Original Article

FREQUENCY AND TYPES OF VARIOUS CRANIOFACIAL CONGENITAL ANOMALIES (CAS) IN 1000 LIVE BIRTHS

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ABSTRACT

Objective: *To determine the frequency and various types of craniofacial congenital anomalies in 1000 live births at District Peshawar of Pakistan.*

Methodology: *The study was carried out in the Gynecology and Obstetrics units of Khyber Teaching Hospital Peshawar and Kulsoom Maternity Home Peshawar during the study period from 01.06.2014 to 30.06.2014. A total number of 1000 pregnant women admitted for the purpose of delivery, in the age group between 16 and 40 years, were included in the study sample. Detailed history was taken on a printed history sheet. Besides preliminary investigations, all the cases were subjected to repeated ultrasound examination throughout the complete duration of pregnancy. After the delivery all the live newborns were thoroughly examined for all types of gross congenital anomalies, especially of the craniofacial region. Newborns suspected of having congenital anomalies other than the obvious ones, were subjected to ultrasonographic examination of the respective system. Echocardiography was done in newborns suspected of having cardiovascular anomalies.*

Results: *In the study population of 1000 cases, a total number of 36 cases were detected to have congenital anomalies at the time of birth, including anomalies of the craniofacial region. Thus the incidence/ frequency of Congenital Anomalies were 3.6%. Congenital Anomalies were detected in almost every system of the body. Out of the 36 cases, Craniofacial congenital anomalies were detected in 14 (38.88 %), of which clift lip and palate were reported in 35.71% cases.*

Conclusions: *Craniofacial congenital anomalies are fairly common in this region requiring serious attention and search for the possible risk factors. Many of the possible risk factors can be avoided, provided the necessary precautions are taken in time.*

Key words: *Congenital anomalies, Craniofacial, Frequency, Types.*

INTRODUCTION

Congenital abnormalities (CAs) have been described under so many headings e.g., congenital malformation, birth defects and congenital anomalies. Congenital anomalies take place as a result of abnormal or mal development of pharyngeal apparatus during fetal life. All of the fore mentioned terms represent the similar problems. These descriptive terms are used for structural, functional, metabolic and behavioral problems, present in the fetus or newborn[1]. Congenital anatomic abnormalities are classified into various main groups. These are congenital malformations, disruption, deformation and dysplasia, polytrophic field defect, sequence, syndrome and association[2,3].

Congenital abnormalities in the new born have both social and economic impact on the family in particular and the community in general. Incidence of CAs varies in different countries of the world[4,5]. General incidences of CAs range from 1.5 to 3% of all births. Syndromes (multiple related anomalies with a known cause) constitute about 1%[6]. Craniofacial anomalies are often a component part. In the developed countries, CAs lead to 20-30% of perinatal deaths, while 50% of the babies die in infancy. It also leads to severe mental and physical handicaps in 50% of the affected children[1,7].

There is statistical evidence that congenital malformations in 80 – 90% of the cases occur in infants without any risk factor[8]. The scientific community

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advocate repeated ultrasonographic screening of the whole obstetric population to allow prenatal diagnosis of structural birth defects.

The most important causes of pregnancy loss and complications include chromosomal abnormalities, placental abruption or umbilical cord complications, infections, the placental damage by the mother immune system, increase in maternal age, uteroplacental insufficiency, diabetes, immunological rejection, drugs and teratogenic exposure[9-13].

The purpose of this study was to find out the frequency of CAs in live birth and to design a preventive strategy on the basis of such findings.

METHODS AND MATERIALS

This cross sectional study was carried out to assess the frequency and types of various craniofacial congenital anomalies in the offspring of 1000 pregnant females.

The study was conducted in the Gynecology and Obstetrics units of Khyber Teaching Hospital Peshawar and Kulsoom Maternity Home Peshawar. A total of 1000 gravid females between the ages of 16-40 years, admitted in these hospitals for the purpose of delivery (both O.P.D/Emergency Cases) were included in the study population. After the delivery all the live born babies were examined for all types of gross congenital anomalies in general and craniofacial anomalies in particular.

A printed Performa was used for detailed history, inquiring about the total duration and progress of the present pregnancy and outcome of the previous pregnancies (if any). A thorough history was taken, of any drugs intake, infections such as toxoplasmosis, rubella, cytomegalovirus, herpes simplex, and HIV (TORCH), exposure to investigative radiations, Diabetes or any other medical problems during the present pregnancy. All the cases were subjected to repeated ultrasonographic examination to look for the progress of pregnancy and any fetal congenital anomaly at an early stage. Cases with abortions, miscarriages and still births were excluded from the study population.

RESULTS

Out of 1000 patients, the age range distinguish was gravid females between 16-40 years. CAs (especially of the chromosomal origin e.g. Down syndrome) were more common in the elderly multigravida as compared to the young group. There was male predominance, with male to female ratio of 2-1.6.

In this study population of 1000 gravid females, 36 (3.6%) live born babies were detected to have congenital anomalies of the various organs at the time of birth, including anomalies of the craniofacial region. Out of the 36 cases, various types of Craniofacial congenital anomalies were detected in 14 (38.88 %). Distribution is given in Table-1.

Among the craniofacial anomalies, Cleft Lip and/or Cleft Palate were among the most frequent craniofacial anomalies, constituting almost 35.71% of the total. Other craniofacial CAs in order of frequency are given in Table-2.

DISCUSSION

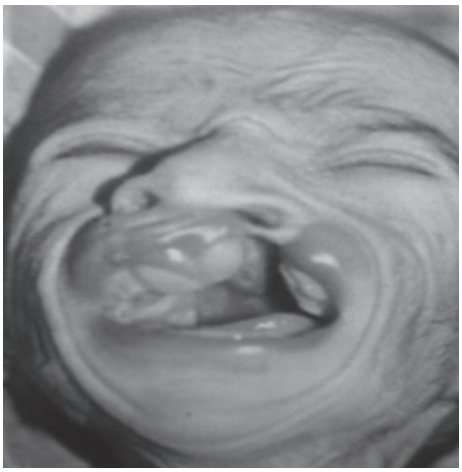
The present study reveals that CAs are fairly common in this part of the world. Incidence of the congenital anomalies in general and that of the craniofacial anomalies in particular varies in different locations of the world. The general incidence of congenital anomalies in this study group was 3.6% with craniofacial anomalies constituting 1.4% and 38.88%

TABLE-1: SYSTEM WISE DISTRIBUTION OF CONGENITAL ANOMALIES

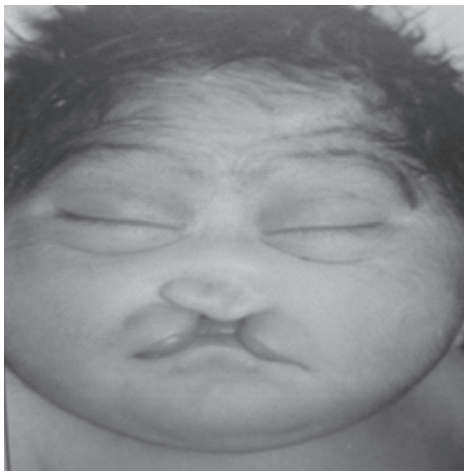
System/Region Affected	n	%
Craniofacial	14	38.89
Musculoskeletal	8	22.22
Cardiovascular	5	13.89
Genitourinary	5	13.89
Gastrointestinal	3	8.33
Chromosomal	1	2.78
Total	**36**	**100**

TABLE-2: DISTRIBUTION OF CRANIOFACIAL ANOMALIES

Types Of Congenital Anomalies	n	%
Cleft Lip and/or Palate	5	35.71
Multisystem involvement	3	21.42
Encephalocele	1	7.14
Anencephaly	1	7.14
Intracranial calcification	1	7.14
Hydrocephalus	1	7.14
Down Syndrome	1	7.14
Congenital cataract	1	7.14
Total	**14**	**100**



Cleft Lip



Cleft Plate

of the total live birth and of the total anomalies respectively. Similar findings were recorded in a study conducted at Tehran Islamic Azad University, Iran, which revealed the frequency of the general conginital amomilies to be 3.5%[14]. The data is also inagreemt with a study done in Atlanta, USA in 1990, where it was found that 3.1% live births were having CAs[15]. However a lower incidences were reported by Verma et al[16] (2.72%) and Martinez[17] (2.02%) respectively.

The incidence of CAs was higher in the male as compared to female subjects in the present study. The ratio was 2:1.6. Male predominance has been established as they have less resistance to oxidative threats compared to females. The finding is consistent with the study conducted by Taksande et al[18] on 9386 cases in India presenting with 179 cases of CAs and the studies conducted by Di Renzo et al[19] and Ever IM et al[20].

As far as the nature of congenital anomalies is concerned, cleft lip and/or palate, both isolated and/or part of the multisystem involvement, were the commonest in our study group accounting for 35.71% of the total number of craniofacial anomalies. In a number of other studies on live born babies cleft lip and/or palate anomalies occured most commonly[21,22,23]. CNS anomalies including Anencephaly, Encephalocele, Hydrocephalus and Intracranial calcification were also the major contributor to the craniofacial anomalies in the present study group. In number of studies conducted in Brazil, Central Nervous System was the most frequent site involved in congenital anomalies[24,25]. The variation in the incidence can be due to difference in exposure to various environmental and or genetic factors in various populations, their habitat and socio economic status.

RECOMMENDATIONS

Craniofacial congenital anomalies are fairly common in this part of the world. There is strong need to find out the causative factors responsible for these anomalies, in order to take the preventive measures in time, to help decrease the burden of the problem in the community. There is need for multidisciplinary approach, both for the prevention and treatment of craniofacial congenital anomalies. Central Nervous System anomalies can effectively be minimized by folic acid intake during and even before the start of pregnancy. Repeated ultrasonography is suggested in all pregnant females throughout pregnancy in order to detect the problem in time.

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