Prevalence of Gross Congenital Malformations at Birth in the Neonates in a Tertiary Care Hospital

Waqas Jehangir, Farooq Ali, Taimoor Jahangir, Muhammad Sajjad Masood

ABSTRACT
Objectives: To determine prevalence, pattern of distribution of congenital anomalies in newborn.
Design: Cross-sectional observational study.
Place of Study: This study was carried out in the Department of Gynecology and Obstetrics, Ward 18, Nishtar Hospital, Multan.
Patients and Methods: This was a cross sectional study. All the women giving birth to babies included. Demographic details, associated risk factors and the type of congenital anomalies in babies were recorded. Diagnosis of congenital anomalies was based on clinical evaluation of newborn by experienced neonatologist.
Results: A total of 611 consecutive neonates delivered in Nishtar Hospital Multan, were subjected to full clinical evaluation. Social data included parental consanguinity and social class. The prevalence of malformations in the 611 hospital live births and stillbirths was 2.95%. Malformed neonates (18) were classified into 06 groups according to the system affected using World Health Organization classification of congenital malformations. The most common anomalies were: central nervous system (38.88%), cleft lip and cleft palate (11.11%), musculoskeletal system (5.55%), ear, face and neck (5.55%), gastrointestinal tract (5.55%) and having more than one defect (33.33%). Parental consanguinity was found in 31.79% of all cases and in 55.5% of malformed cases, thus illustrating the deleterious effects of consanguinity.
Conclusion: The commonest associated risk factor was consanguineous marriage the frequency of which may be reduced by creating awareness regarding the avoidance of consanguineous marriages. CNS related malformations were the most prevalent anomaly detected and early prenatal diagnosis is helpful in decreasing the indirect prevalence of perinatal mortality by offering early termination.
Keywords: Congenital Malformations. Consanguineous Marriage

INTRODUCTION
Congenital malformations have been known and recognized for centuries. It is a stimulating problem for research study because of the high frequency of their occurrence and the devastating effect they may have on the individual and his/her family. Considerable variation in frequency in different populations has been reported, from as low as 1.07% in Japan [1] to as high as 3% in Taiwan [2]. This wide variability could be due to the different methodologies used in the different studies. The primary objective of this study was to determine the prevalence of congenital gross malformations in neonates in Ward 18 of Nishtar Hospital Multan, and to classify the etiologies of malformations in neonates, in order to allow proper genetic counseling, early management and rehabilitation.

SUBJECTS AND METHODS
This was a cross sectional study. This hospital serves both urban and rural area and was therefore chosen as an example of an average public health hospital. All the consecutives babies delivered over the four months period were included in this study. They were examined at birth to identify major congenital defects. Malformations were classified into systems. According to World Health Organization (WHO) recommendations [5], every neonate was given a complete clinical examination. A proforma was completed for every neonate, including clinical and anthropological examination. Social data were obtained from the parents of the neonate, including educational level and occupation of the father and mother, parental consanguinity, number of children in
the family and obstetric history. Anthropometric measurements were made according to the international biological program [6].

RESULTS

The study included 611 consecutive neonates delivered in Ward 18 of Nishtar Hospital Multan during the 4 months period. Of 598 deliveries, 611 neonates were delivered i.e. 598 singletons, 13 sets of twins. Of the 611 neonates, 51 were stillbirths. 431 neonates were full term and 129 were pre-term. The prevalence of congenital malformations in the 611 hospital live births and stillbirths was 2.95%. The congenitally malformed neonates (18) were diagnosed and classified into 06 groups according to the affected system using WHO classification. The percentage was calculated from the total malformed number (18).

Results were categorized as follows:

- Central nervous system anomalies, 07 cases (38.88%)
- Musculoskeletal anomalies, 01 case (5.55%)
- Cleft lip and/or cleft palate, 2 cases (11.11 %)
- More than one defect, 06 cases (33.33%)
- Ear, face and neck anomalies, 01 case (5.55%)
- Gastrointestinal anomalies, 1 case (5.55%)

The frequency and distribution of the 18 birth defects are shown as above. The commonest detected anomalies were those of the central nervous system (38.88%). Of the 18 affected neonates, 13 (72.22%) were live born and 05 (27.77%) were stillborn. Five (27.77%) malformed infants died in the neonatal period. Two cases (11.11 %) had a history of affected relatives of the same or different condition. Other important epidemiological results are shown in Tables 2 and 3. The data in the tables are explained in the discussion.

DISCUSSION

In the present study, the prevalence of congenital malformations among 611 hospital live births and stillbirths was 2.95%. This is much higher than reported in Egypt among live births and stillbirths: 1.16% in Alexandria [3], 1.58% in Cairo [4]. It is higher still than that reported in other populations (12.7/1000) in WHO centers in 16 countries [3]. Other studies among live-born neonates showed different prevalence figures: in Spain (20.23/1000) [7], in Libyan Arab Jamahiriya (23.8/1000) [8], in India (27.2/1000) [9]. On the other hand, the prevalence was lower than that reported previously from a hospital in Teheran (35/1000) for major congenital malformations [10]. The prevalence in the present study is close to that noted in Atlanta, USA (31/1000) among live births only [11].

These variations in prevalence might be explained by social and racial influences which are commonly known in genetic disorders. Also, the results vary according to the background of the investigators and the type of sample studied. In the present study, the number of stillbirths was 51/611 (8.35%), and the number of malformed stillbirths was 05/18 (27.77%). This means that most of the severe congenital malformations were incompatible with life.

Most of the malformed stillbirths in the present study were cases with central nervous system anomalies (38.88%). This is similar to the observations of Rasmussen et al. [11]. The number of neonatal deaths were 46/598 (7.69%) in the normal neonates and 05/18 (27.77%) in the malformed neonates, which confirms that malformations and genetic disorders are a major cause of neonatal death.

Sex was not found to have a role in the occurrence of congenital malformations (Table-1).

<table>
<thead>
<tr>
<th>Table-1 Congenital Malformation With Respect to Sex</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total no. of malformed male neonates</td>
</tr>
<tr>
<td>Total no. of malformed female neonates</td>
</tr>
<tr>
<td>Total no. of malformed Neonates</td>
</tr>
</tbody>
</table>

Regarding birth order (Table-2), there was significant relation between birth order and the prevalence of malformations.

<table>
<thead>
<tr>
<th>Table-2 Congenital Malformation With Respect to Birth Order</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total no. of malformed Neonates from primigravida</td>
</tr>
<tr>
<td>Total no. of malformed Neonates from multigravida</td>
</tr>
</tbody>
</table>

However, our study showed that parental consanguinity was an important cause for most of the malformations (Table-3).
Table-3
Congenital Malformation With Respect to Parental Consanguinity

<table>
<thead>
<tr>
<th>Description</th>
<th>No.</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total no. of malformed neonates with parental consanguinity</td>
<td>10</td>
<td>55.5%</td>
</tr>
<tr>
<td>Total no. of malformed Neonates without parental consanguinity</td>
<td>08</td>
<td>44.5%</td>
</tr>
<tr>
<td>Total no. of malformed Neonates</td>
<td>18</td>
<td>100%</td>
</tr>
</tbody>
</table>

Birth defects in the offspring of first-cousin parents were higher than in the offspring of non-consanguineous parents, which must be taken in account when counseling consanguineous couples. Regarding term of pregnancy (Table-4), there was significant relation between term of pregnancy and the prevalence of malformations. Congenital malformations were much more in pre-term neonates than full term neonates. Regarding weight of neonates (Table-5), there was no significant relation between weight of neonates at birth and congenital malformation.

Table-4
Congenital Malformation With Respect to Term of Pregnancy

<table>
<thead>
<tr>
<th>Description</th>
<th>No.</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total no. of malformed neonates at term</td>
<td>03</td>
<td>16.66%</td>
</tr>
<tr>
<td>Total no. of malformed neonates at pre-term</td>
<td>15</td>
<td>83.34%</td>
</tr>
<tr>
<td>Total no. of malformed Neonates</td>
<td>18</td>
<td>100%</td>
</tr>
</tbody>
</table>

From this study we emphasize that accurate and early diagnosis of congenital malformations is the key to proper management of cases. Premarital counseling is advised, especially in the presence of parental consanguinity and family history of a congenitally malformed child. Because of the high frequency of neural tube defects as revealed by our investigation, we recommend their proper prenatal diagnosis both by abdominal ultrasonography and maternal serum α-fetoprotein at the population level. We also recommend providing periconceptional vitamins and folic acid to all pregnant women. Prenatal diagnosis for other malformations by ultrasonography at around 16 weeks of pregnancy should be a routine procedure.

Table-5
Congenital Malformation With Respect to Weight at birth

<table>
<thead>
<tr>
<th>Description</th>
<th>No.</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total no. of malformed neonates with weight less than 2.5 kg</td>
<td>06</td>
<td>33.33%</td>
</tr>
<tr>
<td>Total no. of malformed neonates with weight between 2.5-3.5 kg</td>
<td>10</td>
<td>55.56%</td>
</tr>
<tr>
<td>Total no. of malformed neonates with weight above 3.5 kg</td>
<td>02</td>
<td>11.11%</td>
</tr>
</tbody>
</table>

Total no. of malformed neonates | 18  | 100%       |

We recommend that all neonates should be thoroughly examined and investigated for congenital malformations. A Pakistani registry of congenital malformations is needed.

REFERENCES

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