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ORIGINAL ARTICLE

Frequency of Various Congenital Anomalies and Associated Maternal Risk Factors, an Experience at Tertiary Care Hospital

JAVARIA ASLAM[1], SADAF KHURSHID[2], SHEEBA REHMAN[3], SHAHNILAH ZAFAR[4], IRAM ASLAM[5], SARA GULBAZ[6]
[1]*Specialist in Indus Hospital Lahore*
[2]*Senior Registrar University of Lahore Teaching Hospital Lahore*
[3]*Assistant Professor Gyne and Obs Rai Medical College Sargoodha*
[4]*Senior Registrar Gyne and Obs Faima Memorial Hospital Shadman Lahore*
[5]*Associate Professor of Gyne Independent Medical College Faisalabad*
[6]*Consultant in Indus Hospital Lahore*
Correspondence to: Javaria Aslam, Email: Dr.javaria.2002@gmail.com, Cell: 03347946510

ABSTRACT

Objective: To determine frequency of congenital anomalies in children and associated risk factors of mothers.
Setting: Department of obstetrics and gynecology Rai Medical College Sargodha.
Study design: Descriptive observational study.
Duration: July 2022 to December 2022.
Methodology: All babies with congenital anomalies born within study duration and their mothers were included in this study. Anomalies of neonates were evaluated after proper clinical evaluation by a neonatologist. A questionnaire was designed to evaluate maternal risk factors. All collected data was analyzed using SPSS version 20.
Results: Total 1900 babies delivered during this study period and out of them congenital anomalies were found in 31 (1.6%) cases. Most common anomaly was hydrocephalus in 10(32.2%) and meningomyelocele in 6(19.3%) cases. Most common maternal risk factor was consanguineous marriage in 15(48.3%) cases. Most commonly mothers with the age of 19-30 years were having babies with congenital anomalies.
Practical Implication: This study helps us to understand the importance of avoiding consanguineous marriages, screening of high risk cases using various diagnostic techniques like ultrasonography during 16-20 weeks, use of folic acid supplements and in case of detecting congenital anomalies option of selective termination of pregnancy can be offered to the parents. Proper awareness of the parents and their counselling after birth of a baby with anomaly can prevent its recurrence in their future pregnancies
Conclusion: Most common congenital anomaly reported in this study was hydrocapahlus followed by meningocele and most common maternal risk factor associated with these anomalies was consanguineous marriage.
Keywords: Congenital anomaly, consanguineous marriage, Maternal risk factor, Neural tube defects

INTRODUCTION

Frequency of con genital anomalies is increasing with the passage of time causing increased mortality rate of newborns.[1] Congenital anomalies include metabolic abnormalities, structural defects and chromosomal abnormalities. Structural defects may be grossly visible or microscopic that either can be detected in utero by various investigations or after birth.[2] According to a study 6-9% deaths occur annually due to congenital abnormalities in Pakistan.[3] Congenital malformations may be present as a single or group of abnormalities which occur due to dysmorphogenesis.[4] Structural defect occur usually in isolated congenital abnormality due to single defect in morphogenesis while combined abnormalities occur due to multiple morphogenic defects happening during the development of the baby.[5] Diabetes mellitus, genetic mutations and hereditary history are main risk factors of congenital abnormalities.[6] Maternal risk factors should be ruled out and preconception screening should be done to avoid such abnormalities. In such cases protective measures can be devised before conception such as use of folic acid plays important protective role in preventing congenital abnormalities. According to a recent study about 65-75% cases of congenital abnormalities have multifactorial cause.[7]Various factors can increase its risk such as consensual marriages causes autosomal recessive chromosomal disorders. Increased maternal age is directly associated with increased rate of birth defects such as Down's syndrome.[8] Gestational diabetes mellitus and family history of diabetes mellitus increases the risk of congenital abnormalities. Infection and high grade fever during first trimester is a predisposing factor to central nervous system anomalies.[9] Early detection of congenital anomalies can be done by ultrasonography and serum markers that is common practice in developed countries but not so common in developing countries. Ultrasound examination can detect congenital anomalies in 70-80% cases.[10]This study will help us to understand frequency of congenital anomalies in our population and its maternal risk factors so that before pregnancy or during conception necessary measures may be taken to avoid the risk factors and anomalies may be prevented.

MATERIALS AND METHODS

This is a cross sectional study conducted in the department of obstetrics and gynecology Rai Medical College Sargodha. Study was started in July 2022 and completed after six months duration in December 2022. Study sample was calculated using WHO sample size calculator. Sample selection was done by nonprobability consecutive sampling technique. History was taken from the mothers regarding age, parity, any infection associated with rash during pregnancy, cousin marriages, history of previous abortion, large size baby previously, diabetes mellitus, hypertension, family history of congenital anomalies and drug history during first trimester. After taking history proper physical examination was done. Physical examination of the baby with any congenital anomaly was also performed. Baseline laboratory investigations like complete blood count, renal and liver function tests, blood sugar, Rh factor and blood grouping were done. Echocardiography of the heart and ultrasound of abdomen and pelvis was also done in all babies for detecting any congenital anomaly. Serum markers in few mothers were tested during pregnancy. A self-made proforma was designed in which all relevant data was documented. Data was analyzed using SPSS software version 20. Percentages were determined for qualitative variables and means with standard deviation were calculated for quantitative variables. Chi square test was applied on the data. P-value less than 0.05 was considered significant.

RESULTS

Total 1900 babies were studied out of them congenital anomalies were found in 31 (1.6%) cases. Most common anomaly was hydrocephalus in 10(32.2%) and meningomyelocele was second common anomaly in 07(22.5%) cases. Ages of the mothers were 16-37 years with mean age of 25.74 ± 3.2 years. Most commonly

mothers with the age of 19-30 years (64.5%) were having babies with congenital anomalies.

Table-1: Congenital anomalies of Central Nervous System (%=n/31)

Congenital anomalies	Number of patients	Percentage
Hydrocephalus	10	32.2%
Meningomyelocele	07	22.5%
Anencephaly	06	19.3%
Meningocele	03	9.7%
Encephalocele	02	6.4%
Microcephaly	02	6.4%
Spina bifida	01	3.2%

Talipes equinovarus was the most common anomaly in musculoskeletal system found in 03(9.7%) cases. Most common anomalies in the babies were found related to central nervous system (n=31) followed by musculoskeletal system (n=6), gastrointestinal system (n=4), genitourinary system (n=4), miscellaneous anomalies (n=3) and facial anomalies (n=3). In 11(35.5%) cases only one system was involved while in 20(64.5%) cases more than one body systems were having anomalies. Most common risk factor of congenital anomalies in mothers was consanguineous marriages in 21(67.7%) cases. Central nervous system involvement was found in all the babies with congenital anomalies.

Table-2: Congenital anomalies of Musculoskeletal System (%=n/31)

Anomalies	Number of patients	%age
Talipes equinovarus	3	9.7%
Chondroplasia	1	3.2%
Syndactyli	1	3.2%
Polydactyli	1	3.2%

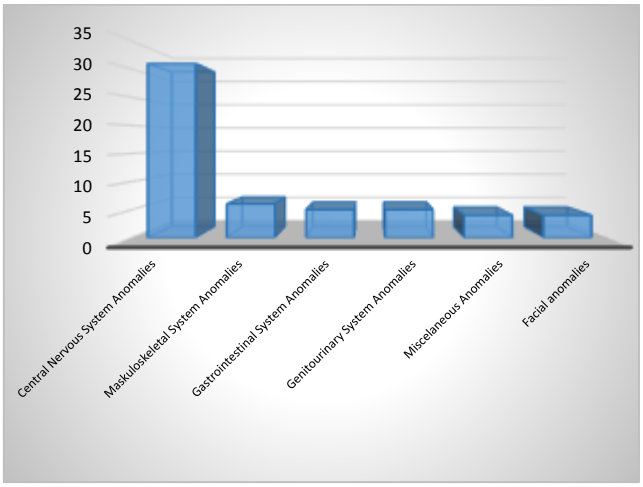

Figure-1: Frequency of congenital anomalies of various body systems

Table-3: Frequency of maternal risk factors (%=n/31)

Risk Factors	No	%age
Consanguineous marriage	21	67.7%
Fever/ infection during pregnancy	11	35.5%
H/O of still birth	07	22.5%
H/O abortion	05	16%
H/O diabetes mellitus	04	12.9%
H/O drug intake in pregnancy	04	12.9%
Family H/O of diabetes mellitus	04	12.9%
H/O cardiac disease in mother	04	12.9%
H/O congenital anomalies previously	02	6.4%
H/O smoking by mother	02	6.4%
H/O drug intake by mother during pregnancy	01	3.2%

DISCUSSION

Congenital anomalies are increasing in rate with the passage of time.[11] Its most important risk factor is cousin marriages which increase the frequency of congenital anomalies from 2-3% to 5-8%.[12] In UAE frequency of cousin marriages is 54% hence incidence of congenital anomalies is 10.5% that is a very high rate.[13] In Egypt frequency of cousin marriages is 37% and incidence of congenital anomalies is 11.4%.[14] According to a recent study conducted in Pakistan prevalence of congenital anomalies was 40% in families with history of cousin marriage as compared to 26% in non-related parents.[15] In our study congenital anomalies were found in 31(1.6%) cases. Most common anomaly was hydrocephalus in 10(32.2%) and meningomyelocele was second common anomaly in 07(22.5%) cases. Ages of the mothers were 16-37 years with mean age of 25.74 ± 3.2 years. Most commonly (64.5%) mothers with the age of 19-30 years were having babies with congenital anomalies. In our study 12.9% mothers were diabetic as compared to 25% as reported in other previous study. Congenital anomalies incidence is 6-13% in diabetic mothers as compared to 1-3% in non-diabetic mothers.[16]This high rate of anomalies can be reduced by proper metabolic control before conception and during pregnancy especially during the period of organogenesis of the fetus. Recent studies have been reported that pre-gestational diabetes mellitus increases chances of anomalies in fetus by 3-5 times.[17] Central nervous system related defects were most common in this study reported in 18/1000 live births. According to a study conducted in India neural tube defects are most common anomalies detected in 4-15/10000 live births and in USA its incidence is 1/2000 live births.[18] Hydrocephalus and meningomyelocele were most common neural tube defects in our study reported in 32.2% and 22.5% cases out of total 31 cases with birth defects. Neural tube defects can be avoided by using folic acid during pregnancy and regarding this public awareness is necessary. If anomalies detected early during pregnancy then option of selective pregnancy termination can be given to the parents.[19]Many diagnostic investigations can help in early detection of congenital anomalies like ultrasonography, amniocentesis, chorionic villus sampling and biochemical tests.[20] Hereditary genetic mutations are another important cause of birth defects. In our study most of the babies (64.5%) with birth defects were born to the mothers with the age of 19-30 years as compared to other studied which reported 32% mothers with age more than 35 years.[21]As most of the mothers were young that may be the cause that no case of downs syndrome was detected in this study. Incidence of cardiovascular anomalies was low in this study that may be due to lack of proper neonatal follow-ups and diagnostic tools.

Medical resources, diagnosis, and treatment must improve in developing countries. There are limited resources: access to medical and health resources; knowledge about disease; awareness, trainings, and awareness about health. Health literacy is mandatory for any disease and facilitates the patients access to resources, databases, and trainings about the disease.[22-28] This study helps us to understand the importance of avoiding consanguineous marriages, screening of high risk cases using various diagnostic techniques like ultrasonography during 16-20 weeks, use of folic acid supplements and in case of detecting congenital anomalies option of selective termination of pregnancy can be offered to the parents. Proper awareness of the parents and their counselling after birth of a baby with anomaly can prevent its recurrence in their future pregnancies.

CONCLUSION

In this study incidence of congenital anomalies was 16/1000 live births. Consanguineous marriages are main risk factor of congenital anomalies in Pakistani population. Neural tube defects were the most common anomalies reported in this study. Mostly babies with anomalies were born to young mothers with age <30 years. Early detection of anomalies using screening techniques and counselling of the parents and selective pregnancy termination in case of anomalies detection can reduce rate of birth defects.

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