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Research Article

Surveillance of congenital malformations and their possible risk factors in a teaching hospital in Punjab

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ABSTRACT

Background: Congenital malformations (CMF) are third most common cause of perinatal mortality in India making their prediction and prevention highly eminent. The present study was designed to analyze the frequency, distribution and patterns of CMF among babies born in a teaching hospital in Punjab, along-with factors contributing towards their occurrence.
Methods: This was a prospective study in which the incidence of CMF was determined in 1554 consecutive deliveries conducted at Gian Sagar Medical College, Banur, from July 2010 - June 2011. Diagnosis of CMF was based on prenatal ultrasound or clinical evaluation of the newborn by the neonatologist. Various risk factors associated with occurrence of CMF were studied.
Results: The incidence of CMF was 4.44% (n=69). Out of the babies born with CMF, 49 (71.01%) were still born, and major defects were present in 68.11%. CNS and urogenital system were most commonly involved, while 8.6% babies had multiple anomalies. High age and parity, parental consanguinity, bad obstetrical history, inadvertent drug ingestion enhanced risk; with no effect of radiation exposure and antenatal infections.
Conclusions: Incidence of CMF in our population was 4.44%: frequently associated with increasing age, parity, consanguineous marriage, maternal disease and drug ingestion.

Keywords: CMF, Incidence, Risk factors

INTRODUCTION

CMF represent defects in morphogenesis during early fetal life. One of the most traumatic experiences both for the pregnant female, and for the obstetrician is to deliver a congenitally malformed fetus. Advances in medicine and advent of better nursing facilities have led to a gradual decline in the infectious diseases and malnutrition, thus moving CMF higher up as a cause of great importance in early infancy, accounting for the deaths of nearly 2 out of every 1000 infants (US statistics). There is considerable ethnic and geographical variation in the incidence of birth defects. Various population and hospital based studies from different parts of India show that 2.5% of new-borns have a birth defect, both minor and major, thus ranking it as 3rd most frequent cause of perinatal mortality in India.[1-4] CMF may be due to genetic, environmental or a combination of both these factors and unknown causes. However, the prediction and prevention of congenital anomalies has been the obstetrician's challenge for a long time. Since there does not seems to be any practical way out to prevent these problems, the only realistic approach seems to be one of early diagnosis. Although a great deal of literature is available from western countries on this subject, there is a paucity of reports from Indian populations. The present study was thus proposed to analyze the incidence, distribution and pattern of CMF encountered in the Gian Sagar Medical College and Hospital, Ramnagar, Banur, Patiala, a teaching hospital in Punjab, India, between July

2010 and June 2011, and to identify the possible risk factors.

METHODS

This was a prospective study in which the incidence of CMF was determined in 1554 consecutive deliveries conducted at the institute.The hospital is the main referral centre in Punjab, which caters for both government and private hospitals of the area. CMF in this study is defined as structural abnormality found at birth and during the first week of life. Major anomalies included all those defects causing serious structural, cosmetic and/or functional disability requiring surgical or medical treatment. Diagnosis of CMF was based on prenatal ultrasound or clinical evaluation of the newborn by the neonatologist. .The study was conducted after taking informed consent from the patient. The association of maternal age, parity, socio demographic details, obstetric history, complications in present pregnancy, and baby's sex, gestational age, birth weight, Apgar score along-with clinically obvious malformations were recorded on a predesigned performa. Cases were also analysed for the presence or absence of risk factors viz maternal age 20 or less and 35 or more at the expected date of delivery, history of previous birth of a child with congenital malformations, history of congenital malformations in the family, history of two or more spontaneous abortions, history of consanguinity, alcoholism and smoking and poor socioeconomic status, maternal disease during pregnancy, any chronic disease in mother requiring prolonged medication, oligohydramnios, polyhydramnios, intrauterine growth restriction, uncontrolled diabetes mellitus in the peri-conceptional period, contact with infections and teratogens e.g. rubella, CMV, etc. History of exposure to radiation/ chemicals, or any fever was also noted. The types of anomalies identified were then classified by the diagnostic standardization of congenital malformation from the International classification of diseases (ICD-10) codes. Karyotyping was not done because it is not available in our institute, and it being a costly investigation and most of our patients belonging to poor socioeconomic status, were not able to afford it. Socioeconomic status was calculated according to Kuppuswami's criteria. No autopsy examinations were performed due to lack of parental consent for the same. Data was analysed using SPSS 15, and incidences of total and system specific malformations were calculated and strength of association of different factors calculated by chi-square method and odds ratio. The level of significance was set at $p < 0.05$.

RESULTS

As described in the tables. Table 1 shows incidence of CMF in live-born & still-born, incidence was much higher in still-born babies.

Table 1: Total incidence of congenital malformation (CMF) during study period.

	Total (n=1554)	Babies with CMF (n=69)	CMF Percentage
Live born	1467	20	1.363
Still born	87	49	56.32

Table 2: Incidence of CMF according to system involved as per the International Classification of Disease (ICD10).

Malformation system	No. of CMF (percentage)
Central nervous system	**39 (52)**
Hydrocephaly	9 (12)
Meningomyelocele	10 (13.33)
Anencephaly	19 (25.33)
Microcephaly	1 (1.33)
Genitourinary system	**15 (20)**
Hypospadias	9 (12)
PUV	4 (5.33)
Undescended testicle	1 (1.33)
Ambiguous genitalia	0
Epispadias	1 (1.33)
Digestive system	**6 (8)**
Imperforate anus	1 (1.33)
Atresia of esophagus with TE fistula	2 (2.67)
High arched palate	0
Omphalocele	1 (1.33)
Gastroschisis	2 (2.67)
Musculoskeletal system	**7 (9.33)**
Polydactyly	2 (2.67)
Clubfoot	4 (5.33)
CDH	0
Syndactyly	1 (1.33)
Brachydactyly	0
Clubhand	0
Rizomelia	0
Chromosomal abnormality	**3 (4)**
Down's syndrome	3 (4)
Cleft lip and cleft palate	**3 (4)**
Cleft lip with cleft palate	1 (1.33)
Cleft lip	2 (2.67)
Cleft palate	0
Eye, ear, face and neck	**none**
Cardiovascular	**2 (2.67)**
Respiratory system Choanal atresia	**0**
Total	**75 (100)**

Table 3: Socio-demography of mother and infants in study.

		All mothers	Mothers with CMF babies	Incidence of CMF (%)
Mother				
Age	<20	105	9	8.57
	20-35	1371	29	2.12
	>35	78	31	39.74
Parity	1	378	6	1.58
	2	552	10	1.81
	3	432	20	4.63
	≥4	192	33	17.19
ANC*	Booked	691	22	3.18
	Unbooked	863	47	5.45
Residence	Rural	1293	66	5.10
	Urban	251	3	1.20
SE status (class)#	I	9	0	0
	II	522	5	0.96
	III	1023	64	6.26
Religion	Hindu	734	31	4.22
	Sikh	799	34	4.26
	Muslim	21	4	19.04
	Christian	0	0	0
	Others	0	0	0
Infant				
Sex	Male	786	37	4.71
	Female	768	32	4.17
Birth weight	<2.5kg	831	41	4.93
	≥2.5kg	723	28	3.87
POG**	<14weeks	121	11	9.09
	14-28 weeks	217	38	17.51
	28-36weeks	423	18	4.26
	≥37 weeks	793	2	0.25

*Antenatal care; #Socio economic Status; **POG=Period of gestation

Table 2 shows distribution of CMF according to the organ system involved. Central nervous system & genitor-urinary system the most commonly involved organs.

Main risk factors associated with CMF were consanguineous marriage, inadvertent drug use, substance abuse, DM and polyhydramnios.

Table 5 shows comparison of our data with various studies on prevalence of CMF.

Table 4: Risk factors/associated conditions.

	All Mothers	Mothers with CMF babies	Percentage of CMF (%)
Consanguineous marriage	9	6	66.67
Family history of CMF baby	4	1	25
Bad Obstetric history	15	3	20
Previous child with CMF	7	1	14.29
Inadvertent drug ingestion	5	3	60
Substance abuse	2	1	50
Radiation/chemical exposure	0	0	0
Contact with infections	7	0	0
Maternal disease in pregnancy	1304	14	1.07
Diabetes mellitus	18	3	16.67
Epilepsy	7	2	28.57
Heart disease	4	0	0
Renal disease	4	0	0
Fever in pregnancy	5	0	0
Anemia	1266	9	0.71
Obstetric complications			
Threatened Miscarriage	3	0	0
Hyperemesis gravidarum	2	0	0
Antepartum Hemorrhage	41	1	2.44
HDP	716	2	0.28
IUGR	91	3	3.30
Oligohydramnios	119	6	5.04
Polyhydramnios	11	4	36.36

DISCUSSION

Most children who are born with major CMF and survive infancy are affected physically, mentally or socially, or can be at increased risk of morbidity due to various health disorders. Thus primordial and primary prevention are vital to decrease incidence of CMF and the morbidity associated with it.

In our institute total number of CMF during the study period was 4.44%. This is significantly higher than the WHO survey of a large series of consecutive births across 24 centers of the world.[5] Also within India, our figures are comparable with those reported from Delhi, Wardha and Hyderabad, but higher than the incidence

reported from Lucknow, Allahabad, Varanasi and Calcutta.[6,7] This wide disparity may be attributed to- the increasing number of referral cases coming to the hospital, due to current ease of detection owing to the rapid improvements in prenatal diagnosis, inclusion of minor anomalies, geographical, racial and ethnic factors, different survey methods used, and other parameters. Moreover, hospital statistics are many a times insufficient and do not replicate the true incidence of birth defects in a community.

Table 5: Frequency of congenital malformation (CMF) in various studies.

Location/ reference	CMF rate per 1000 birth	CMF rate per 1000 live births
Yazd, Iran[12]	28.33	27.95
Tehran, Iran [13]	-	35
Tehran, Iran [14]	-	24.1
Arak, Iran[15]	-	10.4
Gorgan, Iran [16]	-	10.1
Oman [17]	24.6	-
Bahrain[18]	-	27
Arab Emarates[19]	7.92	7.89
Beirut, Lebanon[20]	16.5	-
Maharashtra , India [21]	12.8	10.8
Simla, India[22]	17.8	1.3
Punjab, India (present study)	44.4	1.36

Amongst the total 87 still born babies during the study period, 49 had CMF ,thus making the incidence much higher in still born (56.32%) as compared to the live born babies (1.36%), which is in concordance with earlier published reports across the world.[6-11]

Major birth defects were present in 47 (68.11%) cases and majority of babies 63 (91.30%) had isolated single malformations. Hence, our incidence of CMF involving multiple systems is lower than earlier reported studies. Table 2 shows the ICD-10 classification of the different types of CMF encountered and their frequency Central nervous system (CNS) defects were the most common conditions encountered in the series, accounting for around half (52%) of the birth defects recorded, followed by genitourinary malformations (20%) and musculoskeletal defects (9.33%). Cardiovascular anomalies, being present in 2.67% of the subjects, were the least common amongst all anomalies. Of the CNS defects, anencephaly was the most common (25.33%), followed by hydrocephalus (12%) and meningocele/meningomyelocele (13.33%), whereas microcephaly (1.33%) was the least common anomaly. Incidence of neural tube defects (NTD) have been reported to be 1-8% in different geographical areas and anencephaly is the most common type of NTD accounting for 50-65% of these cases in the literature, which was akin to our observation.[10] The male/female ratio for anencephaly is classically l/4 and in our study also, this ratio was found to be 1/3. Though research has suggested the lack of vitamins, especially folic acid, as a possible cause of NTDs, besides interaction of genetic and environmental factors and infections, at present only the protective effect of folic acid supplementation in the pre-conceptual period against NTD seems to be proven. Most obstetricians thus prescribe folic acid to women in pregnancy with a past history of NTDs. Hypospadias was the commonest genitourinary malformation observed while clubfoot accounted for more than half of the cases with musculoskeletal system involvement. Trachea-esophageal fistula was the commonest gastrointestinal system malformations (4%) found. Cleft lip with or without cleft palate, and Downs syndrome was identified in 3 babies each. Within India, high frequency of NTDs were reported from Delhi and Davangere[23-25] while a research observed gastrointestinal malformations as the commonest malformation.[26]

8.57% of the infants with anomalies were born to adolescent mothers and 39.74% were seen in elder mothers. The incidence of CMF was found to be 17.19% in women with parity ≥4, being significantly higher than anomalous babies with birth order of one or two. A possible explanation for higher birth order is that women at risk for poor pregnancy outcome, i.e. severe CMF, will keep trying until they have normal offspring, resulting in a high birth order. However, since high parity tends to be more prevalent in lower socioeconomic groups, the effect of this parameter on fetal morbidity could not be isolated from confounding influence of socio economic factors. Also, a higher incidence of CMF was observed in babies of unbooked mothers than those booked for antenatal care (5.45% vs 3.18%). 66 out of the total 69 mothers with malformed babies dwelled from rural areas of the region. The incidence of CMF was significantly higher in the lower strata (6.26%) and it decreased as the socioeconomic status of women improved. Also significantly higher number of anomalous newborns was born among Muslim couples (19.04%) than Hindu and Sikh (4.22% & 4.26% respectively) (Table 3).

The incidence of CMF was found to be apparently higher in male babies as compared to female babies, though the difference was not significant. However female preponderance (11/19) was observed in anencephalic babies, while PUV and hypospadias were only seen in male infants. Mishra et al and Mathur et al.[3] have also noted higher incidence among male babies while Swain et al. and Mohanty et al. have not observed any difference between the two sexes.[2] The birth weight distribution of the malformations revealed 41 babies with low birth weight and 28 babies with normal birth weight, which was a statistically significant observation. Also, CMF were seen with a higher frequency among preterm babies (30.8% vs. 0.25%) than their term counterparts, as quoted in literature before. This can be attributed to the lower growth potential of malformed babies, which may

result in abortion or preterm delivery. Also, they have higher chances of being still born or dying later in the neonatal period. Similar to previous researchers, a higher incidence of CMF was found among offsprings of consanguineous marriage (66.67%). One third of these were first cousins while remaining were more distant relatives. One case (25%) had a history of affected relatives of the same or different condition. Only fourteen women with malformed babies had pre-existing medical conditions. Of them, epilepsy (28.57%) followed by diabetes mellitus (16.67%) were the most predominant. A previous bad obstetrical history was noted as a high risk factor for increased chances of CMF, as among the 15 mothers with the same, 20 % experienced anomalous births. One woman with malformed baby had already a child with CMF, who was having downs syndrome. In the present study, history of drug intake could be elicited in only 3 (60%) mothers with CMF, out of which one was epileptic, while remaining two took some medicine from quacks before conception for having a male baby; however no specific association could be established as details of the drugs were not available. The existence of mutagens with high incidence of consanguinity may be the cause for the malformations seen in our study population. One woman gave history of smoking during peri conceptional period. No mother gave positive history of exposure to the radiation in the early pregnancy in the study.

Amongst the obstetric complications associated with mothers with malformed fetuses, one had APH due to placenta previa, six had oligohydramnios, out of which three had growth restriction associated with same, two had hypertensive disorders; and four women had polyhydramnios, out of which 50% cases had anencephaly. In those cases where no cause was found for the occurrence of CMF, those may be genetic or multifactorial etiologies. Five of the malformed infants died in the neonatal period, taking the total toll attributed to CMF to 54 during the period of study.

Thus, in times to come, CMF may emerge as an even more important perinatal problem contributing, sizeably, to perinatal morbidity and mortality. the advantage of discovering a CMF in the antenatal period enables us to terminate such pregnancies which may be expected to reduce the incidence of congenital anomalies at birth. With greater emphasis on 'small family norms' and population control it is necessary to identify CMF so that intervention programmes can be formulated.

In this study, we tried to determine the CMF incidence and identify their types in our population, risk factors associated with them in an effort to possibly reduce the incidence. Early detection and effective management of life threatening malformations will help in alleviating the sufferings and effectively controlling the associated morbidity and mortality. Premarital counseling is advised, especially in the presence of parental consanguinity and family history of a congenitally malformed child avoiding consanguineous marriages may help in bringing down the incidence drastically. Because of the high frequency of NTDs as revealed by our investigation, we recommend their proper prenatal diagnosis as per the Guideline. And provision of periconceptional vitamins and folic acid to all pregnant women. However, further research to evaluate the role of diet, cultural and other environmental factors in the pursuit of determining the cause of fetal malformations is essential. Besides larger multicentric studies are needed to determine the exact congenital anomaly distribution of our country. Widespread health education in the population will thus go a long way in awakening awareness to the preventable etiological factors of fetal malformations.

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