



Original Article

Pattern of Congenital Anomalies in A Tertiary Care Centre

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Abstract

Aim of the Study- *The aim of this study is to identify the common patterns of anomalies seen in the hospital population of a tertiary care centre in Kerala, India.*

Materials and Methods- *This descriptive study was carried out in the obstetric unit of a tertiary care centre in Kerala for a period of one year. All patients who delivered or who had a second trimester abortion [from 14 weeks] were included in the study. All the fetus and the newborns were examined for the presence of congenital anomalies and mothers were interviewed for socio-demographic variables.*

Results- *During the study period, 15227 babies were born, of which 379 had congenital malformations, making the prevalence 2.48 %. 78 newborns (20.58%) had multiple anomalies involving more than one system. Out of the 379 cases, 248 (65.4 %) survived. The predominant system involved was Musculo-skeletal system (24 %) followed by central nervous system (21.4 %). Out of the 131 perinatal losses, 47 cases (35.9%) were due to CNS anomalies. Talipes (15.3 %) was the most common one in musculoskeletal group while hydrocephalus (7.9%) was highest in the central nervous system anomalies. The study found that congenital anomalies were associated with low birth weight, prematurity, malpresentation, consanguinity and polyhydramnios.*

Conclusion - *In spite of high institutional deliveries in Kerala, India, prevalence of congenital anomalies remains high. Increased awareness about preventable risk factors may help in reducing the incidence of congenital anomalies.*

Keywords: *Congenital anomaly, prevalence, neonates.*

Introduction

A congenital abnormality is an abnormality of structure, function or body metabolism that is present at birth and results in physical or mental disability or is fatal ^[1]. There are more than 4000

known birth defects. The long term disability caused by congenital anomalies may have a significant impact not only on the child's wellbeing and development but also on families, health care systems and societies ^[2]. Even though

the incidence of congenital anomalies varies from place to place, approximately 3-7 % of children are born with birth defects ^[2,3]. In India the incidence is around 2.5 % ^[4] and accounts for 8-15 % of perinatal deaths and 13-16 % of neonatal deaths in India ^[5,6].

In Government Medical College, Trivandrum, nearly 15000 deliveries take place every year. This study was planned to analyze all the congenitally abnormal children delivered at our hospital over a period of one year.

Material and Methods

The study was a descriptive prospective study which was done in the obstetric unit of government medical College, Trivandrum, Kerala, India. It included all women who were terminated for a congenital anomaly of the fetus or who delivered an anomalous baby after 14 weeks of gestation. All the live born, still born, intrauterine deaths and neonatal deaths were included in the study. All the fetuses induced following detection by ultrasound and those babies detected to have congenital anomalies by postnatal examination were included in the study.

Relevant history including familial and gestational factors were recorded in a preplanned Performa. The babies were examined immediately by pediatrician after delivery and sex, birth weight and type of anomaly were recorded. The live babies were admitted to NICU for observation, investigation, evaluation and management. System wise classification of anomalies were performed and multiple major congenital anomalies were counted only once by the system for the most serious anomaly.

Statistical Analysis

The data was entered in to master sheet and necessary statistical tables were constructed. To test hypothesis, statistical tests like chi- square test and Odd's ratio were used.

Observations and Results

Total babies born in the study period were 15,227[including the second trimester abortions from 14 weeks of gestation]. Total babies with congenital abnormality were 379(2.48 %).

Demographic characteristics of the patients with fetus having congenital anomalies

| | | |
|-------------------|--------|------|
| Maternal age | number | % |
| < 20 years | 22 | 5.8 |
| 20-35years | 298 | 78.7 |
| >35 years | 59 | 15.5 |
| Gestational age | | |
| Term | 248 | 65.4 |
| Preterm | 131 | 34.6 |
| Gender | | |
| Male | 211 | 54.4 |
| Female | 164 | 42.3 |
| Unidentified | 4 | 3.3 |
| Parity | | |
| Primi | 222 | 58.6 |
| Multi | 157 | 41.4 |
| Type of pregnancy | | |
| Singleton | 370 | 97.6 |
| Twin | 9 | 2.4 |
| Birth weight | | |
| <2.5 | 179 | 47.2 |
| ≥2.5 | 200 | 52.8 |

Table 1: Pattern of anomalies observed

| SYSTEM | Number | % |
|------------------------|--------|------|
| Musculoskeletal system | 91 | 24 |
| Central nervous system | 81 | 21.4 |
| Genitourinary system | 62 | 16.4 |
| Gastrointestinal tract | 54 | 14.2 |
| Cardiovascular system | 35 | 9.2 |
| Syndromes | 12 | 3.2 |
| Miscellaneous | 44 | 11.6 |
| Total | 379 | 100 |

Musculoskeletal were the commonest in the present study constituting around 24 %.

Table 2: Perinatal outcome of congenital anomalies

| Outcome | cases | % |
|----------|-------|-------|
| Mortal | 131 | 34.57 |
| Survival | 248 | 65.43 |
| Total | 379 | 100 |

Table 3: Pattern of anomalies noted in mortality group

| System | Number of cases | Mortal | | Survival | |
|-----------------|-----------------|--------|------|----------|------|
| | | number | % | Number | % |
| CNS | 81 | 47 | 58 | 34 | 42 |
| CVS | 35 | 8 | 22.8 | 27 | 77.2 |
| GIT | 54 | 31 | 57.4 | 23 | 42.6 |
| Genitourinary | 62 | 12 | 19.3 | 50 | 80.7 |
| musculoskeletal | 91 | 9 | 9.8 | 82 | 90.2 |
| syndromes | 12 | 4 | 33.3 | 8 | 66.7 |
| miscellaneous | 44 | 20 | 45.5 | 24 | 54.5 |
| Total | 379 | 131 | 34.6 | 248 | 65.4 |

Central nervous system anomalies constitute to the maximum perinatal mortalities.

Table 4 :- Musculoskeletal anomalies

| Musculoskeletal abnormalities | Count | % to Systems | % to total |
|-------------------------------|-------|--------------|------------|
| CTEV and valgus | 58 | 63.7 | 15.3 |
| Polydactyly | 8 | 8.8 | 2.1 |
| Achondroplasia /dwarfism | 8 | 8.8 | 2.1 |
| Syndactyly | 4 | 4.4 | 1.1 |
| Craniosynostosis | 4 | 4.4 | 1.1 |
| Congenital dislocation of hip | 4 | 4.4 | 1.1 |
| Genurecurvatum | 3 | 3.3 | 0.8 |
| Osteogenesis imperfect | 2 | 2.2 | 0.5 |
| Total | 91 | 100 | 24 |

Among the musculoskeletal anomalies, CTEV was found to be the commonest.

Table 5: Central nervous system anomalies

| CNS anomalies | Count | % to systems | % to total |
|---------------------------------|-------|--------------|------------|
| Hydrocephalus | 30 | 37 | 7.9 |
| Myelomeningocele | 11 | 13.6 | 2.9 |
| Hydrocephalus +myelomeningocele | 8 | 9.9 | 2.1 |
| Microcephaly | 7 | 8.6 | 1.8 |
| Anencephaly | 6 | 7.4 | 1.6 |
| Cystic hygroma | 5 | 6.2 | 1.3 |
| Dandy Walker | 4 | 4.9 | 1.1 |
| Vermian agenesis | 3 | 3.7 | 0.8 |
| Encephalocele | 2 | 2.5 | 0.5 |
| Teratoma | 1 | 1.2 | 0.3 |
| Arnold Chiari Malformation | 1 | 1.2 | 0.3 |
| Porencephalic cyst | 1 | 1.2 | 0.3 |
| Holoprosencephaly | 1 | 1.2 | 0.3 |
| Arachnoid cyst | 1 | 1.2 | 0.3 |
| Total | 81 | 100 | 21.4 |

Among the 81 cases of central nervous system anomalies, the most common anomaly was found to be hydrocephalus.

Table 6: Gastrointestinal anomalies

| GIT anomalies | Count | % to systems | % to total |
|---------------------------|-------|--------------|------------|
| Diaphragmatic hernia | 14 | 25.9 | 3.7 |
| Cleft palate | 11 | 20.4 | 2.9 |
| Cleft lip and palate | 9 | 16.7 | 2.4 |
| Esophageal atresia | 7 | 13 | 1.8 |
| Lower GI atresia | 5 | 9.3 | 1.3 |
| Omphalocele | 4 | 7.4 | 1.1 |
| Jejunal atresia | 2 | 3.7 | 0.5 |
| Tracheoesophageal atresia | 1 | 1.9 | 0.3 |
| Cleft lip | 1 | 1.9 | 0.3 |
| Total | 54 | 100 | 14.2 |

Among gastrointestinal tract anomalies, cleft lip and cleft palate together or in isolation constitutes the maximum.

Table 7: Cardiovascular system anomalies

| CVS anomalies | Count | % to systems | % to total |
|--------------------------|-------|--------------|------------|
| VSD | 14 | 40 | 3.7 |
| ASD | 7 | 20 | 1.8 |
| PDA | 4 | 11.4 | 1.1 |
| Congenital heart block | 3 | 8.6 | 0.8 |
| Congenital heart disease | 1 | 2.9 | 0.3 |
| Ebstein anomaly | 1 | 2.9 | 0.3 |
| TGA | 1 | 2.9 | 0.3 |
| Dextrocardia | 1 | 2.9 | 0.3 |
| Rhabdomyoma | 1 | 2.9 | 0.3 |
| Hypoplastic left heart | 1 | 2.9 | 0.3 |
| Dyke David of mason | 1 | 2.9 | 0.3 |
| Total | 35 | 100 | 9.2 |

Among the cardiovascular anomalies, left to right shunt defects e.g.: VSD, ASD, and PDA constitute the maximum number and among which VSD is more common.

Table 8: Genitourinary system anomalies

| GUS anomalies | Count | % to systems | % to total |
|----------------------------|-------|--------------|------------|
| Hydronephrosis | 25 | 40.3 | 6.6 |
| Renal Agenesis | 10 | 16.1 | 2.6 |
| Cryptorchidism | 5 | 8.1 | 1.3 |
| Hypospadiasis | 4 | 6.5 | 1.1 |
| Congenital hydrocoele | 4 | 6.5 | 1.1 |
| Ambiguous genitalia | 4 | 6.5 | 1.1 |
| Polycystic kidney | 4 | 6.5 | 1.1 |
| PUJ obstruction | 3 | 4.8 | 0.8 |
| Micropenis | 2 | 3.2 | 0.5 |
| Bladder outlet obstruction | 1 | 1.6 | 0.3 |
| Total | 62 | 100 | 16.4 |

Among the genitourinary system anomalies, hydronephrosis was found to be commonest.

Table 9: Syndromes

| Syndromes | Count | % to systems |
|----------------|-------|--------------|
| Downs | 8 | 66.7 |
| Pierre Robbins | 2 | 16.6 |
| turners | 1 | 8.3 |
| Trisomy 18 | 1 | 8.3 |
| Total | 12 | 100 |

There were 44 miscellaneous anomalies among which the maximum were hydrops fetalis (13), facial dysmorphism(6) and congenital rubella.

Discussion

This study was done to find out the incidence of congenital anomalies in a tertiary care centre in Kerala. With improved control of infections and nutritional deficiency diseases, congenital anomalies have become important causes of perinatal mortality in developed countries and will soon become an important determinate of perinatal mortality in developing countries. The pattern and prevalence may vary over time and with geographical location and there are no reliable estimates of the number of children who were born with a serious congenital disorder due

to genetic or environmental causes. In this hospital based study, the incidence of congenital anomalies was 2.48 % of the total number of fetus and babies born after 14 weeks of gestation in our hospital during a period of one year. These findings are comparable to similar studies from India, which reported an incidence of 2.72 % and 1.9 % [7,8]. Although similar results were obtained from studies from other countries also, the incidence in our hospital is actually lower as we have also included the second trimester abortions and still births. Our hospital being a tertiary care centre, which is strictly referral, usually gets

complicated cases and hence the prevalence in the hospital cannot be projected in to total population. Out of the 379 cases, 248 (65.4 %) survived. 25 cases were terminated in the second trimester itself, following ultrasound detection, 9 were macerated stillbirths and 67 were early neonatal deaths.

The most common anomaly detected was musculoskeletal malformations. 91/ 379(24 %) had some or other musculoskeletal malformations. Of the musculoskeletal deformities, majority was contributed by talipes equinovarus. These findings were similar to the results in other studies^(14,15,17). This was followed by central nervous system anomalies. Among the 81 cases of CNS anomalies, hydrocephalus was the commonest. 47 out of 81 (58%) were lethal. Many studies have shown that CNS malformations were associated with high perinatal mortality⁽¹²⁾.

Genitourinary system malformations were found in 62 cases (16.4 %) of which hydronephrosis were 25 in number. Gastrointestinal tract malformations were found in 54 cases of which 31(54.7%) were lethal. There were 14 cases of congenital diaphragmatic hernias and all were neonatal deaths. Cleft lip and cleft palate were the most common non lethal malformations.

Cardiac anomalies were found to be 9.2%. This is comparable to studies conducted in JIPMER, Pondicherry⁽¹⁵⁾. Swedish Malformation Registry has recorded that cardiovascular malformations occurred in 24 % of the infants with associated defects and ventricular septal defects were the most common heart anomalies. In the present study also ventricular septal defects were found to be the maximum number of cases.

There were 12 cases of clinically identifiable syndromes. 8 were down's syndromes. There were 13 cases of hydrops fetalis. This high number is perhaps because of the antenatal detection and referral. Males were more affected in the study which was similar to other studies. This was similar to other studies^(10,16).

Among the 379 newborns, 78 (20.58 %) were found to have multiple anomalies. Mishra and

Bhaveja found multiple anomalies in 37.6 % of cases and Swain S et al reported multiple anomalies in 18.8 % of babies.

The study found that congenital anomalies were more in babies of consanguineous marriage, was associated with low birth weight, preterm labour, Malpresentation and polyhydramnios. This is similar several previous studies^(9,10,11).

Despite Kerala being a state with high female literacy rates and with more than 99 % institutional deliveries⁽¹⁸⁾, the incidence of anomalies remains high. It is important to increase awareness about the consequences of consanguineous marriages and about the need of diabetic control before conception. Women in reproductive age group should be counseled about the benefits of folic acid supplementation especially preconceptional in the high risk group. Rubella vaccination should be recommended for adolescent girls and in the early postpartum period.

Conclusion

The increasing proportion of fetal and infant morbidity and mortality due to congenital fetal malformations in our practice compelled us to study about the prevalence of congenital abnormalities. This study indicates the high prevalence of congenital abnormalities in our locality and points toward the need to maintain a congenital malformation registry. As the prevalence in the hospital [being a tertiary care centre], cannot be projected in to total population, community base studies are needed to determine the exact prevalence of congenital anomalies and their associated factors.

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References

1. World Health Organization. Section on congenital anomalies. [Cited on 2012 Oct]. Available from: <http://www.who.int/mediacentre/factsheets/fs370/en/>
2. Howson CP, Christianson AC, Modell B: Controlling birth defects: reducing the

- hidden toll of dying and disabled children in low-income countries. Dis Control Prior Proj. 2008.
3. Singh A, Gupta RK: Pattern of congenital anomalies in newborn: a hospital based prospective study. JK Sci. 2009, 1: 34-36.
 4. Parmar A, Rathod SP, Patel SV, Patel SM: A study of congenital anomalies in newborn. NIJRM. 2010, 1: 13.
 5. Bhat BV, Ravikumara M. Perinatal mortality in India-Need for introspection. Indian J Matern Child Health. 1996;7:31-3.
 6. Agarwal SS, Singh U, Singh PS, Singh SS, Das V, Sharma A, et al. Prevalence and spectrum of congenital malformations in a prospective study at a teaching hospital. Indian J Med Res. 1991;94:413-9.
 7. Chaturvedi P, Banerjee KS. Spectrum of congenital malformations in newborns from rural Maharashtra. Indian J Pediatr. 1989;56:501-7.
 8. Taksande A, Vilhekar K, Chaturvedi P, Jain M. Congenital malformations at birth in Central India: A rural medical college hospital based data. Indian J Hum
 9. Gupta RK, Singh A, Gupta R. Pattern of congenital anomalies in newborn at birth: A hospital based prospective study. Proceedings of the 42nd National Conference of Indian Academy of Pediatrics (Pedicon); Jan 6-9; Kolkata, India. 2005. Genet. 2010;16:159-63.
 10. Sarkar S, Patra C, Dasgupta MK, Nayek K, Karmakar PR. Prevalence of Congenital Anomalies in Neonates and Associated Risk Factors in a Tertiary Care Hospital in Eastern India. *Journal of Clinical Neonatology*. 2013;2(3):131-134. doi:10.4103/2249-4847.119998.
 11. Swain S, Agrawal A, Bhatia BD. Congenital malformations at birth. Indian Pediatr. 1994;31:1187-91.
 12. Duru Shah: Perinatal audit. A report produced by the perinatology committee of the FOGSI – 1998.
 13. Dutta V, Chaturvedi P. Congenital malformations in rural Maharashtra. Indian JPediatr. 2000;37:998-1001.
 14. Suguna Bai NS, Mascarene M, Syamalan K, Nair PM. An etiological study of congenital malformation in the newborn. Indian Pediatr. 1982;19:1003-7.]
 15. Bhat VB, Babu L. Congenital malformations at birth- A prospective study from south India.. Indian J Pediatr 1998;65: 873-881.
 16. Mohanty C, Mishra OP, Das BK, Bhatia BD, Singh G. Congenital malformations in newborns: A study of 10,874 consecutive births. J Anat Soc India. 1989;38:101-11.
 17. El Koumi MA, Al Banna EA, Lebda I. Pattern of congenital anomalies in newborn: a hospital-based study. *Pediatric Reports*. 2013;5(1):e5. doi:10.4081/pr.2013.e5.
 18. Family Welfare Statistics" (PDF). Statistics Division, Ministry of Health and Family Welfare, *Government of India*. p. 182.