Prevalence and Distribution of Congenital Malformations in Newborns in Tertiary care Hospital

Tallat Farkhanda¹, Sehrish Sabir², Hina Gul³

ABSTRACT

Objectives: The aim of this study is to determine the prevalence and distribution of congenital anomalies in live newborns and to study maternal and perinatal risk factors in a tertiary care hospital.

Methodology: This cross-sectional descriptive study was carried out in the department of gynecology and obstetrics from 1st January 2016 to 31st December 2016. All the babies born with congenital anomalies during this period were included. All stillborn were excluded from this study. The newborns were examined for the presence of congenital anomalies.

Results: Out of 8909 live births in Benazir Bhutto hospital, Rawalpindi 47 newborns were diagnosed with congenital malformations, making the prevalence rate of 52.75 per 10,000 live births. Out of 47 babies diagnosed with birth defects, nine died during the first week of life. The predominant system involved was a musculoskeletal system (36.2%) followed by CNS (31.9%). Dimorphic features (21.3%) were the most common anomaly seen in the musculoskeletal group followed by achondroplasia and limb deformities (6.4%).

Cases of congenital anomaly were found in 0.62% of multiparas, whereas in primiparas, the proportion was only 0.34%. Most of the women were aged between 20 and 30 years (74.5%) with only 8% of the mothers were over the age of 30 years. 354 mothers had breech delivery and nine of them showed some congenital anomaly (2.54%) in their babies, whereas in babies born with the cephalic presentation the prevalence was only (0.44%). Congenital anomalies were more likely to be associated with low birth weight, prematurity, multiparity and cesarean delivery.

Conclusion: Regular antenatal visits and prenatal diagnosis are recommended for prevention, early intervention and even planned termination and appropriate treatment after birth when needed.

Keywords: Congenital anomaly, Prevalence, Types of anomalies.

Introduction

Congenital anomalies are also known as birth defects, congenital disorders or congenital malformations. According to WHO congenital anomalies can be defined as structural or functional anomalies (for example, metabolic disorders) that occur during intrauterine life and can be identified prenatally, at birth, or sometimes may only be detected later in infancy, such as hearing defects.⁴ Congenital anomalies affect remarkable proportion of newborn population and contribute significantly to the childhood mortality and hospital admissions.⁵ An estimated 303 000 newborns die within 4 weeks of birth every year, worldwide, due to congenital anomalies. Congenital anomalies can contribute to long-term disability, which may have significant impacts on individuals, families, health-care systems, and societies.⁶ Expenditures of hospitalization...
and treatment procedures for these children impose a large excess burden on health system and their families. Some congenital anomalies can be prevented. Vaccination, adequate intake of folic acid or iodine through fortification of staple foods or supplementation, and adequate antenatal care are just three examples of prevention methods. Although approximately 50% of all congenital anomalies cannot be linked to a specific cause, there are some known genetic, environmental and other causes or risk factors.

**Methodology**

This cross-sectional descriptive study was carried out in the department of gynecology and obstetrics from 1st January 2016 to 31st December 2016. All the babies born with congenital anomalies during this period were included. All still born were excluded from this study.

The newborns were examined and assessed systematically for the presence of congenital anomalies. Diagnosis of congenital anomalies was based on clinical evaluation of newborn babies by the obstetrician and pediatrician. System-wise distribution of the anomalies was performed. For each case, a detailed antenatal and maternal history including the age of the mothers, parity were obtained by reviewing the maternal and labor ward records and by interviewing the parents.

Birth weights >2.5 kg were considered to be normal; whereas, birth weights <2.5 kg were termed as low birth weight (LBW). Babies born at <37 completed weeks (i.e., <259 days), calculated from the 1st day of last menstrual period, were considered as premature. Data was entered into excel data sheet and appropriate statistical analysis was performed.

**Results**

Out of 8909 live births in Benazir Bhutto hospital, Rawalpindi 47 newborns were diagnosed with congenital malformations, making the prevalence rate of 52.75 per 10,000 live births. Among all the newborns, 57 babies were born of multiple gestations. Gender distribution of birth defects was 61.7% in male babies whereas 38.29% in female counterparts. Out of 47 babies diagnosed with birth defects, nine died during first week of life.

The predominant system involved was a musculoskeletal system (36.2%) followed by CNS (31.9%). Dimorphic features (21.3%) was the most common anomaly seen in the musculoskeletal group followed by achondroplasia and limb deformities (6.4%) in each group and likewise hydrocephalic (23.4%) and spina bifida (4.3%) in CNS system. Rare disorders like cystic hygroma and congenital ichthyosis was also seen in (2.1%) each. (Figure 1)

<table>
<thead>
<tr>
<th>Table I: Prevalence of congenital anomalies and parity of mothers.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Parity</td>
</tr>
<tr>
<td>Primiparous</td>
</tr>
<tr>
<td>Multiparous</td>
</tr>
</tbody>
</table>

It has been seen that more than two third of the mothers were aged between 20 and 30 years (74.5%) with only 8% of the mothers were over the age of 30 years. In the present study, 354 mothers had breech delivery and nine of them showed some congenital anomaly (2.54%) in their babies, whereas in babies born with cephalic presentation the prevalence was only (0.44%).(table. II)

<table>
<thead>
<tr>
<th>Table II: Prevalence of congenital anomalies in breech/cephalic presentation.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Presentation at birth</td>
</tr>
<tr>
<td>Breech</td>
</tr>
<tr>
<td>Cephalic</td>
</tr>
</tbody>
</table>

This percentage was about 6 times less babies born via cephalic presentation and was highly significant. Prematurity and LBW was found to have a higher risk of congenital anomalies. The occurrence was about 4.5 times more in case of preterm delivery as compared with the term ones, making it statistically significant. Mode of delivery was also significantly associated with a congenital anomaly and it was about 10 times more in case of cesarean deliveries.
Discussion

Congenital malformations are rapidly emerging as one of the major worldwide problems as they can result in long-term disability, which may have significant impacts on individuals, families, health-care systems, and societies. The exact causes of malformations remain unknown in a large number of the cases. 6

In the present study, the prevalence of congenital malformations in the live newborns were 52.75 per 10,000 live births which is less as compared with the studies from Iran, which reported the incidence of 112.89 per 10,000 live births. In a metaanalysis conducted in Iran from 1992 to 2014 the overall prevalence of congenital anomalies based on the random effect model was determined to be 2.3%. 7 There are also reports from India with the almost double prevalence of congenital anomalies. In the study by Cosme et al. in the city of São Paulo from 2010 to 2014 prevalence of 17.9 cases for every 1,000 live births throughout the time period studied. 9 Different parts of the world representing the different frequency of congenital malformations. 3, 10 In our study the prevalence of congenital anomaly would have been more than the present rate if we could have included the abortions and stillbirths. Through a simple review of the documents, the reason for such a difference in the prevalence of all types of congenital diseases in reports of different studies can be the difference in the prevalence of serious congenital anomalies among various races which environmental factors sometimes have an impact on their incidence. 11

In the present study, out of 47 babies diagnosed with birth defects, nine died during first week of life. Whereas in another study out of 17 alive babies 7 babies died few minutes after birth and 10 babies got admitted in the neonatal ward of which 9 babies also died within one hour following admission. 26

With regard to the pattern of congenital anomalies in the study, the most common system involved was a musculoskeletal system (36.2%), followed by CNS (31.9%), and syndromic babies with multiple anomalies (10.6%), cardiovascular system (8.5%), genitourinary (6.3%), skin (2.12%) etc., Basavanthappa et al. Found that musculoskeletal malformations were the commonest malformation and accounted for 27.5% of all the malformations in a hospital of South India. This was followed by cutaneous 19.16%, genitourinary 15.83%, gastrointestinal 12.5%, neurological 10%, and cardiac malformations 5.83%. 12 Vatanka et al reported that the highest prevalence rates of anomalies were related to musculoskeletal anomalies (27.5%). Other studies also supported our results. 7, 13, 14, 15, 16, 17, 18

In a study in West Bengal, by Pal et al., the cardiovascular, musculoskeletal, and genitourinary system were found to be most commonly involved. 19 Some studies however recorded higher incidence of CNS malformations followed by GIT and musculoskeletal system, 6, 9, 20 whereas Suguna Bai et al. 21 reported GI malformations as the most common one.

More male babies with congenital anomalies than females were noted in the present study 61.7% and 38.29% respectively. A male preponderance was similar to the other studies. 6, 7, 22, 23 It may be because of the fact that the females were afflicted with more lethal congenital malformations and could not survive to be born with signs of life.

Association of LBW with an increased risk of congenital malformations is very well documented. 22 According to our study results the prevalence of congenital anomalies was significantly higher in preterm babies as compared with the full-term babies, which is in conformity with the previous studies reported from India, Iran and Egypt. 18 Devassy U K also reported a greater incidence of congenital malformations among newborns with prematurity and LBW. 25 Mode of delivery also showed a significant association with congenital anomalies in this study with the cesarean section being more commonly associated than normal delivery. Manal et al. reported more than half (55%) of babies with congenital anomalies were born via cesarean section. 27

The relationship between maternal age and babies born with congenital malformations, in our study, revealed that a majority of malformed babies were born of mothers aged 21-30 years; though, it was statistically insignificant. Our outcomes are similar to reported by Dutta et al. 20 Suguna Bai et al. and Fateme et al. 21, 26 reported a higher incidence of malformation in the babies born to mothers aged over 35 years.

Our results show a higher prevalence of malformations among multiparas (64%). Our result is consistent with this finding from other studies by Manal B et al. and Jawad S et al where the prevalence of malformations among multiparas was 76% each. 27, 28 On the contrary, a study conducted in Bangladesh by Fateme K showed more prevalence in primigravidas (63.33%). 26
Despite the high prevalence of congenital malformations, there is no awareness about maternal care during pregnancy, educational programs on congenital malformations and the consequences of increased maternal age and parity.

**Limitations:** In our study data have been taken retrospectively from hospital record and there has been no planned effort to discover malformations either at birth or in continued follow up during first year of life. Also only specific types of malformations which were obvious and apt to be noted on any routine examination at birth were included. Hence, the abnormalities detected late after birth were missed from data. Secondly, we could not include the abortions and stillborns, because often the abnormalities are not obvious or visible externally. In those cases, a pathological autopsy is warranted and in most of the cases, parental consent is not available for pathological autopsy.

**Conclusion**

This study has highlighted the prevalence and types of congenital anomalies seen in our locality. Regular antenatal visits and prenatal diagnosis are recommended for prevention, early intervention and even planned termination and appropriate treatment after birth when needed.

**References**