Congenital anomalies in North Western Indian population – a fetal autopsy study

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SUMMARY

Better knowledge of unexpected fetal loss is the promise for better parental counseling and for prevention of recurrences. Fetal autopsy can provide a clue to ascertain cause of death in these cases. Variations in the incidence can be attributed to multiple factors. The present study was carried out to help us to develop a database concerning number of autopsies, incidence and types of congenital malformations (CMF) in the North-Western Indian population. The period of study was from January 2010 to November 2011.

Autopsy was carried out on 150 fetuses following guidelines provided by a fetal autopsy protocol. Prior to autopsy, prenatal investigations such as ultrasound and radiographs were procured; a brief maternal and family history was noted. Out of a total of 150 autopsies, 87(58%) were induced abortions and 63(42%) spontaneous abortions. In total, the incidence of CMF was 104(69%) of fetal autopsies. The types of CMF were classified as central nervous system defects (CNS) in 49 (33%), gastrointestinal tract (GIT) disorders in 48 (32%), musculoskeletal (MS) disorders in 31 (21%), genito-urinary (GU) in 25 (17%), and genetic disorders in 12 (8%). Multiple anomalies were present in 40 (27%) fetuses. Anencephaly (meroencephaly) turned out to be the most prevalent anomaly (29%). A few cases showed the occurrence of some uncommon syndromes. Major CMFs manifested very early in intra-uterine life, and could lead to termination of pregnancy (spontaneous or induced) in the 2nd trimester of gestation. Hence the presence of any CMF at the time of birth cannot provide the total percentage of CMF occurring in a given population. The above findings are discussed in the light of the available literature.

Key words: Fetal – Autopsy – Congenital – Major anomalies – Malformations – Fetus – Abortion

INTRODUCTION

Congenital Malformation is a physical, metabolic or anatomic defect which is apparent before birth, at birth, or detected during the first year of life. CMF can present itself in a single organ, system, or may involve multiple organs of the body. Early detection of major anomalies can be indicative of induced abortion to reduce the high morbidity of neonates due to congenital malformations (Siebert and Kapur, 2001).

The evaluation of CMF can be done by ultrasound, maternal serum analysis, etc., prenatally and by autopsy after the fetal death. Fetal autopsies, if done, can provide a clue to ascertain cause of death in these cases. Etiological diagnosis in unexplained fetal deaths is possible with detailed evaluation of fetus. Fetal autopsy is confirmative in 28.6-89%, diagnostic in 10-38%; it provided additional information in 3.9-24% cases; it changed the predicted probability in 18% cases

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(Mohan et al., 2004; Boyd et al., 2004; Sankar and Phadke, 2006; Sankar, 2011). In addition, the data pertaining to demography, socio-economic status, and maternal health is helpful to pinpoint the factors behind the occurrence of fetal loss.

Various studies (Choudhury et al., 1989; Swain et al., 1994; Malla, 2007; Saini and Kumar, 2009; Shamim et al., 2010) have described the incidence of congenital malformations in neonates (still and live born). The studies (Pahi et al., 1998) on the incidence of CMFs in fetuses are less. However there is a marked difference in the percentage of different CMFs in newborn vs. fetuses. Fetuses afflicted with major malformations result in spontaneous or induced abortions. Therefore a large number of malformations do not get counted in studies done at birth.

This study is an attempt to find the incidences of congenital anomalies out of total fetal autopsies in a given time. Further the classification of anomalies may help to identify the root cause of a specific disorder. It will also help us to develop a database containing number of fetal autopsies, incidence and types of CMF, and other related information in North-Western Indian population.

MATERIALS AND METHOD

The present study included a profile of 150 fetal autopsies being routinely done in this institute from January 2010 till Nov, 2011. Prior to the autopsy, the prenatal investigations like ultrasound, radiographs carried out by the respective departments were procured. Consent for the autopsy was taken from the parents/relatives on a Performa prepared in accordance with guidelines provided by the ethics committee which conforms to the provisions of the Declaration of Helsinki in 1995. A brief maternal and family history regarding any history of disease, intake of drugs, socio-economic status, etc., was noted down. The information regarding the prenatal genetic screening, if done, was noted and correlated with the presence of malformation.

Each fetus was sent for radiographic examination to note any skeletal abnormality. The various morphometric parameters including body weight, CRL (crown rump length), and CHL (crown heel length), head, thorax and abdominal circumferences, etc., were measured. The placenta and umbilical cord were examined for any gross abnormality. The external appearance of the fetus was recorded.

The autopsies were performed as per guidelines provided by fetal autopsy protocol (Siebert and Kapur, 2001; WHO, 2007; Agarwal et al., 2008). A longitudinal incision followed by a horizontal incision was given to retract the skin flap. The thoracic and abdominal cavities were opened and any deviation from the normal anatomy was photographed and noted. In relevant cases, tissues from various organs were sent for histological examination.

All the available information was collated to ascertain the cause of death. In this retrospective study, data from 150 fetal autopsies were computed to note the incidence of spontaneous abortions, intrauterine deaths, incidence and type of congenital malformations, and presence of syndromes. Graphs were plotted to correlate the number of cases with gestational age, socioeconomic status, maternal age, and gravidity of mother (Figs. 1-4).

RESULTS

Out of total 150 fetal autopsies, 87 (58%) specimens were from induced abortions, and 63 (42%) from spontaneous abortions and intrauterine deaths. In total, the incidence of congenital malformations was 104 (69%, Table 1). Out of 150 fetuses, 74 were males and 76 female fetuses; out of which the incidence of CMF in males and females was 75% and 83% respectively.

The congenital malformations were grouped according to the system affected, using World Health Organization classification (2007) of congenital malformation (Table 2). In 32.6% fetuses, defects in the central nervous system (CNS) were most prevalent. Gastrointestinal tract (GIT) anomalies were found in another 32%, followed by musculoskeletal (MS) anomalies in 21%, and genitourinary (GU) system in 17% specimens each; genetic disorders were the main cause in 8% of fetal deaths. Multiple anomalies manifested in 27% fetuses, giving rise to various syndromes.

When the gestational age was taken into account, the presentation and subsequent fetal loss

Table 1. Profile of 150 fetal autopsy cases. CMF, congenital malformations

<table>
<thead>
<tr>
<th></th>
<th>Total</th>
<th>Normal</th>
<th>Macerated</th>
<th>CMF*</th>
</tr>
</thead>
<tbody>
<tr>
<td>Spontaneous/missed abortions</td>
<td>63(42%)</td>
<td>19 (30%)</td>
<td>8 (13%)</td>
<td>36 (57%)</td>
</tr>
<tr>
<td>Induced abortions</td>
<td>87(58%)</td>
<td>11 (13%)</td>
<td>8 (9%)</td>
<td>68 (78%)</td>
</tr>
<tr>
<td>Total</td>
<td>150</td>
<td>30 (20%)</td>
<td>16 (11%)</td>
<td>104 (69%)</td>
</tr>
</tbody>
</table>

*congenital malformations
Fig. 1. Correlation between number of cases and gestational age. The number of aborted fetuses was maximum in 18-20 weeks, followed by 20-25 weeks of gestational age.

Fig. 2. Correlation of occurrence of congenital anomalies with socio economic status of parents. The upper-lower income group showed more cases of CMF.

Fig. 3. Incidence of CMF with maternal age. 60% fetuses belonged to the 20-25 year of maternal age group.

Fig. 4. No significant correlation was observed between congenital malformations compared to primi- or multigravida females.

Table 2. Systemic classification of congenital anomalies in autopsy cases: CNS: Central Nervous System; CVS: Cardiovascular system

<table>
<thead>
<tr>
<th>System</th>
<th>Spontaneous/missed abortions</th>
<th>Induced abortions</th>
<th>Total (% of 150)</th>
</tr>
</thead>
<tbody>
<tr>
<td>CNS*</td>
<td>17</td>
<td>32</td>
<td>49 (33%)</td>
</tr>
<tr>
<td>Digestive system</td>
<td>11</td>
<td>37</td>
<td>48 (32%)</td>
</tr>
<tr>
<td>Musculoskeletal</td>
<td>12</td>
<td>19</td>
<td>31 (21%)</td>
</tr>
<tr>
<td>Genito-urinary</td>
<td>9</td>
<td>16</td>
<td>25 (17%)</td>
</tr>
<tr>
<td>CVS &amp; Respiratory</td>
<td>5</td>
<td>7</td>
<td>12 (8%)</td>
</tr>
<tr>
<td>Genetic disorders</td>
<td>2</td>
<td>10</td>
<td>12 (8%)</td>
</tr>
<tr>
<td>Single Umbilical artery</td>
<td>-</td>
<td>5</td>
<td>5 (3%)</td>
</tr>
<tr>
<td>Multiple anomalies</td>
<td>15</td>
<td>25</td>
<td>40 (27%)</td>
</tr>
</tbody>
</table>

*Central Nervous System; **Cardiovascular system

Table 3. Incidence of types of CMF in different gestational age groups

<table>
<thead>
<tr>
<th>Gestational age</th>
<th>Total no. of CMF</th>
<th>CNS</th>
<th>MS</th>
<th>GU</th>
<th>GIT</th>
<th>CVS/RESP</th>
<th>GENETIC</th>
<th>Ass. A</th>
</tr>
</thead>
<tbody>
<tr>
<td>11-15 weeks</td>
<td>7 (5%)</td>
<td>2</td>
<td>-</td>
<td>3</td>
<td>2</td>
<td>2</td>
<td>2</td>
<td>4</td>
</tr>
<tr>
<td>&gt;15-20 weeks</td>
<td>62 (41%)</td>
<td>25</td>
<td>17</td>
<td>12</td>
<td>22</td>
<td>5</td>
<td>4</td>
<td>18</td>
</tr>
<tr>
<td>&gt;20-25 weeks</td>
<td>49 (33%)</td>
<td>16</td>
<td>10</td>
<td>6</td>
<td>14</td>
<td>3</td>
<td>3</td>
<td>10</td>
</tr>
<tr>
<td>&gt;25-30 weeks</td>
<td>16 (11%)</td>
<td>3</td>
<td>3</td>
<td>3</td>
<td>8</td>
<td>2</td>
<td>2</td>
<td>5</td>
</tr>
<tr>
<td>&gt;30-35 weeks</td>
<td>4 (3%)</td>
<td>3</td>
<td>1</td>
<td>1</td>
<td>2</td>
<td>2</td>
<td>1</td>
<td>3</td>
</tr>
</tbody>
</table>

CMF: Congenital Malformations; CNS: Central Nervous System; MS: Musculoskeletal; GU: Genitourinary; GIT: Gastro-Intestinal; CVS: Cardio Vascular System; Resp.: Respiratory system; Ass. A: Associated Anomalies
Fig. 5. A 24+ week female fetus with meningocele.

Fig. 6. A 14 week female fetus with complete rachischisis.

Fig. 7. Bilateral polycystic kidneys in a 28 week male fetus.

Fig. 8. Enlarged cystic urinary bladder with dilated ureters.

Table 4. Classification of defects in the Central Nervous system: VC, vertebral column

<table>
<thead>
<tr>
<th>Condition</th>
<th>Count (Percentage)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anencephaly</td>
<td>14 (29%)</td>
</tr>
<tr>
<td>Rachischisis (Spina bifida)</td>
<td>11 (22%)</td>
</tr>
<tr>
<td>Meningocele</td>
<td>7 (14%)</td>
</tr>
<tr>
<td>Meningomyelocele</td>
<td>10 (20%)</td>
</tr>
<tr>
<td>Encephalocele</td>
<td>9 (18%)</td>
</tr>
<tr>
<td>Hydrocephalous</td>
<td>10 (20%)</td>
</tr>
<tr>
<td>Abnormal flexion of VC*</td>
<td>9 (18%)</td>
</tr>
<tr>
<td>Total</td>
<td>49 (33%)</td>
</tr>
</tbody>
</table>

*vertebral column

Table 5. Classification of defects in the Genitourinary system

<table>
<thead>
<tr>
<th>Condition</th>
<th>Count (Percentage)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Agenesis</td>
<td>8 (32%)</td>
</tr>
<tr>
<td>Congenital polycystic kidneys</td>
<td>6 (24%)</td>
</tr>
<tr>
<td>Horse-shoe kidneys</td>
<td>3 (12%)</td>
</tr>
<tr>
<td>Undifferentiated gonads</td>
<td>2 (8%)</td>
</tr>
<tr>
<td>Hypospadias</td>
<td>1 (4%)</td>
</tr>
<tr>
<td>Oligohydramnios</td>
<td>4 (16%)</td>
</tr>
<tr>
<td>Dilated pelvis/ureter</td>
<td>2 (8%)</td>
</tr>
<tr>
<td>Absent/Cystic urinary bladder</td>
<td>1 (4%)</td>
</tr>
<tr>
<td>Total</td>
<td>25 (17%)</td>
</tr>
</tbody>
</table>
(either by spontaneous or induced abortions) occurred maximum (41%) in >15-20 wks, followed by 33% in >20-25 wks. The fetal loss was 11% in >25-30 wks, and only 5% in 11-15 wks (Table 3). Out of this total loss, CNS defects were the main cause in all the age groups followed by almost equal proportion of musculoskeletal, genito-urinary and GIT disorders (Table 3; Fig. 1).

Anencephaly (Mero-encephaly) turned out to be the most prevalent anomaly in CNS (28.5%); it was often associated with Meningocele (Fig. 5) or Meningomyelocele. Incidence of Rachischisis (spina bifida, Fig. 6) was 22.4% in cases with CNS anomalies. Hydrocephalus was observed in 20.4% cases and in 18% fetuses there was an abnormal flexion of vertebral column (kyphosis, scoliosis, etc.) (Table 4).

Amongst the defects in genito-urinary system, a congenital polycystic kidney (Fig. 7) was found in 24% cases; Agenesis of kidneys/ suprarenal/ gonad was observed in 32% cases. The kidney was horse-shoe shaped in 3 specimens; there was dilated pelvis/ureter in 2 specimens, and in other 3 cases there was abnormal opening of ureter. The urinary bladder had a large cyst in one fetus (Fig. 8). Two specimens presented with undifferentiated gonads (Table 5).

Exomphalos (omphalocele) was found in 12 fetuses (25%) in the GIT defects (Fig. 9). Absence of a digestive organ was present in 7 specimens, whereas there was non/mal-rotation of gut in 7 cases. Other anomalies of GIT included hepato/splenomegaly, etc. (Table 5).

Musculoskeletal anomalies included diaphragmatic hernia in 16%, joint defects in 29%, club foot in 16%, and cleft lip/palate (Fig. 10) in another 16% fetuses. Facial features were dysmorphic in 4 fetuses (Table 7).

Out of total 87 induced abortions, 12 (8%) specimens were triple-test positive when a ge-

**Table 6. Classification of defects in digestive system**

<table>
<thead>
<tr>
<th>Digestive System</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Omphalocele</td>
<td>12 (25%)</td>
</tr>
<tr>
<td>Non/malformation of gut</td>
<td>7 (14.6%)</td>
</tr>
<tr>
<td>Absence (liver/spleen/colon)</td>
<td>7 (14.6%)</td>
</tr>
<tr>
<td>Hepato/Splenomegaly</td>
<td>4 (8.3%)</td>
</tr>
<tr>
<td>Ascites</td>
<td>16 (33%)</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>48 (32%)</strong></td>
</tr>
</tbody>
</table>

**Table 7. Classification of defects in the musculoskeletal system**

<table>
<thead>
<tr>
<th>Diaphragmatic hernia</th>
<th>5 (16%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Club Foot</td>
<td>5 (16%)</td>
</tr>
<tr>
<td>Joint defects</td>
<td>9 (29%)</td>
</tr>
<tr>
<td>Cleft lip / palate</td>
<td>5 (16%)</td>
</tr>
<tr>
<td>Kyphoscoliosis</td>
<td>6 (19%)</td>
</tr>
<tr>
<td>Dysmorphic face</td>
<td>4 (13%)</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>31 (21%)</strong></td>
</tr>
</tbody>
</table>

**Fig. 9.** A case of omphalocele with abdominal visceral herniation enclosed in a transparent amniotic sac.

**Fig. 10.** A 28-week female fetus presenting complete cleft lip associated with complete cleft palate.
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Genetic analysis was done prenatally. The morphological features of 3 cases resembled those of Down’s syndrome including the presence of simian palmar crease, etc., although these features could not be conclusive of presence of Down’s syndrome. Three fetuses had features similar to Arnold Chiari syndrome, including the small posterior cranial fossa, rocker bottom feet, etc. Apart from these, we could find one case each of split notochord syndrome (SNS) (Fig. 11), hand foot genital syndrome (HFGS), Edward’s syndrome, aplasia cutis congenita, and cloacal dysgenesis sequence (Fig. 12). Our limitation with these cases was the lack of postnatal genetic analysis, which could have corroborated the present autopsy findings.

An attempt was made to correlate socioeconomic status and the occurrence of CMFs (Fig. 2). The socioeconomic status was calculated according to Kuppuswamy scale (Kumar et al., 2007), which is based on three variables — education, occupation and income. On this basis, population was divided into upper, upper middle, middle, upper lower, and lower classes. The present study included 54% cases belonging to middle class; 37% from lower class and only 10% from upper class (Fig. 2).

Sexual preponderance was not significant statistically, as 83% female fetuses had CMF as compared to 75% male fetuses. The incidence of CMF was correlated with maternal age (Fig. 3) and status of gravida (Fig. 4). It was observed that 60% of the fetuses belonged to the mothers who were between 20-25 years of age. Also, almost 50% of the mothers were primigravida, followed by 2nd gravida in 21%. Multi-gravida mothers (G4-G8) had only 15% of the congenitally malformed fetuses.

At the time of taking consent, any history of intake of drugs was enquired. Most of the parents/parent replied in negative. It was also admitted that the mother was not taking any kind of nutritional supplements usually given during pregnancy.

DISCUSSION

CMFs are emerging as important component in the perinatal mortality and morbidity with considerable repercussion on the families’ affected. Early diagnosis of life threatening CMF can pave the way for surgical correction or palliation of these infants.

Studies are available (Mohan et al., 2004; Boyd et al., 2004; Sankar and Phadke, 2006) where the presence of CMFs, prenatally diagnosed, are confirmed by fetal autopsy. Autopsies are performed to identify a cause of death, to produce epidemiologic data to further the understanding of the patient’s course and disease. Detailed autopsy, which includes radiological and histological examination, can be a useful tool in the diagnoses. It can also verify the ultrasound findings in the prenatal period, and can corroborate their results. In a teaching hospital, this kind of feedback is essential to maintain good standards.

The incidence of congenital anomalies in the total fetal autopsies was found to be 69% in the present observations. In another study (Puri et al., 2009) done in North India, the incidence of fetal anomalies was 63%, which is similar to the present study. Mohan et al. (2004) described the incidence as 38.7% out of 62 perinatal autopsies.

**Fig. 11.** A rare case of split notochord syndrome in a 24 week female fetus where intestinal loops were seen herniating from the posterior abdominal wall.

**Fig. 12.** Imperforate anus with associated gut anomalies in a 16 week male fetus.
from the same region, which is lesser than the
two abovementioned studies.

Most of the available literature pertains to con-
genital anomalies found at the time of birth (still
or live) (Malla, 2007; Swain et al., 1994; Shamim
et al., 2010; Choudhury et al., 1989). Table 6
shows the systemic incidence of various CMFs in
these studies. Many authors (Grover, 2000;
Mohan et al., 2004; Malla, 2007; Saini and
Kumar, 2009; Sankar, 2011) had stated the de-
fects in central nervous system as the highest
anomaly; the findings are similar to those of the
present study. Some studies (Chinara and Singh,
1982; Datta and Chaturvedi, 2000; Boyd et al.,
2004; Malla, 2007; Karbasi et al., 2009) men-
tioned the musculoskeletal as the most affected
system, whereas a few others (Shamim et al.,
2010; Dutta et al., 2010) reported GIT anomalies
to be the most frequent.

In the present study, the fetal autopsies showed
an incidence of NTDs higher than other anoma-
lies. This observation is in agreement with the
other two fetal studies from the same region
(Mohan et al., 2004; Sankar and Phadke, 2006).
The apparent cause could be that most of the
fetuses afflicted with neural tube disorders were
aborted very early in the gestational period. How-
ever, more incidences of NTD’s could be due to
lower socio economic status and lack of social
awareness. In US, Williams et al. (2005) found
the prevalence of spina bifida and anencephaly
highest among Hispanic births, followed by the
non-Hispanic whites, and the lowest among non-
Hispanic black births. To reduce these NTD’S
among these racial groups, folic acid fortification
programme was launched in US.

Favorito et al. (2004) noted the incidence of UG
(urogenital) anomalies in human male fetuses in
the age group of 10-36 wks of IU life. They found
the incidence of total UG anomalies as 4.2%, out
of which renal agenesis was observed in 1.2%.
They stated that UG anomalies in human male
fetuses are rare. Similar views were expressed by
Grover (2000), as they found the incidence of UG
anomalies in only 3.8% cases. Some other au-
thors (Malla, 2007; Sharma et al., 2009; Saini and
Kumar, 2009; Shamim et al., 2010; Dutta et al.,
2010) noted the incidence as 25.8%, 24.8%,
24.5%, 17.3%, and 26.2%, respectively.

Sanghvi et al. (1998) diagnosed congenital re-
nal malformations in 0.2% of fetuses by antenatal
ultrasound, out of which oligohydramnios was
noted in 31%. The other anomalies found were
dilated urinary system, polycystic kidneys, and
renal agenesis. The incidence of UG anomalies in
the present study was 16.6%. Renal agenesis
was present in 32%, polycystic kidneys in 24%.
Oligohydramnios was associated in 16% renal
anomalies. The genital defects were rare; a few
cases with defined syndromes had hypospadias
or undifferentiated gonads. In comparison to the
above mentioned authors, the absence of kidney/
kidneys in the present series is on a higher side.
This could be explained on the basis that many of

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**Table 8. Comparison of incidence of CMF with the available literature (%)**

<table>
<thead>
<tr>
<th>S.no</th>
<th>Authors</th>
<th>Autopsy</th>
<th>CNS</th>
<th>GIT</th>
<th>GU</th>
<th>MS</th>
<th>Genetic</th>
<th>CVS</th>
<th>Multiple anomalies</th>
<th>Sex preponderance</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.</td>
<td>Sankar et al. (2006)</td>
<td>Fetal</td>
<td>74.2</td>
<td>-</td>
<td>17.2</td>
<td>-</td>
<td>-</td>
<td>4.9</td>
<td>8</td>
<td>No sex diff.</td>
</tr>
<tr>
<td>2.</td>
<td>Malla (2007)</td>
<td>At birth</td>
<td>40</td>
<td>17.3</td>
<td>17.3</td>
<td>18.6</td>
<td>1.3</td>
<td>-</td>
<td>-</td>
<td>More in females</td>
</tr>
<tr>
<td>3.</td>
<td>Swain et al. (2010)</td>
<td>At birth</td>
<td>39.5</td>
<td>10.4</td>
<td>10.4</td>
<td>14.5</td>
<td>4.2</td>
<td>8.3</td>
<td>18.8</td>
<td>No sex diff.</td>
</tr>
<tr>
<td>4.</td>
<td>Shamim et al. (2010)</td>
<td></td>
<td>14</td>
<td>44</td>
<td>24.5</td>
<td>1.7</td>
<td>-</td>
<td>7</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>5.</td>
<td>Grover (2000)</td>
<td>At birth</td>
<td>40</td>
<td>-</td>
<td>3.8</td>
<td>23.8</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>8.</td>
<td>Dutta et al. (2010)</td>
<td>Live births</td>
<td>5.6</td>
<td>26</td>
<td>25.8</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td>More in males</td>
</tr>
<tr>
<td>9.</td>
<td>Al-Jama (2001)</td>
<td>Live births</td>
<td>49</td>
<td>6</td>
<td>14</td>
<td>11.2</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>10.</td>
<td>Kim et al. (2010)</td>
<td>At birth</td>
<td>8.4</td>
<td>11</td>
<td>11</td>
<td>10</td>
<td>-</td>
<td>-</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>11.</td>
<td>Present Study</td>
<td>Fetal</td>
<td>33</td>
<td>32</td>
<td>17</td>
<td>21</td>
<td>8</td>
<td>8</td>
<td>27</td>
<td>No sex diff.</td>
</tr>
</tbody>
</table>

CNS: Central Nervous System; GIT: Gastrointestinal System; GU: Genitourinary; MS: Musculoskeletal; CVS: Cardiovascular System
these studies were done by antenatal ultrasound examination, where the absence of an organ could not be verified as compared to the confirmed autopsy findings.

GIT anomalies constituted 32% in the present observations, and mostly these were represented by exomphalos (omphalocoele), gastrohisis, hepato/splenomegaly, and imperforate anus. Some of the defects were associated with paraumbilical defect of the anterior abdominal wall; some gut anomalies were associated with neural tube defects, and a few cases were part of a syndrome. 54% of GIT defects also had ascites. The incidence of GIT anomalies as reported by various authors (Al-Jama, 2001; Malla, 2007; Saini and Kumar, 2009; Kim et al., 2000) was 6%, 10.4%, 17%, and 1.17/1000. The present data report a higher incidence of GIT anomalies. The agenesis of an organ (liver, spleen, and pancreas) was not very common, as corroborated by other studies as well.

Musculoskeletal deformities included the diaphragmatic hernia, club foot, cleft lip/palate and dysmorphic facial features. Diaphragmatic hernias usually occur on left side (16% in the present study), and the heart and lungs are compressed towards the right due to upward displacement of abdominal organs. The incidence was reported as 1 in 3000-5000 births (Gilbert and Deich, 2004). Various authors (Kim et al., 2000; Al-Jama, 2001; Malla 2007; Saini and Kumar, 2009) have reported the incidence of musculoskeletal anomalies as 10%, 11.2% 15%, and 19%, which is in accordance with present observations.

Racial variations, geographical location and socio-economic status all play an important role in determining the incidence of congenital anomalies in a given population, e.g. most of the studies from Middle East countries refer consanguinity as one of the leading cause of CMF. Various researchers (Kulkarni and Kurian, 1990; Tayebi et al., 2010) have established the fact that consanguinity has a deleterious effect on fetal growth, and it increases the risk of CMF and fetal Loss. Some authors have linked low socio-economic status with consanguineous marriages, and therefore increase in the percentage of CMF is the result of both these factors. The present study did not find even a single case of consanguineous marriage in north-west India. Therefore, the impact of consanguinity on the occurrence of CMF could not be debated.

According to GAPPs (Global alliance to prevent prematurity) 85% preterm births occur in Africa and Asia (Erickson, 1976). This fact can explain the reason of higher incidences of anomalies in the present study as compared to the studies done in other continents.

Fetal factors

In the present study the most vulnerable period of gestation was >15-20 wks followed by >20-25 wks. Although the involvement of many systems might have occurred earlier during the first trimester, USG examination had confirmed the malformations in the second trimester which was followed by Induced abortions. None of the available literature had considered the prevalence of CMF according to gestational age. This observation has been the basis of our hypothesis that calculations of incidence of CMF at the time of birth do not reflect the accurate data.

Few studies (Chinara and Singh, 1982; Malla, 2007) had observed higher incidence of anomalies in male fetuses as compared to females. Raghuramaiah et al (2010) stated that NTDs are thrice more common in females than in males. Contrary to that, no gender preponderance could be established by many authors (Swain et al., 1994; Karbasi et al., 2009; Taksande et al., 2010). Similarly, in the present study the number of male and female fetuses was almost similar, and sexual preponderance was statistically not significant.

Maternal factors

The maternal factors considered in the present study were the age of mother and the number of pregnancy. The incidences of CMF were higher in primigravida; concurrently the age of the mother was also in the age group of 20 to 25 years. A few authors (Taksande et al., 2010) have maintained the higher incidence of CMF in gravida 4 and maternal age also higher than 35 years. Since the present study did not include many cases above 35 yrs. of maternal age, the data is mostly inconclusive. However 47% cases were from primigravida as compared to 9% cases from gravida 4. This observation is consistent with earlier studies. However, various studies (Chinara and Singh, 1982; Datta and Chaturvedi, 2000) were of the opinion that frequency of malformed babies was not influenced either by maternal age or parity of the mother.

According to Chinara and Singh (1982), there was a slight increase in the incidence of malformed babies in higher income groups. However, their division into different socio-economic groups seems to be arbitrary. Socioeconomic inequalities increased the risk of cardiac defects, GIT disorders and multiple anomalies – i.e., anomalies of non-chromosomal origin (Vrijheid et al., 2000). However NTD and oral clefts are not affected by socioeconomic deprivation (Sanghvi et al., 1998). The present study found the incidence to be 54% in middle class, and 37% in lower socio economic class. In India the socio-economic status of the families can cause severe nutritional deficiencies leading to gross congenital anomalies. That is why CNS malformations, particularly the anencephaly (mero-encephaly) and rachischisis, are
the most prevalent anomalies in the present study.

**Conclusion**

Fetal autopsies performed in cases of induced/spontaneous abortions can provide a clue, and can therefore serve as a purposeful tool in counseling the family for future planning.

The incidence of CMF was 69% out of 150 cases of fetal autopsy observed in North-West Indian population in a period of almost two years. The most prevalent CMF was NTD’s (33%) in the form of anencephaly (mero-encephaly) and rachischisis. They were followed by anomalies in the digestive system in 32%, the genito-urinary in 25%, the musculoskeletal in 21%, and genetic in 8%. Twenty seven percent of fetuses had multiple CMF leading to various syndromes. There was no significant sexual preponderance for the occurrence of CMF. Almost 50% of CMF occurred in primigravida and mothers of less than 25 years of age.

It will be erroneous to calculate the incidence of CMF in stillborn /live born fetuses at the time of delivery, as most of the fetuses with major CMF get aborted in the 17-28th weeks of IU life.

It is well known that surveillance and monitoring of congenital conditions is important for identifying patterns of CMF. The emphasis of the present study would be to sensitize the antenatal health care workers to provide an effective antenatal screening even in the tertiary or rural healthcare hospitals, and to recognize social factors for the prevention of birth defects.

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