congenital anomalies on grey scale USG. In our population, the common important risk factors for congenital anomalies include Poor socioeconomic status, lack of folic acid intake and cousin marriages. These risk factors can be reduced by public awareness. USG is a simple, noninvasive and cost-effective investigation and has a significant established role in early detection of congenital anomalies. So we recommend antenatal screening for anomalies by second-trimester USG of all pregnant women.

Abbreviation list

CVS - Cardiovascular system.
CNS - Central nervous system
GUS - Genitourinary system.
MSK - Musckuloskeletal system.
RS - Respiratory system.
USG - Ultrasound.

Conflict of Interest

The authors declare that they have no conflict of interest.

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Ethical approval

This study was done by the approval of ethical committee of the institute.

Contribution of authors

All the authors contributed significantly in manuscript writing. Dr Rubina Mukhtar is main author for manuscript writing (50%). Dr Mukhtar Hussain (10%), M. Ahmad Mukhtar (10%), Dr Isharat (10%), Rubaida Mehmood (10%) and Dr M. Saqib (10%) contributed in Data recording Data analysis and helped in manuscript writing.

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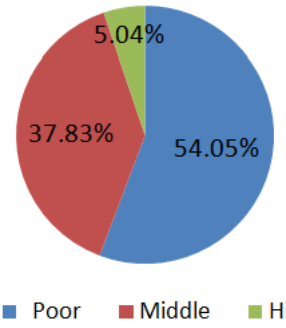
Table 1: Demographic Data of Patients

Total Number of Patients=76		
Types of anomalies	NO.	%
CNS	36	48.6
CVS	2	2.7
GIT	2	2.7
GU	6	8.1
RESP SYS	2	2.7
MSK	10	13.5
MULTISYS	16	21.6
Age of patients		
<20 years	10	13.5
20-25 years	28	37.8
25-30 years	28	37.8
>30-35 years	4	5.4
>35 years	0	0
Gestational Age		
<14 weeks	2	2.6
>14-20 weeks	22	28.9
>20-28 weeks	26	34.2
>28 weeks	26	34.2

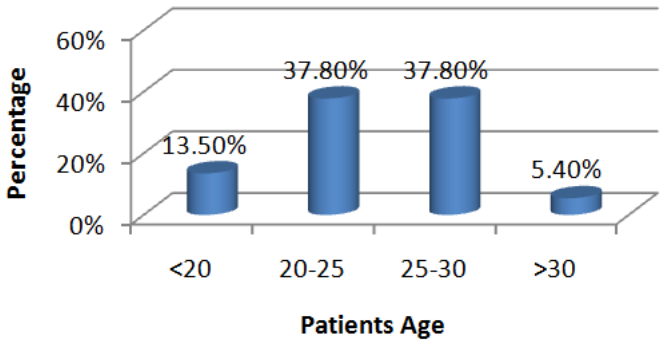
Table 2: Risk factors n=76

Risk Factors	Yes (%)	No (%)
Lack of Folic Acid intake	76(100%)	0
Cousin marriage	60(81.1%)	16(18.9)
Poor socioeconomic status	40(54.05%)	36 (45.95)
Maternal age>35	0	76 (100)
Smoking	0	76 (100)
Previous history of anomalies	18(23.7%)	58(76.3) (76.3)
History of medication of mother	4 (5.2%)	72(94.7)

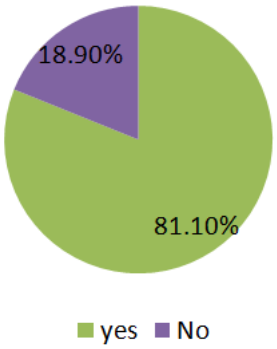
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Graph 1: Socioeconomic Distrubution of Patients



Graph 2: Age Distrubution of Patients



Graph 3: Distrubution of Patients by Cousine Marriage

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