INCIDENCE AND NATURE OF HUMAN CONGENITAL ANOMALIES

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ABSTRACT

Objectives: To determine the relative incidence and nature of various congenital anomalies (CAs) in 1000 live births at District Peshawar.

Material and Methods: The study sample included 1000 cases, in the age group between 15 and 45 years, admitted in Gynecology and Obstetrics unit of the Kalsoom Maternity Hospital, Peshawar for the purpose of delivery during the study period from June 2013 to June 2014. Complete history was taken on a printed Performa. Besides the preliminary investigations all the cases were subjected to repeated ultrasonic examination in order to look for the progress of pregnancy and any fetal congenital anomaly. After the delivery all the live newborns were thoroughly clinically examined for all types of gross congenital anomalies (CAs). Newborns suspected of having cardiac or intracranial congenital anomalies were subjected to echocardiography and ultrasonographic examination respectively.

Results: Out of 1000 cases, 36 cases were found to have congenital anomalies at the time of birth. Therefore the total incidence/ frequency of CAs in the study group were 3.6%. 22.22% of the CAs belonged to musculoskeletal system. 19.44% were those of central nervous system and similarly 19.44% were of the genitourinary system. The other systems involved were, cardiovascular system 13.88% and craniofacial anomalies 11.11%. Multiple anomalies were detected in 08.33% of the cases while gastrointestinal tract and chromosomal anomalies contributed to 2.77% each.

Conclusions: It was concluded that there is a fairly high incidence of CAs in our population deserving serious attention. Many of the possible risk factors are avoidable if necessary precautions are taken in time. The condition can be detected at an early age in the intrauterine period by ultrasonic examination and possible preventive and curative measures can be taken in time.

Key Words: Congenital anomalies, Newborn, Incidence, Nature.

INTRODUCTION

Pregnancy is a very complex process. Many factors can affect the pregnancy outcome. There are only 25-30% chances of getting pregnant during in each menstrual cycle[1]. For a blastocyst to get implanted is another complex process. So much so that only 70-75% of the blastocysts get implanted. Out of these implanted blastocysts, in only 48% the pregnancy progresses while the remaining 58% do not survive till the second week of gestation[2]. There are so many other factors, including genetic[3] and environmental[4,5,6] factors, which can affect not only the progress of pregnancy but its outcome also. The general incidence of congenital anomalies varies considerably in various populations. This accounts for 1.5 to 3% of all births. Almost 20-30% of perinatal deaths in the developed countries are due to congenital anomalies and 50% of the babies with congenital anomalies die in infancy. About 50% of the affected children suffer from severe mental and physical handicaps[7]. Congenital anomalies can effectively be diagnosed on ultrasonography. As 80-90% of the cases of CAs occur without any risk factor[8], it is mandatory to have repeated ultrasonographic examination of the whole obstetric population in order to have prenatal diagnosis of structural birth defects. By avoiding many of the known risk factors, the burden of CAs can be minimized and thereby its social and economic impact on the concerned families and the community can be reduced.

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MATERIAL AND METHODS

The study was conducted to assess the general incidence and nature of congenital anomalies in this region. The study sample included 1000 females in the age group between 15 and 45 years, admitted in Gynecology and Obstetrics unit of Kalsoom Maternity Hospital, Peshawar for the purpose of delivery during the study period. Detailed history was taken on a printed Performa from all the admitted cases regarding the total duration of pregnancy, any abnormal features or findings, infections, drugs intake, exposure to radiations (X-rays etc.) or any episode of rise of temperature during pregnancy. The number and outcome of previous

pregnancies (if any) was inquired. Repeated prepartum ultrasonography of each case was done to know the progress of pregnancy, fetal congenital abnormalities and various contributing risk factors. All the cases were followed till delivery. All the Live newborns were methodically examined for any obvious congenital anomalies. Suspected cases were subjected to ultrasonography and echocardiograph to look for any intracranial and cardiac anomalies respectively.

RESULTS

A total number of 36 cases were identified in the whole study population of 1000 cases, to have congenital anomalies at the time of birth. Therefore the total incidence/ frequency of CAs in the study group were 3.6%, excluding CAs in stillbirths (Table 1).

Congenital anomalies were identified in various systems of the body. CAs detected in the whole study population are given in Table 1 and Table 2. Majority of the CAs in the study population were detected in the musculoskeletal system, constituting 0.8%. Central nervous system and genitourinary system were the other commonly involved systems contributing 0.7% each. The other systems involved being cardiovascular system 0.5% and craniofacial anomalies 0.4%. Multiple anomalies were detected in 0.3% of the cases while gastrointestinal tract and chromosomal anomalies contributed to 0.1% each.

Numbers of congenital anomalies were detected on ultrasonic and echocardiographic examination, including 3 cases of Ventricular septal defect, 2 cases each of Hydronephrasis/Hydroureter and Hydrocephalus and one case each of Intracranial calcification, Polycystic Kidney, Pulmonary Stenosis and Transposition of great arteries. (Tables 3).

DICUSSION

The data accumulated in this study has been set out in simple tabular form and the information given in them is comprehensive. Further detail has been discussed in the text only where it seemed essential to explain or to stress what seemed of particular interest or importance.

The present study covers several aspects of the problem in this part of the country. The study indicates that the problem is fairly common in the new born. The overall incidence of the problem was 3.6% (36/1000) which is very similar to the findings observed by Hematyar M and Khajouie P (2004) in a study carried out at Tehran Islamic Azad University, Iran, with the frequency of 3.5% (35/1000) live births[9]. The data is also close to the one noted in Atlanta, USA i.e. 3.1% (31/1000) in live births[10]. Some of the studies have reported lower

Table 1: The nature and frequency of CAs detected in 1000 cases

Types of Congenital Anomalies	n	%
Club foot/ feet	05	0.5
Newborns with multiple anomalies	03	0.3
Ventricular septal defect	03	0.3
Meningocele/ Encephalocele/Anencephaly	03	0.3
Cleft lip/Cleft palate	03	0.3
Hydronephrosis/ Hydroureter	02	0.2
Hydrocephalus	02	0.2
Ambiguous genitalia	01	0.1
Congenital cataract	01	0.1
Congenital wrist drop	01	0.1
Cryptorchidism	01	0.1
Down syndrome	01	0.1
Erb's palsy	01	0.1
Hydrocele	01	0.1
Hypospadia	01	0.1
Imperforate anus	01	0.1
Intracranial calcification	01	0.1
Polycystic kidneys	01	0.1
Arthrogryposis	01	0.1
Polydactaly	01	0.1
Pulmonary stenosis	01	0.1
Transposition of great arteries	01	0.1
Total	36	3.6

Table 2: System wise incidences of congenital anomalies in 1000 cases

System involved	n	%
Musculoskeletal	8	0.8
CNS	7	0.7
Genitourinary	7	0.7
CVS	5	0.5
Craniofacial	4	0.4
Multiple systems	3	0.3
GIT	1	0.1
Chromosomal	1	0.1

Table 3

Types of congenital anomalies	n	%
Ventricular septal defect	03	0.3
Hydronephrosis/Hydroureter	02	0.2
Hydrocephalus	02	0.2
Intracranial calcification	01	0.2
Polycystic Kidney	01	0.1
Pulmonary stenosis	01	0.1
Transposition of great arteries	01	0.1

Detected on Ultrasound and Echocardiograhy

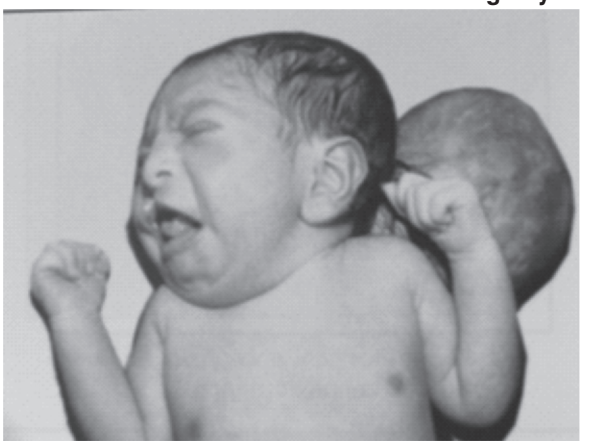

Fig 1: Meningoencephalolocele

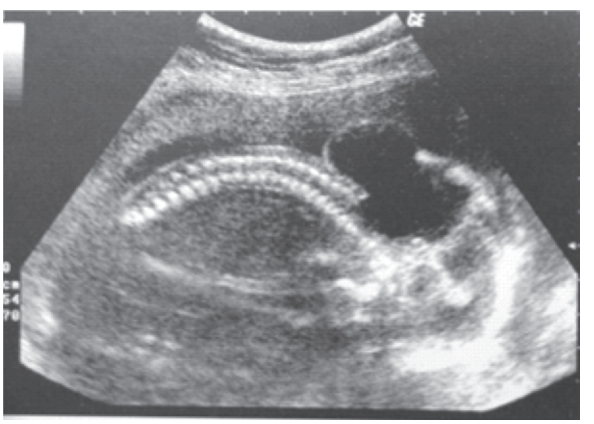

Fig 2: Fetal Anencephaly on ultrasound

incidence figures than our finding. Such figures include, 2.72% (27.2/1000) in India[11] and 2.02% (20.2/1000) in Spain[12].

In the present study male subjects were affected more as compared to females. Their ratio being 5:4. Males having less resistance to oxidative threats as compared to females are more vulnerable. Similar finding has been observed in the study conducted on 9386 cases in India presenting with 179 cases of CAs 13 and the studies conducted by Di Renzo et al in 2007[14] and Inge M et al 2009[15].

Regarding the nature of concerned congenital anomalies, musculoskeletal anomalies were the commonest, constituting for 22.22% of the total number of cases. Musculoskeletal anomalies take the major share in some of the other studies also, including a large study conducted on 2365 births in Uganda[16], on 1000 live births in Iran in 2004 and on 2000 live births in Jammu[17]. CNS anomalies were the second higher in frequency in the study group because some of the cases with CNS anomalies were still born and so were not included in our study. This confirms the finding that there is increase rate of congenital anomalies in still births. The disparity in the incidence in various study groups can be due to variation in exposure to various causative factors like environmental and or genetic factors and poor living standards of a community[18].

RECOMMENDATIONS

In view of the facts described in the present study it is highly recommended that all the pregnant females should be subjected to antenatal ultrasonography for early detection and timely management of the congenital anomalies. Recognition of all the risk factors is obligatory for the possible preventive measures to be taken to help minimize the burden of the problem.

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AUTHOR'S CONTRIBUTION

Following authors have made substantial contributions to the manuscript as under:

Hamid A: Creation of idea and collection of data.

Inayat Q: Supervised the research.

Khan J: Data analysis and statistics.

Authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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