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CONGENITAL MALFORMATIONS

DR. SHAMAMA HASAN
FCPS (Paeds)
Children Complex
MH Rawalpindi

DR. HUMA FARRUKH, FCPS
Classified Child Specialist
CMH, Quetta

DR. AMEEN ULLAH
Registrar Pediatrics
MH Rawalpindi

Dr. Nadeem Sadiq, MCPS, FCPS
Classified Child Specialist
CMH Muzaffarabad

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ABSTRACT...Objective: To determine frequency, pattern of distribution of congenital malformations in newborn. **Design:** Cross-sectional observational study. **Place and Duration of study:** Study was carried out in the Department of neonatology, Military Hospital Rawalpindi during the period of January 2008 to Dec 2008. **Patients and methods:** All the newborns were examined and mothers were interviewed. Baby's antenatal record was also noted and recorded in a Performa. Antenatal ultrasonography record was also recorded. Any internal malformation was also noted. Babies who were found to have any malformations were screened for the congenital malformations. Screening was done with latest laboratory tests , radiological evidences and chromosomal studies for suspected trisomies. **Result:** Congenital anomalies were noted in a Performa. Neural tube defect (NTD) was found to be the commonest type of anomaly. Among the most frequent NTD were hydrocephalus and menigomyelocele. Following that were the cardiac anomalies and genitourinary tract anomalies. Trisomy 21 was detected in two of the babies while one newborn had trisomy 13. **Conclusion:** NTD were the most prevalent anomaly detected and early prenatal diagnosis is helpful in decreasing the indirect prevalence of perinatal mortality by offering early termination. Folic acid supplementation should be advocated before conception

Key words: Congenital anomalies, consanguineous marriages. Neural tube defects.

INTRODUCTION

All the newborn, born with congenital anomalies, during the study period were included in the study. Examination of the newborn was done by a consultant pediatrician and findings recorded in pre-designed performa according to the different systems affected by the anomaly. Any neonate with congenital malformation was thoroughly screened using not only the conventional methods but also the latest diagnostics were used like cytogenetics, renal scans and Doppler scans. Record for antenatal ultrasonography was also sought for the detection of some internal anomalies like cardiovascular and gastrointestinal system. Mothers of the newborn were interviewed. Frequencies and pattern of congenital anomalies were determined. Statistical analysis was done by SPSS version 10.

PATIENTS AND METHODS

A cross sectional observational study was carried out in which all the newborns born at Military Hospital Rawalpindi over a period of 1 year were observed. These newborns were examined and investigated.

Any newborn with any congenital malformation was recorded in a Performa. After complete examination of

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Correspondence Address:
Dr. Shamama Hasan
A -6, First Avenue DHA 1
Rawalpindi
shamamaomar@yahoo.com.ca

the baby including all the systemic examination, the findings were recorded. Babies were also investigated for any structural anomalies of the internal structures.

The investigations included skeletal surveys, 2D echocardiography, abdominal and cranial ultrasonography, CT scans of brain, chromosomal analysis, renal scans and serum markers like 17 OHP, testosterone levels. Trisomies were screened with chromosomal studies and karyotyping. Mothers were also interviewed extensively that included consanguinity, previous births, history of abortions or miscarriages and a detail account of present pregnancy. All the previous antenatal scans were also recorded in a Performa.

RESULTS

Among 11520 babies born over a period of 1 year 196 had congenital malformations 1.70%. Of all the congenital malformations neural tube defects were the commonest. 42 newborns had anomalies related to CNS 21.4%. Most frequent of the neural tube defect were hydrocephalus and meningocephalus 61.9%.

Following that were the cardiac defects 17.34% and the genitourinary defects 7.6%. Two newborns had trisomy 21 and one presented with trisomy 13.

About 1/4th of the malformed babies were born to consanguineous parents. It was also observed that mothers who were not taking folic acid supplements faced the problems more than the mothers who were taking it before conception.

Table-I. Congenital malformations in newborns over a period of 1 year

Congenital Anomalies	Total No of births 11520	
	No	%
Present	196	1.701%
Absent	11324	98.29%
Total	11520	100.0%

PIE CHART. Showing the Percentage of Congenital Anomalies in newborns

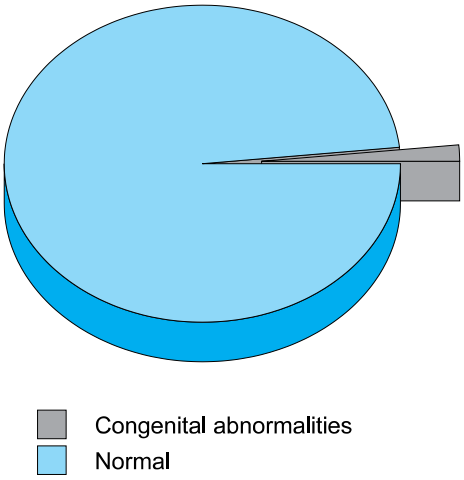

Table-II. Patterns of congenital malformations

Anomalies	No of cases	Anomalies	No of cases
Skeletal	9 (4.59%)	MCA	13 (6.6%)
Genitourinary	15 (7.6%)	Syndromes	11 (5.61%)
CNS	42 (21.4%)	Cleft LIP/Palate	6 (3.06%)
CVS	34 (17.34%)	Cutaneous	5 (2.55%)
RS	3 (1.53%)	Ambigous Genitalia	3 (1.53%)
GIT	9 (4.59%)	Others	46 (23.4%)

DISCUSSION

Congenital abnormalities are an important cause of perinatal mortality. In Pakistan, about 6-9% perinatal deaths are attributed to congenital anomalies. Congenital anomalies refer to structural defects, chromosomal abnormalities, metabolic errors and hereditary disease present at birth. It may occur as an isolated defect or as multiple malformations. Isolated congenital anomaly, is the structural defect, which can be traced down to one localized error in morphogenesis while multiple congenital malformations result from two or more

different morphogenetic errors, which occur during development of individual[3].

The total incidence of congenital anomalies in this study was found to be 1.7% in a total of 11520 babies (Table- I).

Pre-conception screening and counseling offer an opportunity to identify maternal risk factor and to recommend proper protective measures even before the pregnancy begins as pre-conception daily intake of folic acid offer a protective effect[4]. The advanced diagnostic modalities used for prenatal diagnosis of congenital include high resolution sonography, biochemical screening, screening for congenital infections, cytogenetic techniques, pre-implantation diagnosis of genetic diseases and percutaneous umbilical cord sampling. In developed countries, early detection of congenital by offering early selective termination of pregnancy[9].

High resolution sonography by an expert sonologist can diagnose up to 70 – 80% of fetal anomalies in second trimester of pregnancy[10].

Therefore, the prevention and management of congenital anomalies include dealing with known risk factors, early prenatal diagnosis, counseling the couple regarding termination of pregnancy and offering medical treatment or corrective surgery in anomalies by maternal serum marker and sonography has decreased the incidence in neonates utero or after birth.

This study was conducted to determine the frequency and pattern of distribution of congenital malformations in new born.

In our study babies were thoroughly examined and their findings were recorded in a Performa. According to the pattern of congenital malformation the babies were investigated like skeletal surveys for skeletal malformations, cranial ultrasounds and CT scans for NTDs, 2D echocardiography for cardiac malformations, chromosomal analysis for suspicion of trisomies, renal scans for genitourinary malformations, and 17OHP for ambiguous genitalia. The investigations were also recorded in performas.

In accordance with other studies, NTD was the commonest anomaly found in this study which is comparable to a study from Karachi showing 63%[17].

NTD was also reported as most common birth defects from India as 4 to 15 / 10000 live births and in United States as 1/2000 births[18,19].

Hydrocephalus and anencephaly were the commonest NTD found in this study which is in accordance with other reports[11].

Ensuring maternal folic acid supplementation during periconceptional period can lower the incidence of this anomalies[21,22]. Apart from folic acid supplementation, early diagnosis of NTD and advising early termination of affected pregnancies with lethal anomalies will help in decreasing the existing prevalence rate at birth.

Again genetic counseling and early prenatal diagnosis would help in lowering the prevalence in newborn by destroying the fetus before viability. Rate of cardiac malformation in this study was also quite high which is the same to another local study giving a frequency of 4/1000 births. Increased rate of detection of cardiac anomalies is possible by incorporating four chamber views of the heart[23].

Thus, by creating awareness among the studied population regarding the avoidance of consanguineous marriage, periconceptional use of folic acid supplementation, screening of high risk cases and early prenatal detection by routine use of ultrasonography around 16 – 20 weeks and offering termination of pregnancy in cases of lethal anomalies, the perinatal morbidity and mortality can indirectly be reduced.

CONCLUSIONS

The commonest anomaly detected was neural tube defects either isolated or with other malformation.

Frequency of consanguineous marriages was found as the significant associated risk factor for congenital malformations. Avoidance of consanguineous marriage is important in reducing the prevalence by decreasing the hereditary factor, however, this clashes with our cultural norms. Screening of high risk cases, routine preconception and folic acid supplementations, early prenatal diagnosis and termination of fetus with lethal anomaly before attaining viability will reduce the perinatal morbidity and mortality.

RECOMMENDATIONS

Use of folic acid prior to conception and before the first trimester can prevent neural tube defects.

Malformation scan can be used to detect lethal congenital anomalies.

Antenatal testing like amniotic fluid testing and CVS can be used to detect certain lethal congenital anomalies.

Increased maternal age is associated with increased incidence of congenital anomalies; hence maternal education and family planning play a very important role in prevention of congenital anomalies.

Public awareness is of utmost importance regarding antenatal scans, supplementations and consanguinity.

Media campaign should help in this regard.

LHWs and LHVs should be trained to educate people regarding these issues.

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