PATTERN OF CONGENITAL MALFORMATIONS AND THEIR NEONATAL OUTCOME

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ABSTRACT

Objective — *To determine the pattern of major congenital malformations in neonates admitted in a tertiary care NICU and evaluate their early outcome.*

Study design — *Descriptive study.*

Place & Duration of study — *Department of Pediatrics, Liaquat National Hospital & Medical College Karachi, from January to December 2009.*

Methodology — *A total of 431 neonates were admitted in NICU including those referred from outside. Children with major congenital malformations were identified by clinical examination and confirmed by appropriate radio-diagnostic methods. These neonates were immediately referred to the surgical team for intervention.*

Results — *A total of 57 neonates were admitted during the study period. Thirty one were males and 26 females. Fetal anomalies were diagnosed correctly in 17 cases out of a total of 19 inborn deliveries on maternal ultrasound while it was missed in one fetus and incorrectly diagnosed in one case. A total of 48 patients had surgery out of which 4 (8.3%) died in the neonatal period. Five cases were booked for elective surgery beyond the neonatal period. Out of 4 neonates with congeintal heart disease one case was referred outside, one neonate died preoperatively while 2 infants were managed conservatively.*

Conclusions — *Due to detection of fetal anomalies, early surgical intervention, and intensive neonatal care, most infants can be rescued after a successful primary operation.*

Key words — *Congenital, Malformations, Neonate.*

INTRODUCTION:

Congenital malformations are morphologic defects that originate in the prenatal period as a result of genetic mutations, chromosomal aberrations and/ or adverse intrauterine environment. A congenital physical anomaly is abnormality of structure of any body part that can be present at birth or become clinically manifest anytime later in life. There is a wide variety of fetal problems which range from relatively minor abnormalities to major structural defects.[1] Minor anomalies involve non vital organs with little or no functional effects. They do not cause any distress in the newborn and usually there is no urgency for their correction especially in the neonatal period. In contrast, major or severe anomalies impair function or are of significant cosmetic value. They may even be life threatening. Thus they require immediate correction. If not corrected early major anomalies could also impair the child's well being and development.

A prenatal diagnosis is possible in 2nd trimester on

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maternal sonography.[2, 3] As such, neonatal surgical interventions can be taken soon after birth. The corrective procedures not only try to restore the structure but also the function. The cosmetic effect is also improved.

According to international data about 2-3% of babies are born with significant congenital birth defects. Limited data is available on the incidence, pattern and neonatal outcomes of congenital anomalies from Pakistan. However, with improvements in strategies for neonatal survival in Pakistan the problem of congenital anomalies and related complexities are likely to emerge soon.[4] This study was therefore undertaken in a tertiary care neonatal unit to determine the pattern of congenital malformations and their outcome.

METHODOLOGY:

This descriptive study was carried out at Liaquat National Hospital and Medical College (LNH), Karachi, on all the neonates admitted in NICU from January to December 2009. All cases with major congenital anomalies were enrolled. A detailed history for any risk factor was taken. A thorough physical examination was performed. Confirmation of internal defects was done by various imaging modalities i.e., radiography, ultrasound, echocardiography, and CT scan. The anomalies diagnosed on prenatal maternal sound were confirmed by appropriate radio diagnostic method soon after birth.

The neonatal management consisted of initial stabilization of vital signs, ventilatory support if indicated, prevention and treatment of infection and correction of metabolic derangements. Surgical intervention was done as soon as the general condition of the baby permitted and postoperative neonatal mortality was noted.

RESULTS:

There were 431 admissions out of which a total of 57 cases had congenital malformations and enrolled in the study. Amongst those, 19 were inborn and 38 were outborn. There were 31 males and 26 females. All patients were actively managed surgically and medically. Reports of fetal ultrasound were available in all the 19 inborn cases. The anomalies were identified in 17 cases while it was missed in one case and incorrectly diagnosed in another.

Based on clinical examination and relevant investigations all cases were categorized into organ specific involvement. Frequency of specific organ involvement was determined and is given in table I. The most common anomalies were of gastro intestinal tract followed by genito urinary malformations. Regarding neonatal outcome, out of 57 cases, 48 were operated in LNH and 1 case of congenital heart disease was referred elsewhere as the child required urgent intervention. In a total of 48 operated cases, 4 died (8.3%) post operatively. Three neonates with congenital heart disease were managed conservatively out of which one baby died preoperatively. Five children with cleft lip, skin and musculoskeletal anomalies were planned for elective surgery beyond neonatal period and discharged.

DISCUSSION:

The present study indicates that congenital anomalies are important paediatric problem constituting 13% of total admissions in a tertiary care neonatal unit. The high number of congenital anomalies at LNH may be due to the fact that overall few centers offer pediatric surgery in Karachi, LNH being one of them.

The incidence of anomalies in our own hospital deliveries was 15.8 /1000 in live births, while other studies from Pakistan have described the frequency of anomalies in either total or still births. A study from Liyari General Hospital[5] reports it to be 11.4/1000 total births while a study from a university hospital in Sindh has shown it to be 16% in still births.[6] This variability from different centers may be due to various risk factors associated with congenital anomalies such as ethnicity, geographical distribution, consanguity, socio-cultural and nutritional factors. An Iranian study that included children from birth to eight years reports the prevalence of all congenital conditions to be 29.4/1000 live births, which impair the function with or without structural defects.[7]

The pattern of malformations is also different from other neighboring regions. In our study the most common pattern of anomalies was GIT defects. In studies from Iran musculoskeletal anomalies rank as the commonest,[8,9,10] while a study from India also reports the same.[11] Another Indian study reports CNS anomalies to be the most frequent.[12] All these are hospital based studies which may not reflect the overall status of the problem. Community studies need to be undertaken for getting a better picture of the problem.

Due to availability of prenatal diagnosis, detection of a malformation facilitates early surgical intervention. However foetal ultrasound may not pick up all cases.[13 14] Other related issues are emotional stress for parents who need counseling for early surgical intervention for better outcome. Another option is termination especially for conditions incompatible with life, for which appropriate laws are required.

Several congenital defects are surgically curable with complete recovery, while in others some improvement

Table I: Specific Malformations According to Systems Involved

System	Malformation	No. of Malformation	Percentage
Gastro-intestinal Tract	Esophageal Atresia with TEF	2	3.5
	Anorectal Malformation	5	8.8
	Intestinal atresia/stenosis	4	7
	Cleft lip/cleft palate	2	3.5
	Malrotation	1	1.7
	Diaphragmatic Hernia	3	5.3
	Hirschsprung's Disease	3	5.3
	Combined Defects	5	8.8
Total		25	44%
Genitourinary system	Hydronephrosis	3	5.3
	Hypospadias	3	5.3
	Epispadias	1	1.7
	Prune belly syndrome	1	1.7
	Inguinal hernia	4	7
	Ovarian cyst	1	1.7
	Urogenital sinus	1	1.7
Total		14	24.5%
Central Nervous System	Meningocele/ Meningomyelocele	4	7
	Hydrocephalus	3	5.3
	Sacrococcygeal terratoma	1	1.7
Total		8	14%
Cardiovascular System	Transpostion of great arteries	1	1.7
	Acyanotic disease	3	5.3
Total		4	7%
Respiratory tract	Tracheal stenosis	1	1.7
	Choanal atresia	1	1.7
Total		2	3.3%
Musculoskeletal System	Club foot	1	1.7
	Polydactyly	1	1.7
Total		2	3.5
Total (eye, skin)		2	3.5
Grand Total		57	100%

in function and quality of life can be achieved. However, often there is a necessity of repeated operations and hospitalization which increases the financial burden. Neonatal outcomes were generally satisfactory after surgery, which was offered in 48 cases with mortality in four. The main factor resulting in post operative mortality was found to be delayed referral since all postoperative deaths occurred in neonates referred from outside. The other associated contributory factors were prematurity and infections.

W.H.O and other international bodies also address the burden of congenital malformations and their outcomes and are prioritizing this as a public health issue.[15] Major focus is on early recognition and development of new surgical techniques. Improvement in training facilities and monitory grants may also be beneficial for improving the outcome in developing countries.

CONCLUSIONS:

The study gives an overview of pattern of congenital anomalies in a tertiary care center. Surveillance and monitoring of congenital conditions is important for identifying patterns of malformations. A nation wide surveillance can recognize the disease burden in pre and post natal period and related risk factors. This will be helpful for strategic planning to improve the outcomes.

REFERENCES:

1. Puri P, Diana DE. Preoperative assessment In Prem Puri (editor) Newborn Surgery 2nd edition London Arnold 2003:46

2. Balakumar K. Antenatal ultrasound screening for anomalies among singletons result of a prospective study. Calicut Med J 2004:2;1-10

3. Singh S, Shergill A, Singh R. Role of Ultrasound in detection of antenatal fetal malformations. Ind J Radiol Imag 2006;16:831-34.

4. Robert L. Brunt of environmental causes of human congenital malformations: The pediatricians role in dealing with these complex clinical problems caused by a multiplicity of environmental and genetics factors. Pediatrics 2004;113:957-68.

5. Parveen F, Tayab S. Frequency and pattern of distribution of congenital anomalies in the newborn and associated maternal risk factor. J Coll Physicians Surg Pakistan 2007;17:340-3.

6. Khaskheli M, Balouch S, Khushk IA, Shah SS. Pattern of fetal deaths at a university hospital Sindh, J Ayub Med Coll 2007;19:32-4.

7. Movafagh A, Zadah P, Javari MH et al. Occurrence of congenital anomalies and genetic diseases in a population of Ghazvin Province, Iran. Pak J Med Sci 2008;24:80-5.

8. Ali A, Shafikhani Z, Abdulahi M. Congenital malformations among live births at Arvand Hospital Ahwaz , Iran – A Prospective study . Pak J Med Sci 2008;24:33-37.

9. Khatemi F, Mamorri GA. Survey of congenital major malformations in 10,000 newborns. Iranian J Pediatrics 2005;5:315-20.

10. Golalipour MJ, Ahmadpour-kacho M, Vakili MA. Congenital malformations at a referral hospital in Gorgan, Islamic Republic of Iran. East Mediterr Health J 2005;:11:707-15.

11. Grover N. Congenital malformations in Shimla. Indian J Pediatr 2000;67:249-51.

12. Singh A, Gupta R K Pattern of Congenital anomalies In Newborn: A hospital based prospective study. JK Science 2009;1:34- 6.

13. Munim S, Nadeem S, Khuwaja NA. The accuracy of ultrasound in the diagnosis of congenital abnormalities. J Pak Med Assoc 2006;16:6-9.

14. Pitukki S, Chittacharoen A, Jetswangsri T, Panburana P, Jaovisidha A, Roungsipragarn R et al. The value of mid-trimester routine ultrasonoraphic screening in antenatal detection of congenital malformations. J Med Assoc Thai 2009;92:748-53.

15. Boyle CA, Cordero JF. Birth defects and disabilities: a public health issue for the 21st century. Am J Public Health. 2005:95:1884-6.