CONGENITAL ANOMALIES THE OBSTETRIC PERSPECTIVE

MENON V. K. • BHARUCHA K. E.

SUMMARY

This study comprises 52 cases of congenital foetal anomalies diagnosed (either antenatally or postpartum) between August 1992 and July 1993 at the Sassoon General Hospitals, Pune. The patients were interviewed and possible high risk factors acting upon the present pregnancy were scrutinised. Sixty-five percent of the cases were registered antenatally with us 40% were primigravidae with a mean age of 24 yrs. 53.8% of the labours were preterm. Of the 30 live-births, 12 died in the neonatal period. The male : female ratio of the offspring was 3:2. Twenty three percent of the cases were diagnosed on USG at a mean age of 30 wks. and the findings correlated well with the postpartum gross appearance. The majority of the anomalies seen were neural tube defects (45%). There were 5 cases with minor anomalies and 6 cases with multiple anomalies. Polyhydramnios was associated with anomalies in 32.7% and IUGR detected in 15%. The clinical impression of a normal pregnancy was falsely noted in cases like meningocoele, encephalocoele and congenital diaphragmatic hernia. No significant history regarding environmental influences as a factor in the genesis of foetal anomaly could be elicited from our study group.

INTRODUCTION

The increasing proportion of foetal and infant morbidity and mortality due to congenital foetal malformations in our practice compelled us to study the associations of foetal anomalies and the circumstances in which they occured. The role of prenatal diagnosis is emphasised in the early detection of foetal malforma-

Dept. of Obst. & Gyn. & Family Welfare B.J. Medical College, Pune.

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tion which might be amenable to prenatal therapy or may require termination of the pregnancy.

Congenital anomalies : the obstetric perspective

This study comprises 52 cases of congenital anomalies both major and minor. The analysis revealed the following clinical profiles. Sixty-five percent of the cases had registered for antenatal care in our hospital while 35% arrived as emergencies in labour.

Forty percent of them were primigravidae. They were equally distributed throughout the reproductive age group with a mean age of 24 yrs.

Environmental influences like drug and radiation exposure and substance abuse were not found to be responsible for the genesis of congenital anomalies in our series. None of the group could either recall having suffered from significant febrile illness. There was one patient with the history of congenital anomaly in the previous pregnancy, one known diabetic and five cases with a history of a consanguinous marriage (second degree).

Six patients gave a history of a previous spontaneous abortion. The anomalies seen in this group were neural tube defects (3 cases) T-O fistula, congenital diaphragmatic hernia and cystic hygroma. There were 2 patients with oligohydramnios.

Thirty two percent of the cases were picked up on account of polyhydramnios. The diagnosis of anomaly was made antepartum in 23%, intrapartum in 7.7% and postpartum in 69.2%. Of the anomalies diagnosed post-partum, 75% were major anomalies.

Ultrasonographic Diagnosis

An antenatal USG diagnosis of congenital foetal anomaly was made in 12/52 (23%) cases. One case of hydrocephalus was missed on USG Scan done at 32 wks. of gestation. The main indications for doing a USG scan being -

Clinical evidence of polyhydramnios (8 cases), unsure dates/Clinical suspicion of IUGR (3 cases), Breech presentation (1 case)

The mean age at which the USG was done was 30 wks. The postpartum gross findings correlated well with the antenatal diagnosis.

Types of Anomalies

Among the major anomalies, 45% were neural tube defects, in addition to hydrocephalus, congenital diaphragmatic hernia, T-O fistula, sacrococcygeal teratoma and osteogenesis imperfecta. Minor anomalies like polydactyly and external pinna deformities were also seen. Multiple anomalies in the form of syndromes, eg. Meckel Gruber, and associations, eg. VATER, were seen in 6 cases.

Obstetric Outcome

Preterm deliveries accounted for 53.8% of cases of which 5 cases were induced labours.

Full term deliveries occured in 44.2% cases and there was one case with a postdated pregnancy. Two pregnancies were terminated by Caesarean section for an obstetric indication. The congenital anomalies associated with breech presentation were neural tube defects (60%), hydrocephalus (30%) and others (10%).

Pregnancy Outcome

Abortion accounted for 1.9% of cases and stillbirths accounted for 40.3% of the cases. Of the 30 live births, 12 i.e. 40% were neonatal deaths. The male to female ratio of the offspring was 3:2.

DISCUSSION

In our study we had a relatively young age group being affected, (mean age 24 yrs.) among which 40% were primigravidae. This has serious implications regarding risk for future pregnancies as well as the psychological stress that a woman, who is pregnant for the first time, has to go through.

According to Walkinshaw and Burn (1962), pregnancies may be predictably at risk for the development of congenital anomaly or unpredictably so. No significant history was elicitable from our patients regarding the influence of environmental factors like drug and radiation exposure or substance abuse. We had two cases with evidence of intrauterine infection with rubella, confirmed by positive maternal titres.

We had one case of a major anomaly in a woman who was a pregestational diabetic but screening for abnormal blood sugar levels did not give positive results in the cases studied.

Consanguinity has been associated with a tenfold increase in the risk of recessive disorders in consanguinous marriages (second degree) were observed in 96% of our cases.

Three percent of severe IUGR's have chromosomal anomalies. In Suijders et al study of karyotyping on 458 foetuses with IUGR between 17 and 37 wks. gestation, abnormal karyotype was found in 19%. We were able to detect IUGR in 4 cases both clinically and with USG confirmation.

Chamberlain et al state that a liquor pool greater than 8 cm. is associated with a malformation rate of 4%. This is 8 times that of a pregnancy with normal liqour volume. In the present study polyhydramnios was diagnosed in 37.5% cases. In the series by Golan et al. Of 41 congenital malformations, in 28 new borns in 197 cases of polyhydramnios, he found central nervous system malformations in 4.5%, cardiovascular anomalies in 4.5%, musculoskeletal anomalies in 4.1% and urinary and reproductive tract anomalies in 3.6%. Associated with polyhydramnios we had CNS malformations in 14 (82.3%), musculoskeletal in one case, gastrointestinal tract anomaly in one case and one case of a sacrococcygealteratoma.

Downs Syndrome, Anencephaly, Spina bifida, CTEV and low birth weight are associated with previous foetal loss (Paz J. E. et al 1992). We found mainly neural tube defects in this group.

Incidentally 2 of our cases were under treatment for Primary infertility. This has tragic undertones in that a couple eager to conceive finally does so only to produce an anomalous child.

The major bulk of the diagnosis (approx. 70%) were made postpartum. The mean gestational age at which an antenatal USG diagnosis of foetal anomaly was made was 30 wks. An earlier antenatal diagnosis would allow us the option of therapeutic termination of the pregnancy before the age of viability. In our set up, where antenatal USG exami-

nation of every patient is not fesible, the obstetrician often relies on clinical acumen and the patients history in screening out high risk cases to be evaluated further ultrasonographically. Clinical suspicion played a role in detecting anomalies in 21 cases of IUGR and polyhydramnios. However major anomalies like encephalocoele, meningocoele and congenital hernia were missed when the clinical impression was a pregnancy normal in terms of appropriate growth and liquor volume.

Of the 30 livebirths, 12 died in the neonatal period. The cases that left the nursery alive were non-life threatening conditions like CTEV, congenital dislocation of hip, etc.

Sixty percent of the affected foetuses were male. This is of social significance in our country where great importance is attached to the birth of a healthy male child.

Concluding Remarks

Though counselling regarding future pregnancies must be done by a qualified medical geneticist, pregnancies complicated by congenital anomalies of the foetus are of supreme importance to us as obstetricians who have to look after the perinatal welfare of the baby as well as the psychological stresses that the patient and her family have to go through.

This study conducted at a tertiary hospital has revealed that to develop a multidisciplinary approach for the successful management of pregnancy there is a greater need for public education, and also amongst the obstetric fraternity, in the screening techniques of prenatal diagnosis.

We have since improved our pick up-rate of congenital foetal malformations diagnosed antenatally with the experiences gained in this field.

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