FETAL CONGENITAL ANOMALIES - AN ANALYSIS

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SUMMARY

Analysis of fetal congenital anomalies showed that the overall incidence was 1.5% in our hospital. Neural tube defects were the most common anomalies encountered. Prenatal diagnosis was possible in 51.5% while in another 12% anomalies were suspected on sonography. Almost 78% were major anomalies. Hence outcome for the babies has been poor. Only 25% have normal survival. Our general perinatal mortality rate for the same period was 78.8 per thousand. However congenital anomalies accounted for only 7.7% of all perinatal deaths. Maternal serum alpha feto protein screening and sonography, besides identifying anomalies, also help in detection of high risk pregnancies. This should help in decreasing the perinatal mortality.

INTRODUCTION

Birth of a malformed baby is an unfortunate event for any family and society. With advances in perinatal medicine and improved perinatal outcome in the West, the focus is now on prenatal diagnosis and fetal therapy because congenital anomaly is an important cause of perinatal mortality and morbidity today. However in India, abruptio placenta, low birth weight and eclampsia are still responsible for majority of the perinatal deaths. The aim of this study was to find out incidence of congenital anomalies, its outcome and its contribution to the perinatal mortality.

MATERIAL AND METHODS

Congenital anomalies were studied in all deliveries and second trimester abortions occuring between January 1992 and December 1993. Detailed antenatal history with stress on exposure to teratogens in the first trimester and family

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history were recorded. Relevant investigations to rule out diabetes and TORCH group of infections were done. All the malformed babies and abortuses were examined in detail. Postmortem of dead babies was possible only in a few as many were macerated and consent was not available in some. Other causes of perinatal mortality were also studied in the same period.

RESULTS AND OBSERVATIONS

The overall incidence of congenital anomalies was 1.5% in 2652 deliveries and 76 second trimester abortions. There were a total of 87 anomalies in 40 babies (Table I). Twenty one had isolated anomalies; 4 had multiple anomalies of the same system while in another 15 there were multiple anomalies involving different systems.

Although craniospinal and musculoskeletal defects occured with equal frequency, the latter were mainly minor defects like talipes, polydactyly and scoliosis. There were only 2 major skeletal dysplasias-one thanatophoric dysplasia and another short rib polydactyly syndrome. Both of these were suspected prenatally on ultrasound but the exact diagnosis regarding the type of dysplasia was made only after delivery. One abortus had phocomelia. Among the craniospinal defects there were 5 isolated hydrocephalus, 1 with absence of occipital bone and the rest (12) were neural tube defects. None of the babies with craniospinal defects survived. Major anomalies incompatible with life and requiring major surgery were seen in 31 (77.5%) of cases while 9 (22.5%) had minor anomalies.

As expected 63% were unbooked but gave history of having had antenatal checkup elsewhere. Only 3 had not had any check up. Seven cases (17.5%) had not had ultrasound antenatally. Among the 33 scanned, 17 (51.5%) were diagnosed and another 4 (12%) were suspected to have an anomaly. In 12 (36%) diagnosis was missed inspite of the scan. Among those missed significant ones were 2 cases of spina bifida and one each of diaphragmatic hernia, renal agenesis and cleft palate. Others were minor ones like talipes, polydactyly and ambiguous genitalia.

Among the 33 patients scanned pre-

Table I

Congenital Anomalies

Craniospinal defects	18	Abdominal wall defects	4
Musculoskeletal	18	GI tract	7
Face, eyes, ears & mouth	15	Renal system	4
Neck	2	Ext. genitalia	8
Heart	3	Trisomy 21	2
Pulmonary hypoplasia	2	Skin	4

natally, 23 were scanned in the third trimester as many of them reported only then. Atleast 4 major anomalies were diagnosed on routine scanning. Polyhydramnios and oligoamnios were seen in 25% of the women with anomalies. Almost 32% had preterm labour; breech presentation and IUGR were seen in 17% and 14.6% respectively. Single umbilical artery was seen in 2 cases. Threatened abortion in the 1st trimester was reported by 3 women. Fifty percent (20) of the babies were females while in 3, genitalia were ambiguous.

Cause for teratogenesis could not be determined in the majority. There was significant history of drug intake in only one patient who had gone to a general physician with continuous vomiting between 5th-7th week of gestation. She was given Tab Domperidon, Inj. Ranitidine and Tab Mebendazole. The abortus at 2nd trimester had phocomelia, omphalocele with right sided cleft lip and palate. Three women had married their first cousins. Seventeen (42.5%) were primigravidas. In the remaining, only 2 had recurrent anomalous babies. One was a 3rd gravida, who lost her first baby due to congenital heart disease and unilateral anophthalmia. Her second baby is alive with anophthalmic left eye and microphthalmia of the right eye which can only perceive light. In her third pregnancy, anophthalmia was diagnosed by USG at 22 weeks and pregnancy was terminated. Another woman had congenital ichthyosis with collodion membrane in two successive pregnancies. Eighty percent were between 20-30 years of age, only one woman was above 40 years of age and she had a trisomy 21 baby. All had singleton pregnancy and none were diabetic. Three had conceived after investigations for infertility and one of them was clomiphene induced pregnancy.

Late abortions, perinatal and infant deaths occured in 70% of congenital anomalies (Table II). Among the 12 survivors, 2 are mentally retarded; one with trisomy 21 and another with cerebral palsy and spastic diplegia. The baby with trisomy 21 has been abandoned by the parents.

Total general perinatal mortality during the study period was 78.8 per thousand. Table III shows that abruptio placenta was the leading cause of perinatal mortality while congenital malformation was responsible for only 7.7% of deaths. No cause was apparent in 23% of cases. Majority of these were unbooked cases who reported only after loss of fetal movements. Postmortem was not possible in macerated babies. There were

Table II

Outcome of Congenitally Abnormal Babies

Outcome	Number	Percentage
Late abortions	5	12.5
Terminations	3	7.5
Still births	9	22.5
Neonatal deaths	7	17.5
Infant deaths	4	10
Survivors	12	30
Total	40	100

Table III

Causes of Perinatal Deaths
n=209

	Number	Percentage
Abruptio placenta	54	25.8
Unexplained stillbirths	49	23.5
Prematurity	31	14.8
* NND (various causes)	23	11
Hypertensive complications	22	10.5
Congenital anomaly	16	7.7
** IUD (various (causes)	14	6.7
Total	209	100

* Neonatal deaths - births asphyxia, meconium aspiration, 2nd of twin & cot death.
** Intrauterine deaths - cord prolapse, IUGR, rupture uterus & placenta praevia.

8 caesareans in the anomaly group. In 4 patients, anomalies were suspected on scan but caesarean had to be done for other obstetric reasons. In the caesarean group there were 2 neonatal deaths and 2 infant deaths.

DISCUSSION

The incidence of congenital anomalies in the general population is said to be around 2% (MJ Whittle 1991). Our incidence of 1.5% is low, partly because postmortem has not been done in all unexplained stillbirths. Neural tube defects were the most common defects encountered. Definitive etiology for teratogenesis can not be determined in the majority of cases. Brent et al (1993) reported that in 20-25% fetal anomalies were genetic and in another 10% environmental causes were responsible.

The outcome for babies with anomalies is very poor. Among the 12 survivors only 12 can lead a meaningful life. Despite the poor outcome the contribution of congenital anomalies to general perinatal mortality is low as depicted in Table III unlike in the West, where 20-25% of the perinatal losses occur due to anomalies (MJ Whittle 1991). Here abruptio placenta, low birth weight and unexplained still births still make a significant contribution to perinatal mortality. From this data it may seem that our restricted resources need to be used for bringing down perinatal mortality due to these causes rather than be used for a large scale antenatal screening programme for detection of anomalies. Second trimester MSAFP determination

and ultrasound scanning are the popular screening methods in the developed countries today. However the advantages of these methods is not restricted to the diagnosis of anomalies alone. Elevated MSAFP is said to be a marker for high risk pregnancy conditions like IUGR, preterm birth, PIH, abruptio placenta and high perinatal mortality, in the absence of structural defects (Katz et al 1990). So this will prove beneficial in developing countries where the incidence of these conditions is high. Similarly early scan is useful to confirm gestational age, check viability, rule out multiple pregnancy and assess fetal growth.

IUGR, preterm labour, breech presentation, oligoamnios and hydramnios are often seen in pregnancies with congenital anomalies (Sheriyar et al 1987). These can serve as clues for detailed scans to rule out anomalies. Sharma & Vijayvergia (1989) found high incidence (26%) of congenital anomalies in IUGR babies. Ganapat, Bhide and Sawant et al (1991) found congenital anomalies in 16.6% of breech babies as opposed to 1.1% in the control group. Incidence of breech presentation is partly high because many of the congenital anomaly pregnancies have preterm labour.

Majority of our women had anomalies detected in the third trimester because they reported late to the hospital. But they had early antenatal check up in peripheral clinics. Therefore it may be worthwhile to make the practitioners aware of the significance of early scanning in pregnancy. Luck CA (1992) in a prospective study of 4 years evaluated the effectiveness of routine ultrasound scanning in unselected, general population. He observed that early anomaly scan brought down the perinatal mortality and morbidity significantly. Besides, saves a lot of anxiety for the couple and their family. Any standard obstetric care today should include anomaly screening as a part of routine antenatal care.

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