

Pattern of major congenital anomalies and their outcome

Malhotra P¹, Thapar K²

¹Dr Preeti Malhotra
Associate Professor, Paediatrics
drpreetimalhotra@yahoo.com

²Dr Karuna Thapar
Professor & Head, Paediatrics
drpreetimalhotra@yahoo.com
Sri Guru Ram Das Institute of
Medical Sciences and Research
Vallah, Amritsar, Punjab, India

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Correspondence to:
Dr Preeti Malhotra
drpreetimalhotra@yahoo.com

ABSTRACT

Background: Congenital anomalies are one of the most common causes of morbidity in developed and developing countries. Early detection and prevention of birth defects is necessary to further reduce the morbidity and mortality in neonates.

Objective: The study was to know the common congenital anomalies and their outcome in a tertiary referral centre.

Material and Methods: Study included 859 admissions in neonatal ICU Department of Paediatrics in a tertiary care hospital from January 2012 to December 2012. Neonates with congenital anomalies were diagnosed by clinical examination and appropriate investigations with exclusion criteria of minor anomalies. Medical or surgical intervention was done accordingly.

Results: Out of 859 admissions 59 were diagnosed to be suffering from congenital anomalies. The results showed a prevalence of congenital anomalies of 6.8% and out of that major cases were of GI disorders in the tune of 75% followed by nervous system disorders to the tune of 19%. 84.7% patients were males and 15.3% were females which could be because of the gender bias in the society though we cannot confirm it.

Conclusion: As compared to other studies GI disorders appear to be more common and by improvement in antenatal, postnatal diagnosis, early referral to tertiary hospital and early intervention most of these infants can be saved.

Key words: Congenital anomalies, birth defects, antenatal, gastrointestinal defects, prevalence

Introduction

Health is a multidimensional concept that is difficult to capture in a single measure. Common indicators like infant mortality rate, life expectancy at birth, anthropometric measures or nutritional status are used to assess the health status of population. But Most of the studies focus on the infections and their effects during infancy and childhood. Very few studies have been conducted to know about the morbidity rates and outcomes due to congenital anomalies. Congenital anomalies affect approximately 1 in 33 infants leading to 6.6% deaths in infants and causing significant morbidity in children. Ever since the discovery of Penicillin by Ian Fleming a lot of antibiotics have been introduced along with development in immunology and medicine also the understanding of the preventive aspect of the

infective diseases has lead to significant reduction in the morbidity and mortality of infectious diseases.^[1] But these have little impact on congenital anomalies but with the advancement of antenatal ultrasonography and availability of trained paediatric surgeons the congenital anomalies are being identified and treated well. Congenital anomalies also referred as birth defects, affect approximately 1 in 33 infants and results in approximately 3.2 million birth defects related disabilities every year.^[2, 3] Congenital anomaly can be defined as abnormality of physical structure or form seen at birth or few days/weeks after birth that has surgical, medical or cosmetic relevance. Community based study by Indian council of Medical Research (ICMR).^[4] Reported that congenital malformations accounted for 6.6% of neonatal deaths in the rural as

well as urban slum communities. The variability of incidence of malformation in various parts of the country could be due to inaccurate detection at birth or later or it can be due to various methodologies used. [5] Maternal ultrasonography can diagnose these anomalies prenatally in 2nd trimester of life. [6, 7, 8] and intervention of congenital malformations in the intrauterine life is gaining popularity now a days. Neonatal surgical intervention is done usually soon after birth, these procedures are not only to restore the structure but function also. As the diagnosis of congenital malformation invokes an emotional parental response [9] So the life threatening congenital malformations must be identified by thorough clinical examination because early diagnosis and surgical correction or palliation of these infants offers the best chances for survival.

This type of study was conducted in the view to find out the prevalence of congenital anomalies affecting various organ systems. This study also becomes important in this part of Punjab as people's desire for particular sex may lead to consumption of various drugs or other products during antenatal period which may harm the fetus or its organs. Environmental factors, genetic factors also affect the developing fetus. [10, 11] So this type of study may help to document the outcome and pattern of abnormality.

Materials and methods

This retrospective descriptive study was carried at SGRDIMSR, VALLAH, Amritsar which is a rural tertiary care teaching hospital, catering all the inborn, out-born babies and referrals from the various hospitals and periphery. The study included all the neonates being admitted to the neonatal nursery for evaluation, observation, investigation and management from January 2012 to December 2012. All the cases with major anomalies were enrolled

and minor anomalies like polydactyly, CTEV (congenital talipes equino varus) etc were excluded. Detailed history and thorough physical examination was done. Various imaging modalities like radiography, ultrasound and CT Scan/MRI were done as per requirement. The anomalies diagnosed on pre-natal ultrasonography were confirmed clinically or by appropriate radio-diagnostic methods soon after birth. The neonates were managed accordingly either medically or surgically. Post-operative outcome in terms of morbidity and mortality was noted. Analysis of the data was done using simple statistical method of recording number and percentage of cases.

Results

Total 859 neonates were admitted during the study period. Out of which 59 patients had congenital malformations making prevalence as 6.8%. There were 50 (84.7%) males and 9 (15.3%) females suffering from congenital anomalies. (Fig. 1) All patients were managed actively both medically and surgically. Reports of fetal ultrasonography were available in 40 cases (67.7%). Only 7(17.5%) cases of congenital anomaly were confirmed antenatally. Based on relevant clinical examination and investigations, all cases were categorized into organ specific involvement. The pattern of congenital anomalies in terms of frequency is shown (Table 1, Figure 2).

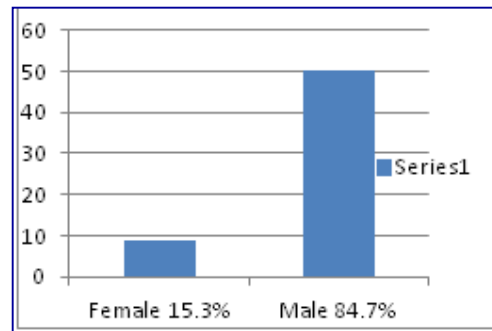


Fig. 1 Male to Female distribution

Table 1: Distribution of Congenital Anomalies according to System

Type of anomaly	No of Patients	Percentage
GASTROINTESTINAL		
ANNULA PANCREAS WITH DUODENAL ATRESIA	1	1.69%
ARM WITH TOF	1	1.69%
DUODENAL ATRESIA	1	1.69%
HIRSHSPRUNG	4	7.00%
ILEAL BAND	1	1.69%
ILEOILEAL INTUSSUSEPTION	6	11.00%
INESTINAL ATRESIA/STENOSIS	9	16.00%
INTESTINAL MALROTATION	1	1.69%
ARM	10	17.00%
OMPHALOCELE	1	1.69%
DIAPHRAGMATIC HERNIA	5	9.00%
TOF	4	7.00%
CNS		
MENINGOCELE/MENINGOMYELOCELE	8	14.00%
HYDROCEPHALUS	2	4.00%
SACROCOCCEGEAL TERATOMA	1	1.69%
RENAL		
B/L HYDRONEPHROSIS	1	1.69%
OTHERS		
CLEFT LIP/PALATE	2	
MUSCULOSKELETAL		
CLUB FOOT	1	1.69%

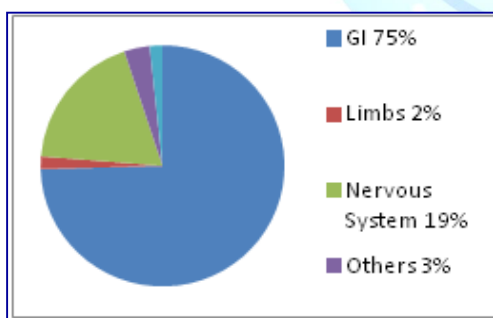


Fig. 2 System wise prevalence

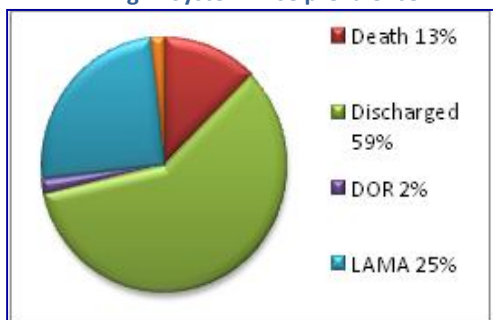


Fig. 3 Final outcome

The most common anomalies were of Gastrointestinal system 44(74.57%) cases followed by central nervous system anomalies 11(18.64%) and 4(6.77%) of miscellaneous cases. Regarding outcome (Figure 3) 37(62.7%) neonates were operated successfully and discharged well 11(18.6%) left against medical advice 8(13.5%) patients died pre operatively due to various reasons and 3(5%) patients died post operatively. The observations in the present study did not found any significant relationship to gravida. Primigravida were 30(50.8%) and multigravida were 29(49.1%), in comparison to other studies which showed higher incidence of congenital malformations among multigravida mothers. In our study there was no particular relationship with the blood

group of the patient. Incidence of congenital malformations in term babies was 45(76.2%) and in pre-terms 14(23.8%).

Discussion

Our study indicates that congenital anomalies are important paediatric problems constituting 7.3% of total neonatal admissions in one year. This is much higher than the incidence reported by Madi et al in Kuwait who reported it to be of 1.25%^[12] and almost similar incidence was reported by Swain et al of 1.2% in India. Similar incidence of 1.2% was reported from United Arab Emirates^[13] Alexandria and Cairo reported a incidence of 1.16% and 1.58%^[14, 15] but was still higher than the WHO reported incidence in other populations of other countries where it was reported to be 12.7 per 1000 in 16 countries^[14] Variable prevalence figures are reported from different countries; Spain showing prevalence of 20.23 per 1000,^[16] Libyan Arab Jamahiriya 20.23 per 1000,^[17] India 27.2 per 1000,^[6] where as Federal Republic of Germany showed it to be 6.9% for major congenital anomalies and 35.8% for mild errors of morphogenesis among live births, still births and abortions.^[18] Our figures match the German study and this almost double than the previous studies by Singh et al and Desai et al.^[19] Our hospital is a tertiary care and a teaching hospital in periphery. The high incidence of congenital anomalies seen in our institution may be because our hospital caters the patients from primary health centers and some other hospitals in the periphery. Pattern of congenital anomalies vary from region to region. In our study the most common pattern of congenital anomalies was GIT defects. Some studies report CNS anomalies to be more frequent. Male preponderance was more in our study as also reported in other studies^[1, 5, 20, 21] this needs to be further evaluated due to prevailing social structure resulting in high

female feticide rate. Prenatal ultrasonography leads to early detection of malformations and facilitates early surgical intervention. However, fetal ultrasonography may not pick up all cases. In our study 40 patients got ultrasonography reports but only in 7 patients anomalies were identified. There may be multi factorial reasons for these namely lack of awareness among people to get level 3 ultrasound during antenatal period, poverty, lack of proper health services in periphery. The actual incidence may be quite high as we studied only those congenital malformations that were brought to the hospital to get treatment.

We need to involve and make aware all the health care workers who are providing maternal and child health care working in government or private sector so as to quantify exact prevalence rate of congenital malformations involving any particular system. Mortality and morbidity can be because of sepsis, infections, prematurity or delay in treatment due to late admissions after diagnosis due lack of money, poor background and lack of awareness. This study definitely helps to know the pattern of congenital anomalies and their outcome in this area so that strategies for prevention, early detection and timely management can be sort out.

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