Pattern of Congenital Anomalies in the Newborn

Department of Pediatrics, Combined Military Hospital Okara

Abstract

Background: To determine the patterns of congenital anomalies in newborns.

Methods: In this descriptive study all newborns, whether alive or stillborn were screened for congenital anomalies, soon after birth. Detailed history was taken. Investigations were done when required to confirm certain defects.

Results: Out of total 50 patients 54% were males, with a male to female ratio of 1.2:1. Most of the patients were term babies (58%). Thirty eight babies (76%) were alive whereas 12 (24%) were stillborn. Antenatal diagnosis was done in 11 (22%) cases. Nervous system was the commonest system involved with 20 cases having some nervous system malformation followed by musculoskeletal system with 14 cases. Neural tube defect was the commonest. Three cases of congenital heart disease were seen with two cases each of Down's syndrome and facial malformation.

Conclusion: Neural tube defect, as the commonest congenital defect, emphasizes the importance of vitamin supplementation, especially folic acid, during pregnancy as a preventive measure.

Key Words: Congenital anomalies, Birth defects, Neural tube defects.

Introduction

Birth defects are defined as abnormalities of structure, function, or metabolism that are present at birth and result in physical, mental disability or mortality. These defects may be isolated or multiple and are due to multiple etiological factors. With the control of infectious diseases and malnutrition, congenital anomalies are now making a proportionally greater contribution to ill health and are currently a leading cause of infant mortality in developed countries. The exact prevalence of congenital anomalies varies in different areas and among different populations. In Pakistan, about 6-9% perinatal deaths are attributed to congenital malformations. Congenital anomalies can manifest as neural tube defects, congenital heart diseases, musculoskeletal defects, gastrointestinal defects, genitourinary defects, face defects and ear anomalies etc. Long-term survival of children with congenital anomalies is significantly lower than that for normal children.

Various risk factors have been identified as contributing to these defects which include genetic factors, maternal age, drug intake, teratogen, radiation exposure, maternal illness/infection, smoking, consanguinity and alcohol consumption. Some of these risk factors such as maternal drug intake, smoking, teratogen exposure, maternal infection, maternal diabetes, radiation exposure and alcohol consumption are avoidable. Neural tube defects are of particular significance as they are amenable to prevention with the use of folic acid during the peri-conception period. In many countries, flour and other food fortification with folic acid has been started which has significantly reduced the frequency of neural tube defects.

Antenatal screening such as maternal serum markers, chorionic villous sampling, amniocentesis, cordocentesis and ultrasound are important in early detection of many of the congenital anomalies. Early detection of anomalies can be helpful in deciding about termination of pregnancy or any therapeutic intervention.

Patients and Methods

This descriptive study was conducted over a period of six months at neonatal unit of Department of Paediatrics at Combined Military Hospital, Lahore in collaboration with obstetrical unit from January to June 2008. All newborns, whether alive or stillborn (excluding aborted fetuses), delivered in Combined Military Hospital were screened for congenital anomalies. Those who were having a congenital anomaly were selected for the study by nonprobability convenience sampling. Detailed history was taken with emphasis on duration of gestation, antenatal detection of anomaly, maternal address and maternal age. Detailed general physical examination of newborn was carried out. Weight and length were recorded. Examination of neurological, cardiovascular, respiratory, abdominal and musculoskeletal system was done. Investigations were done where required to confirm certain defects (echocardiogram in suspected cases of congenital anomalies).
heart disease and X-rays in suspected cases of gut atresias /diaphragmatic hernias). Information of each selected newborn and result of any investigation carried out were recorded.

**Results**

Out of total 50 patients with congenital anomalies 27 (54%) were males and 23 (46%) were females. Male to female ratio was 1.2:1. Most of the patients were term babies (58%). Forty percent were preterm babies and 2% were post-term. Sixteen percent of babies had weight between 0.5-1.5 kg, 44% had weight between 1.6-2.5 kg and 40% had weights between 2.6-3.5 kg. Mean weight of the babies was 2.35±0.74 kg (p value <0.04). Thirty two percent mothers were aged between 19-25, 58% between 26-30 and 10% between 31-36. Mean maternal age was 27.38±4.18 years.

Thirty eight babies (76%) were alive whereas 12 (24%) were stillborn. Antenatal diagnosis was done in 11 (22%) cases. Nervous system was the commonest system involved with 20 cases having some nervous system malformation followed by musculoskeletal system (Table 1).

<table>
<thead>
<tr>
<th>System</th>
<th>Anomalies</th>
<th>No(%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Central nervous system (40%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hydrocephalus</td>
<td></td>
<td>8(16)</td>
</tr>
<tr>
<td>Meningomyelocele</td>
<td></td>
<td>2(4)</td>
</tr>
<tr>
<td>Microcephaly</td>
<td></td>
<td>3(6)</td>
</tr>
<tr>
<td>Anencephaly</td>
<td></td>
<td>4(8)</td>
</tr>
<tr>
<td>Encephalocele</td>
<td></td>
<td>3(6)</td>
</tr>
<tr>
<td>Musculoskeletal (40%)</td>
<td>Syndactyly</td>
<td>3(6)</td>
</tr>
<tr>
<td>Polyactyly</td>
<td></td>
<td>4(8)</td>
</tr>
<tr>
<td>Talipes equino varus</td>
<td></td>
<td>5(10)</td>
</tr>
<tr>
<td>Developmental dysplasia of hip</td>
<td></td>
<td>1(2)</td>
</tr>
<tr>
<td>Arthrogryposis multiplex</td>
<td></td>
<td>1(2)</td>
</tr>
<tr>
<td>Genitourinary (18%)</td>
<td>Ambiguous genitalia</td>
<td>3(6)</td>
</tr>
<tr>
<td>Undescended testes</td>
<td></td>
<td>3(6)</td>
</tr>
<tr>
<td>Hypospadias</td>
<td></td>
<td>3(6)</td>
</tr>
<tr>
<td>Facial (4%)</td>
<td>Cleft lip/palate</td>
<td>2(4)</td>
</tr>
<tr>
<td>Cardiovascular (6%)</td>
<td>Cyanotic heart disease</td>
<td>1(2)</td>
</tr>
<tr>
<td>Syndrome (4%)</td>
<td>Down Syndrome</td>
<td>2(4)</td>
</tr>
</tbody>
</table>

**Discussion**

Major congenital anomalies occur in approximately 2-3% of births with a variable frequency in different populations. In our study, the rate of congenital anomalies was higher among males than females but the difference was not statistically significant (p=0.65). This finding was in contrast with the findings of the study carried out in Gorgan, northern Iran, where the rate was significantly higher among male newborns. Majority of the mothers, in present study, belonged to age group 19 to 30 years and none was above 36 years, which is in contrast to some other studies where 32% of mothers were aged 35 years or above. In our study 38 cases (76%) were alive and 12 cases were stillborn [24%], as compared to a study in Iran where 61% were alive. Most of the babies were term (58%) and 40% were preterm showing an increased tendency of anomalies in term babies in contrast to other reports showing increased tendency of anomalies in preterms. In our study 60% cases had weight below 2.5 kg which is in accordance with other studies showing increased incidence of low birth weight in babies born with congenital malformations. Antenatal diagnosis was done in 11 cases showing a low incidence of prenatal diagnosis, corresponding to a study done in Nigeria in which antenatal diagnosis was done only in 10 % of the patients.

Nervous system defects especially neural tube defects were the commonest anomaly found in this study, (20 cases or 40%), which is comparable to both local and international studies. Musculoskeletal anomalies were encountered in 14 cases (28%), becoming the second most common anomalies which is comparable to another study in Karachi with talipes equino varus being the commonest skeletal anomaly. Rate of cardiac malformation in this study was very low (3 cases), which is in contrast to another study giving a frequency of 4/1000 births. This may be due to under diagnosis because of lack of availability of sophisticated diagnostic technique, lack of routine autopies and neonatal follow-up loss.

Prevention of congenital anomalies can be primary and secondary. Primary prevention includes folic acid and multivitamin supplement, maternal disease prevention/treatment and preconception vaccination especially against chicken pox and rubella. Secondary prevention aims at early detection of congenital anomalies and advising early termination of affected pregnancies with lethal anomalies. These measures are especially useful in neural tube defects whose incidence are significantly decreased in developed countries as a result of preventive measures but are still very high in our study as well as most studies in our region. In our region, the magnitude of this problem can be attributed to several factors including the higher rate of traditional
consanguineous marriages, advanced maternal age, lack of educated mothers with poor knowledge regarding antenatal checkups, difficult access to health care centres and larger family size.\textsuperscript{9, 21}

**Conclusion**

Maternal education, preconception vaccination, vitamin supplementations, maternal disease prevention/treatment, avoidance of unnecessary drug/teratogen exposures, routine antenatal screening and offering early termination in affected pregnancies are measures by which the incidence of different congenital anomalies can be reduced.

**References**