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Congenital anomalies in paediatric surgery in North India

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ABSTRACT

Background: Congenitalmal formation represents defects in morphogenesis during early foetal life. Congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India. The proportion of perinatal deaths due to congenital malformations is increasing as a result of reduction of mortality due to other causes owing to the improvement in perinatal and neonatal care. **Materials and Methods:** A retrospective record based study was conducted in the Paediatric Surgery Department of a Tertiary Care Institute of North India. The records of over a decade (2003-2012) were analysed. A total of 4305 cases of congenital anomalies were recorded in the study. All the data were entered in the excel spread sheet and analysed in SPSS version 17. (Statistical Package for the Social Sciences, a software package used for statistical analysis, officially named "IBM SPSS Statistics"). **Results:**Over the decade, a total of 14264 children were admitted in the paediatric surgery department with various problems. Out of these about one-third children (30.18%) had one or other type of congenital anomalies. This trend remained almost constant over the decade within a range of 26.8-33.6%. About half of the total congenital anomalies belonged to the gastro-intestinal tract, followed by genitourinary tract, central nervous system and other anomalies. All the anomalies were more common in males and were found in children belonging to rural communities. **Conclusions:** All the neonates should be examined with scrutiny for overt as well as occult congenital anomalies and Paediatric surgical care should be considered as an essential component of child health programmes in developing populations. Moreover, it is necessary to establish a registry system for congenital anomalies.

Key words: Congenital, paediatric surgery, malformations

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INTRODUCTION

Congenital malformation represents defects in morphogenesis during early foetal life. According to the World Health Organization, the term congenital malformations should be confined to structural defects at birth.[1] About 3% of newborns have a 'major physical anomaly', meaning a physical anomaly that has cosmetic or functional significance.[2] The leading causes of infant morbidity and mortality in poorer countries are malnutrition and infections, whereas in developed countries they are cancer, accidents and congenital malformations. Congenital anomalies account for 8-15% of perinatal deaths and 13-16% of neonatal deaths in India.[3,4]Patients with multiple congenital anomalies present a relatively infrequent but tremendously difficult challenge to the paediatrician. The proportion of perinatal deaths due to congenital malformations is increasing as a result of reduction of mortality due to other causes owing to the improvement in perinatal and neonatal care. In the coming decades, this is going to be a leading cause of morbidity and mortality in centres providing good neonatal care. The cause of 40-60% of congenital anomalies in humans is unknown. These are referred to as sporadic, a term that implies an unknown cause, random occurrence regardless of maternal living conditions and a low recurrence risk for future children.[5] For20-25% of anomalies, there seems to be a 'multifactorial' cause, meaning a complex interaction of multiple minor genetic anomalies with environmental risk factors. Another 10-13% of anomalies have a purely environmental cause (e.g. infections, illness, or drug abuse in the mother). Only 12-25% of anomalies have a purely genetic cause. Of these, the majority are chromosomal anomalies.[6]

Due to lack of facilities, even for routine medical examination, by a qualified doctor in rural areas, a large number of cases with malformations either die undetected or present with mental or physical handicap later in life.[7] Congenital anomalies contribute

a significant proportion of infant mortality as well as foetal morbidity. As a consequence, it is essential to have basic epidemiological information of these anomalies. Congenital anomaly rates can also used for planning health services. Hence, this study was undertaken to determine the frequency and type of congenital malformation in children presenting in the department of paediatric surgery.

MATERIALS AND METHODS

We conducted a retrospective record based study in the Paediatric Surgery Department of a Tertiary Care Institute of North India. This is the only government tertiary care institute in the state providing services to the entire population of the region. Nearly all paediatric-surgical facilities are provided in this institute. Hence, a reasonable coverage of all patients is expected. The records of over a decade (2003-2012) were analysed. Full record of all the patients admitted in the paediatric surgery department is accurately maintained by the trained hospital staff in the patient record registers. The record includes the identification data, socio-demographic data of all the patients, the diagnosis as per attending surgeon and the treatment provided.

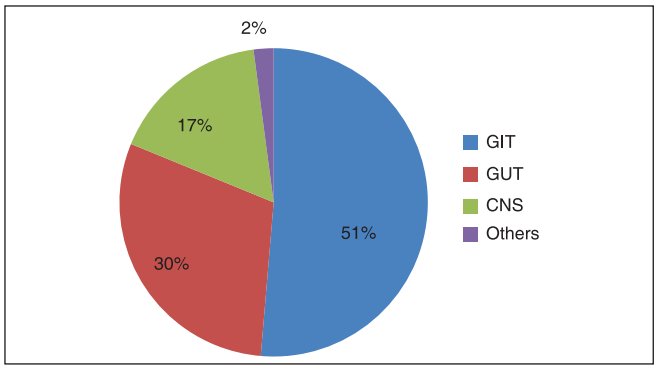


Figure 1: System-wise distribution of congenital anomalies

All the cases attending the paediatric surgery department and diagnosed as having congenital anomalies during the study period were included in the study. Thus, a total of 4305 cases of congenital anomalies were recorded in the study. Information regarding age, sex, residence etc., was noted from the available records by the researcher. Total cases, irrespective of the diagnosis, admitted during the study period were also recorded. Although it was a record based study, but the records were well maintained such that there was no problem in recording the information. All the data were entered in the excel spread sheet. The data were analysed in percentages and proportions. Categorical data were analysed by Chi-square test. All the analysis was done in SPSS version 17. SPSS was purchased and installed by the dept. of Community Medicine during some project.

RESULTS

From January 2003 – July 2012, a total of 14264 children attended the Paediatric Surgery Department with various problems. Out of these about one-third children (30.18%) had one or other type of congenital anomalies. This trend remained almost constant over the decade within a range of 26.8-33.6% [Table 1]. About half of the total congenital anomalies belonged to the Gastro-intestinal Tract (GIT), followed by Genitourinary Tract (GUT), central nervous system (CNS) and other anomalies [Figure 1].

When the year-wise distribution of the anomalies was plotted then the minimum variation was seen in GIT, CNS and other anomalies, but in GUT a dip was found in the year 2005 with the gradual rise in number of cases which peaked in the year 2010 [Figure 2].

When sex-wise distribution was done then GIT anomalies were found more common in males, this difference was found statistically significant ($P < 0.001$). GUT

Table 1: Distribution of paediatric surgical congenital anomalies over the decade

Years	GIT	GUT	CNS	Others	Total anomalies cases	Total cases	Percentage of anomalies
2003	230	105	72	9	414	1229	33.68
2004	238	109	71	3	418	1274	32.18
2005	227	73	88	5	393	1294	30.37
2006	223	97	71	11	400	1378	29.02
2007	234	111	70	17	430	1427	30.13
2008	240	141	63	8	450	1486	30.28
2009	247	167	81	12	501	1661	30.16
2010	229	233	87	8	554	1831	30.25
2011	247	168	88	14	515	1826	28.20
2012 (upto July)	118	81	29	3	230	858	26.80
Total	2210	1285	720	90	4305	14264	30.18

GIT-Gastro-intestinal tract; GUT-Genitourinary tract; CNS-Central nervous system

anomalies were mainly the anomalies of male children. Meningomyelocele and hydrocephalus were more common male children, but the difference was found statistically non-significant ($P = 0.221$), other type of anomalies were also more common in males and the difference was statistically significant ($P< 0.001$) [Table 2].

When distribution of the patient was seen as per their residence, then it was found that most of the patients were from rural communities and this difference was statistically significant in all types of anomalies except CNS anomalies, which did not have any significant association with the place of residence [Table 3].

DISCUSSION

This study was an effort to frame an actual picture of congenital anomalous babies in this tertiary care centre. In this institute a total of 14264 children presented with paediatric surgical problems over a decade, out of which 4305 children had one or other type of congenital anomalies.

The most common system involved in our study was the gastrointestinal system, followed by genitourinary tract. The findings were similar to a study done by Dutta *et al*. in which the malformations involving the gastrointestinal tract (26%) and genitourinary tract (25.8%) were the most common anomalies.[8]

In our study it was found that among GIT anomalies the most common anomaly was anorectal malformation followed by trachea-oesophageal fistula.The most common genitourinary malformation was hypospadias, while Meningomyelocele was the commonest congenital anomaly involving CNS. The results were same as what reported by Gorgan and Tehran.[9,10]

When the distribution of anomalies was plotted over a decade then the minimum variation was seen in GIT,

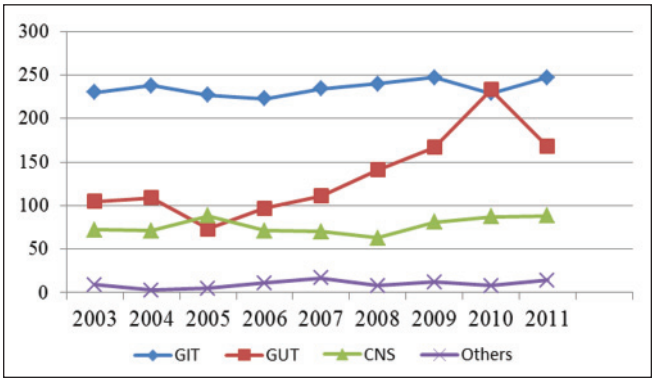


Figure 2: Year-wise distribution of congenital anomalies (2003-2011)

Table 2: Sex-wise distribution congenital anomalies

System involved	Male	Female	Total	*P* value
Gastrointestinal tract anomaly				
Anorectal malformation	662	444	1106	<0.001*
Tracheo-oesophageal fistula	407	201	608	
Intestinal atresia	71	32	103	
Congenital hernia/hydrocele	64	4	68	
Choledocal cyst	12	31	43	
Congenital hypertrophic pyloric stenosis	13	5	18	
Congenital diaphragmatic hernia	36	7	43	
Exomphalos/omphalocele	45	16	61	
Hirschprung's disease	83	15	98	
Imperforate anus	26	9	35	
Miscellaneous	13	14	27	
Genitourinary tract				
Hypospadias	761	NA	761	<0.001*
Undescended testes	502	NA	502	
Miscellaneous	15	7	22	
Central nervous system				
Meningomyelocele	494	203	697	0.221
Hydrocephalus	19	4	23	
Others				
Multiple anomalies	26	5	31	<0.001*
Teratoma	9	21	30	
Polydactyly/syndactyly	7	2	9	
Miscellaneous	17	3	20	

*Significant, NA-Not applicable

Table 3: Residence-wise distribution of congenital anomalies

System involved	Rural	Urban	Total	*P* value
Gastrointestinal tract anomaly				
Anorectal malformation	848	258	1106	0.007*
Tracheo-oesophageal fistula	482	126	608	
Intestinal atresia	75	28	103	
Congenital hernia/hydrocele	50	18	68	
Choledocal cyst	27	16	43	
Congenital hypertrophic pyloric stenosis	10	8	18	
Congenital diaphragmatic hernia	32	11	43	
Exomphalos/omphalocele	54	7	61	
Hirschprung's disease	82	16	98	
Imperforate anus	32	3	35	
Miscellaneous	20	7	27	
Genitourinary tract				
Hypospadias	565	196	761	0.008*
Undescended testes	332	170	502	
Miscellaneous	16	6	22	
Central nervous system				
Meningomyelocele	556	141	697	0.859
Hydrocephalus	18	5	23	
Others				
Multiple anomalies	24	7	31	0.028*
Teratoma	29	1	30	
Polydactyly/syndactyly	8	1	9	
Miscellaneous	13	7	20	

*Significant

CNS and Other anomalies, but a curious dip was found in case of GUT in the year 2005 the reason of which is unclear, more research is needed to clear the reason.

Male preponderance was found in the study when the anomalies were analysed as per the sex of the child. The results were almost similar to other studies.[9,11] Most important reason of male preponderance may be the better care of a male child as compared to the female in India. There is also social embarrassment associated with seeking consultation for the malformed genitourinary organs of the female child. Another reason is that some anomalies of GUT are confined to the male child e.g. hypospadias, un-descended testes and these defects of the GUT are more obvious in males.

When congenital anomalies were studied as per the residence of the child, then it was found that most of the children belonged to the rural communities and rural to urban ratio was 3:1. This may be due to the fact that in India around 70% population lives in rural communities. It might also be suggested that the rural population, which is mostly of low socio-economic background, goes to the government hospitals, while in the urban areas people usually prefer consulting the private doctors.

The difference between the frequency and types of congenital malformation in different parts of the country and reports from other countries may be due not only to genetic background but also to geographic, nutritional and socioeconomic differences. More research, preferably community based, is needed to determine the prevalence and the factors underlying the various types of congenital malformation encountered in this area so that our medical workforce could be acquainted with and our medical set-up geared up for the existing challenges.

SUGGESTIONS TO REDUCE CONGENITAL ANOMALIES

- Improving the diet of women throughout their reproductive years, ensuring an adequate dietary intake of vitamins and minerals such as folic acid and iodine and restricting harmful substances, particularly the abuse of alcohol. Controlling pre-conceptional and gestational diabetes.
- Avoiding exposure to hazardous environmental substances (e.g. heavy metals, pesticides, some medicinal drugs) during pregnancy.
- Improving vaccination coverage, especially with rubella virus, for children and women. This can be prevented through childhood vaccination. The rubella vaccine can also be given at least 1 month prior to pregnancy to women who are not already immune.
- Increasing and strengthening education to health staff and others interested in promoting birth defects prevention.

CONCLUSIONS

Congenital anomalies are a major cause of stillbirths and infant mortality. By thorough clinical examination, the life-threatening congenital malformation must be identified, as early diagnosis and surgical correction of the malformed babies offer the best chance for survival. To draw significant conclusions it is recommended that all neonates should be examined with scrutiny for overt as well as occult congenital anomalies.

RECOMMENDATIONS

- Screening of high risk cases, routine prenatal folic acid supplementation, early prenatal diagnosis and termination of foetus with lethal anomaly before attaining viability will reduce perinatal morbidity and mortality
- Moreover, it is necessary to establish a registry system for congenital anomalies
- More research is needed to determine the factors underlying the various types of congenital malformation encountered in this area
- Paediatric surgical care should be considered an essential component of child health programmes in developing populations.

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