Congenital Malformation - A Biological Accident

(A 6 years 9 months study in a rural based tertiary care hospital)

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Abstract: Background: Birth of live congenitally abnormal child especially at term is a great mental and social trauma to parents, family and to the society. In developed countries it is one of the main causes of infant mortality. It accounts 8-15% of peri-natal deaths and 13-16% of neonatal deaths in India. Aims & Objectives : To find out the incidence of congenital anomalies in live and still births and to find out association of major and minor anomalies to maternal demographic profile and to various obstetrics and medical parameters.
Materials & Methods: This study was carried on rural based tertiary care referral hospital. All the deliveries occurred during 1st September 2004 to 30th June 2011 (81months) comprised 7053 births were enrolled. The new born were examined systematically by obstetrician and pediatrician. System wise distribution of anomalies and correlation with risk factors were analyzed. Results: Out of total 7053 deliveries, 6433 were live births and 620 were still births. Total number of congenital malformed babies were 71 (1.006%). 31 (43.66%) of the 71 malformed babies were still born and 27 (38.02%) were NND (neonatal death). It means out of 100 stillborn, 43 babies had structural malformation. Major malformation was present in 58 and minor anomalies were present in 13 fetus. Marginal Male predominance 40 (56.33 %) was noted. Central nervous system malformation was most common in live births followed by respiratory system and musculo skeletal system. 64% patients had not taken ANC (antenatal care) and in them 92% had major anomalies. 81.3% anomalies were detected by 2nd trimester USG (ultra sonography) and 18% anomalies were not detected by USG out of which 98% were minor anomalies. Conclusion: Congenital anomalies are a major cause of still births and infant mortality. Regular ANC, 2nd trimester USG for early detection and folic acid supplementation for prevention are the important factors to be considered to decrease the burden of congenital malformation. [Phadke et al NJIRM 2011; 2(5) : 94-97]

Key words: Congenital malformation, congenital anomalies

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Introduction: Birth of a live congenitally abnormal child especially at term, is a great mental & social trauma to parents & pauses a heavy financial burden on the family. According to the World Health Organization (WHO document of 1972), the term congenital malformation should be confined to structural defects at birth[1] . Congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India[2,3]. Even though it appears to be the tip of the ice berg, it is a great havoc to the parents. It also stretches the demand of human resources with wastage of financial resources for the country.

The leading causes of infant mortality and morbidity in developing countries are malnutrition, infection, where as in developed countries they are congenital malformation and cancer. The proportion of perinatal deaths due to congenital malformation is increasing as a result of decrease in mortality due to other causes owing to the improvement in perinatal and neonatal care .In coming years , this is going to be the leading cause of morbidity and mortality in centre providing good antenatal and neonatal care. Registry of birth defects will help in study the malformation profile in a geographical locality and under take etiologic studies.

The present study was carried out to find out the incidence of congenital anomalies in live and still births and to find out association of major and minor anomalies to maternal demographic profile and various obstetrics and medical parameters.

Materials and Methods: This study was conducted in department of Obstetric & Gynecology , Shri Krishna Hospital, Karamsad, a rural based tertiary care hospital, after approval from institutional

ethical committee. All the deliveries occurred during 1st September 2004 to 30thJune2011 (6 years 9 months) were included in this study. The study material comprised 7053 births (live & still births). All the newborns were looked for congenital malformation soon after birth and every day till patients were admitted. Relevant information regarding maternal age, parity, socio economic status, locality, religion, gestational age, birth weight and sex of fetus were documented. Detailed antenatal history like ANC care taken, maternal illness, ingestion of drug, exposure to radiation and complications during delivery were recorded. The antenatal ultrasonography findings were noted. A detail general and systemic examination was carried out by obstetrician and pediatrician at the time of birth to detect any malformation. Ultrasounds, 2D echocardiography, X-ray, ECG (electro cardiogram), were done in indicated cases. Autopsy was done on still birth and in neonatal deaths after getting consent from parents. Malformations were divided in major and minor. Major anomalies were those which has direct impact on mortality or morbidity, require medical or surgical intervention or have significant cosmetic concerned. The malformation were divided into central nervous system, muscular skeletal system, cardiovascular system, genitourinary and miscellaneous disorders. Statistical analysis was done.

Results: Total no. of patients delivered during study period was 7053, live births 6433 & still births were 620 . Numbers of babies with congenital malformation diagnosed at birth or within 1st week of life were 71 [1.006%]. Out of 71, still births were 31, live births 40 & NND (neonatal death) were 27. Major anomalies were present in 58 (81.69%) & minor anomalies were present in 13(18.3%) babies. There was male predominance in malformed babies i.e. 40 [56.33%] males to 29 [40.84 %] females. In 2 babies, sex could not be identified .There were ambiguous genitalia .Congenital malformations were seen more significantly in still birth [4.51%] as compared to live births [0.24 %]. It means prevalence of malformation was 1:25 for still birth against 1:416 for live birth in present study.

Table no. 1 shows demographic profile. Mothers in reproductive age 21-30 yr had maximum congenital malformed anomalies [69.01%].Mean age of patient was 28.5years .Sixty three [88.33] were Hindus & 8[11.26 %] were Muslims . Fifty seven (80.28 %) patients were from low socioeconomic class, 46(64.78 %) had not taken ANC (antenatal care) and maximum (42) major malformation baby was found in this group. Forty (70%) patients had USG (ultrasonography) done and 95% major anomalies were detected. Two babies had TOF (Tracheo esophageal fistula) which was missed in USG. Twelve patients were booked patient who were enrolled from 1st trimester and regular USG was done. Out of 12, in 9 congenital malformation was detected during 2nd trimester USG. Rest of 3 were minor malformation detected at the time of birth and otherwise healthy baby.

Table I: Demographic profile (n=71)

		Total no.	%
Age of mother (in years)	Up to 21	2	2.81
	21 to 30	49	69.01
	>30	20	28.16
Parity	Primigravida	19	26.76
	Multigravida	52	73.23
Socio economical status	Low	57	80.28
	Middle	14	19.71
	High	0	0
Gestation	20-28 weeks	20	28.16
	29-36	31	43.66
	>37 weeks	20	28.16
Birth weight	<1000 gms	12	16.90
	1000-1500gms	29	40.84
	1500-2000gms	14	19.71
	>2000gms	16	22.53

Analysis of maternal risk factors was done . Out of 71, co existence of mal presentation and liquor abnormality was found in 32 and 29 had major malformation. The second common risk factor co existing were ecclampsia and severe anaemia.

Association of low birth weight and preterm delivery with increased risk of congenital malformations was very well documented [4]. Our

findings can be compared with this study. 57% babies had birth weight of less than 1.5 kg and 71.82% had preterm delivery.

Table no. 2 represents the distribution of malformations according to the systems involved. Malformations involving the CNS system were maximum i.e.29 [40.84 %].

Table II : System wise distribution of malformation fetuses in our studies

Distribution of system involved	Present study
CNS	29 (40.84%)
Respiratory system	7 (9.85%)
CVS	3 (4.22%)
GIT	4 (5.63%)
Genitourinary	3 (4.22%)
Musculoskeletal	9 (12.67%)
Skin	0
Defect in spine	4 (5.63%0
Eye , ear	2 (2.81%)
Syndrome	6 (8.45%)
Single umbilical artery	2 (2.81%)
Miscellaneous	2 (2.81%)
Total	71

The next most commonly involved system is respiratory system- 7 patients [9.85 %].Out of 71, autopsy was done in only two malformed babies. One showed dolicocephalic head, low set ears , hypo plastic lung ,both polycystic kidney, large suprarenal gland and enlarged spleen. Other one had fetal ascites.

Table no. 3 shows the relation between antenatal care and time of congenital malformation. Out of 71 patient only 12 had taken regular ANC and congenital malformation was detected by USG in all .

Table III Relation between antenatal care & time of detection of anomaly .

ANC	2nd Trimester	3rd Trimester	At Birth
ANC not taken	4	19	23
ANC taken	9	3	1
Irregularly taken	0	8	4

Discussion: The overall incidence of congenital malformation in India ranges from 0.3% to 3.6%[5,6,7] It is higher in centre where autopsy is carried out as a routine[8]. Association of low birth weight with increases of malformation was well documented[4]. Our finding is in accordance. The incidence of congenital anomalies was significantly higher in preterm babies as compared to full term babies[9].

Figure 1 : Congenital fetal ascites with anterior abdominal wall defect with limb abnormalities

In our study – 51(71.82 %) were preterm baby. The incidence of congenital malformation has reported male predominates [4] which was proved in our study. We have 56.3% of male fetus. This study showed that malformed babies were more in low socio economic group. This may be related with malnutrition or micronutrient deficiency. Another cause is, these populations do not take ANC because of unawareness and poverty. So diagnosis becomes delayed.

Central nervous system was the mostly affected system in our study. Kalra et al.study[10] also reported that the CNS defects have the highest incidence .

Conclusions: Our study is a preliminary study to obtain first hand information about the magnitude of problem of congenital malformation in tertiary care referral hospital which is the tip of ice berg. The real incidence of malformation will be definitely more because all patients with malformations are not referred. The incidence of congenital malformation at birth in our study is 1.006%. Incidence of malformation in still birth is

19 times more than live birth. Major malformations were present in 81.69% & CNS abnormalities in 41%. We observed that 65% has not taken ANC & 72% of major malformation was present in this group. USG was not done in 70% patients & 95% major anomalies were detected in this group. Out of 71 patients only 12 had taken ANC from 1st Trimester.

Malformation was detected in 75% on early 2nd trimester & rest 25% were minor abnormalities. In our study group 2 patients who had cardiac malformation in fetuses in two consecutive pregnancies. So if patients come early, chromosomal study in 1st trimester, preconception and 1st trimester folic acid supplementation and other required investigation may be done for prevention or early diagnosis of congenital malformed baby.

We had 45% patients with liquor abnormalities in antenatal time & 90% of them had major malformation. Level 2 targeted USG is must when patient presents with persistent malpresrentation with liquor abnormalities. Now there are medical and surgical therapies (fetal therapy)in utero is available for some malformation detected in time before birth. So preconceptional counseling, early ANC and USG are the corner stone to prevent birth of congenital malformed baby which is a big social burden to parents and society.

The limitations of study : History of consanguinity, autopsy and karyotyping was not done due to lake of fund available and refusal by patients due to ignorance regarding importance of the same.

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