Original Article

Frequency of Congenital Anomalies in Southren Punjab

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ABSTRACT

Objective: To determine the pattern of major congenital malformations in neonates admitted in Nishtar Hospital Multan and evaluate their early outcome.

Study Design: Descriptive study.

Place and Duration of Study: This study was conducted at the Department of Anatomy, in collaboration with department of Gynaecology & Obstetrics and department of Paediatrics Nishtar Medical Institution Multan from March to August 2012.

Materials and Methods: A total of 431 neonates were admitted in Paediatric Wards including those referred from outside and from Labour Room of Nishtar Hospital. 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 with congenital malformations 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 congenital heart disease one case was referred outside, one neonate died preoperatively while 2 infants were managed conservatively.

Conclusion: 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 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.

MATERIALS AND METHODS

This descriptive study was carried out at Nishtar Medical Institution Multan, on all the neonates admitted to paediatric wards from March to August 2012.. 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.

Table No.1: 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%

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 Nishtar Hospital 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 Nishtar Hospital may be due to the fact that overall few centers offer pediatric surgery in Multan.

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 fetal 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 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[16]. Improvement in training facilities and monitory grants may also be beneficial for improving the outcome in developing countries.

CONCLUSION

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.

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