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ORIGINAL ARTICLE

Major Congenital Malformations of Gastrointestinal Tract Among Neonates Presenting to Tertiary Care

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ABSTRACT

Background: Major congenital malformations of gastrointestinal tract (CMGIT), are usually manifested in early neonatal period by signs of intestinal obstruction and may be life threatening. A broad spectrum of Gastrointestinal Tract (GIT) anomalies can be seen including both upper and lower GIT. As compared to other congenital anomalies, GIT anomalies are not well studied and till now very few studies have reported prevalence and pattern of GIT malformations.

Objective: To determine different type of common gastrointestinal anomalies among neonates presenting at tertiary care hospital.

Methods: This descriptive cross sectional study was done at National Institute of Child Health (NICH) Karachi over period of 6 months, from 1-8-17 to 31-1-18. All neonates presenting to emergency department and got hospitalized with CMGIT were enrolled.

Results: There were 35 neonates enrolled with CMGIT. Out of these 16(45.7%) were male and 19(54.3%) were females. Mean age 9.4+3.82 (range 1-20) days. Mean weight was 2.5±0.98 (range 0.5-4.5) kg. Mean duration of symptoms was 5.8+1.72 (range 1-9) days. Among the CMGIT, 12 (34.3%) cases were of Anorectal malformation, persistent Vitelline duct was found in 6 (17.1%), Intestinal atresia in 4(11.4%), Intestinal stenosis in 5(14.3%), Hirshsprung disease in 4(11.4%) and Exomphalos in 4 (11.4%).

Conclusion: Anorectal malformations were most common anomaly followed by persistent vitelline duct. Antenatal follow up and anomaly scan of all neonates should be done.

Keywords: Gastrointestinal anomaly, Neonates, congenital anomaly.

INTRODUCTION

Congenital anomalies, also called as birth defects, congenital malformations or congenital disorders are defined as structural or physical abnormalities present at birth which may be identified antenatally, at birth or in later life and may result in mortality or morbidity resulting in significant impact on patient, family, health care system and society[1-4]. The short and long term impact of congenital anomalies increases in developing countries, due to their under diagnosis and so prevalence is under estimated due to poor reporting and recording of health statistics. There is also variation in the prevalence in different regions of the world due to racial and social differences[1]. According to WHO statistics on causes of child death from 2000-2013, 276000 neonates die within first 4 weeks of life due to congenital anomalies worldwide[5,6]. According to figures from south east region, 8-15% deaths in perinatal period and 13-16% deaths in neonatal period are due to congenital anomalies[7,8].

A fully functional gastrointestinal tract (GIT) is a complex organ which develops from simple digestive tract leading to complex series of events evolving from early embryonic period spanning till birth[9].

Major congenital malformations of gastrointestinal tract (CMGIT) are usually manifested in early neonatal period by signs of intestinal obstruction and may be life threatening[10]. Though CMGIT usually present in early newborn period but may present in later life or even in adulthood[9]. CMGIT can involve both upper and lower GIT. Among upper CMGIT involvement of esophagus like atresia, webs, fistula, vascular rings and duplication[9]. At the level of stomach congenital gastric outlet obstruction, pyloric stenosis and duplication while in duodenum, atresia, duplication and rotation are common[9]. Lower CMGIT include malrotation, intussusceptions, volvulus, fistula, imperforate anus, hirshsprung disease and anal atresia[7].

CMGIT vary among different ethnicities from as low as from 1% to as high as 45.2%[10-12].

Aim of our study is to determine the frequency and pattern of CMGIT in symptomatic newborns admitted at tertiary care center.

METHODS

This descriptive cross sectional study was done at National Institute of Child Health (NICH) Karachi over period of 6 months, from 1-8-17 to 31-1 -18. Ethical approval was taken from institutional ethical committee. All neonates from one to 28 days of life of either gender presenting to emergency department with CMGIT were enrolled. Neonates with other systemic congenital birth defects were excluded. Informed consent after explaining nature of the study was taken from parents or guardians. Detailed history and thorough physical examination was done. Confirmation of the diagnosis was made by imaging modalities as per needed like x-rays, ultrasonography, computed tomography and barium studies. All babies were managed after emergency department at Neonatal and Pediatric Surgery unit as per hospital protocol and surgical intervention was taken as per indication.

Statistical

RESULTS

There were 35 neonates enrolled with CMGIT. Out of these 16(45.7%) were male and 19(54.3%) were females. Mean age 9.4+3.82 (range 1-2) days. Mean weight was 2.5±0.98 (range 0.5-4.5) kg. Mean duration of symptoms was 5.8+1.72 (range 1-9) days.

Among the CMGIT, 12(34.3%) cases were of Anorectal malformation, persistent Vitelline duct was found in 6(17.1%), Intestinal Atresia in 4(11.4%), intestinal stenosis in 5(14.3%), Hirshsprung disease in 4(11.4%) and exomphalos in 4(11.4%).

Stratification of CMGIT was done in two age groups, between 1-9 days and > 9 days. There was significant difference in two age groups in intestinal stenosis, intestinal atresia, persistent vitelline duct and in anorectal malformations (p value <0.05), indicating these diseases more commonly present in early age groups, while there was no significant difference in exomphalos and in hirshsprung disease (p value <0.05) as shown in table II.

In stratification of gender no significant differences were found across all types of gastrointestinal anomalies i.e. (P≤0.05) as shown in table III.

Table 1: Demography And Pattern Of Cmgit N=35

Mean age+SD (range) days	9.4+3.82 (range 1-20)
Mean weight+SD (range) kg	2.5±0.98 (range 0.5-4.5)
Mean duration of symptoms+SD (range) days	5.8+1.72 (range 1-9)
Anorectal malformation N(%)	12 (34.3%)
Persistent vitelline duct N(%)	6 (17.1%)
Intestinal atresia N(%)	4(11.4%)
intestinal stenosis N(%)	5(14.3%)
Hirshsprung disease N(%)	4(11.4%)
Exomphalos N(%)	4 (11.4%).

Table 2: Stratification Of Cmgit According To Age Groups

Gastrointestinal Anomalies		Age groups (in days)		p-value
		1-9 days	>9 days	
Exomphalos	Yes no	5.6% 53.7%	2.3% 38.4%	0.337
Hirschsprung's Disease	Yes no	5.1% 54.2%	4.0% 36.7%	0.793
Intestinal Stenosis	Yes no	9.0% 50.3%	1.7% 39.0%	0.019
Intestinal Atresia	Yes no	9.6% 49.7%	0.6% 40.1%	0.001
Persistence of Vitelline Duct	Yes no	13.6% 45.8%	1.7% 39.0%	0.001*
Ano-Rectal Malformation	Yes no	22.6% 36.7%	7.3% 33.3 %	0.004*

Table 3: Stratification Of Cmgit According to Gender N=35

Gatrontestinal anomalies		Gender		P value
		Male	Female	
Exomphalos	Yes No	4.6% 40.7%	3.4% 51.4%	0.349
Hirshsprung disease	Yes No	5.1% 40.1%	4.0% 50.8%	0.352
Intestinal atresia	Yes No	5.6% 39.5%	4.5% 50.3%	0.239
Intestinal stenosis	Yes No	6.2% 30.9%	4.5% 50.3%	0.352
Persistent vitelline duct	Yes No	7.3% 37.9%	7.9% 46.9%	0.783
Anorectal malformations	Yes No	15.3%	14.7%	0.315

Table 4: Stratification Of Cmgit According To Duration Of Symptoms n=35

Gatrontestinal anomalies		Duration of symptoms		P- value
		1-6 days	>6 days	
Exomphalos	Yes No	1.1% 45.2%	6.8% 46.9%	0.012
Hirshsprung disease	Yes No	1.7% 44.6%	7.3% 46.3%	0.020
Intestinal stenosis	Yes No	2.3% 44.1%	8.5% 45.2%	0.019
Intestinal atresia	Yes No	0.6% 45.8%	9.6% 44.14%	0.0001
Persistent vitelline duct	Yes No	2.8% 43.5%	12.4% 41.2%	0.002
Anorctal malformations	Yes No	15.6% 40.7%	24.3% 29.4	0.0001

Table 5: Stratification Of Cmgit According To Weight Group n=35

Gatrontestinal anomalies		Weight groups (in kg)		P- value
		0.5-2.5	>2.5	
Exomphalos	Yes No	6.8% 45.2%	1.1% 46.9%	0.008
Hirshsprung disease	Yes No	7.3% 44.6%	1.7% 46.3%	0.014
Intestinal stenosis	Yes No	18.6% 44.3%	2.3% 44.8%	0.016
Intestinal atresia	Yes No	17.9% 44.1%	2.3% 45.8%	0.021
Persistent vitelline duct	Yes No	11.3% 40.7%	4% 44.1%	0.013
Anorctal malformations	Yes No	25.6% 26.6%	4.5% 43.5%	0.0001

A significant difference was noted between duration of symptoms and all the types of gastrointestinal anomalies i.e. (P≤0.05) as shown in table IV.

Significant differences were found between weight and all the types of gastrointestinal anomalies i.e. (P≤0.05) indicating most of the congenital anomalies were associated with weight <2.5kg as shown in TABLE V.

DISCUSSION

Congenital malformations can result in long term sequel, disability and is one the emerging health problems causing significant effect on family, society and health care system. In the study in 6 months, 35 cases of CMGIT were enrolled signifying disease burden at tertiary care hospital, reason may be that NICH is only government sector tertiary care hospital in Karachi providing free pediatric surgery services and referrals are also received from whole of the province.

It was found that CMGIT more commonly involved females (54.3%). This is contradictory to other studies where they found more prevalence of congenital anomalies in males[1,5].

Among CMGIT, most commonly found was anorectal malformation seen in 34.3% cases. Similar results have been found from most of the other studies. From Pakistan, Mahmud et al has reported as imperforate anus as 54% and anorectal stenosis as 14% cause of all GIT malformations. And shamim et al has reported anorectal malformation as 20% of GIT malformations. In studies from India it has been reported as anorectal malformation as most common GIT anomaly making 20% of CMGIT[1,5,12]. In study by Ochaga et al has shown anorectal malformation as 66% of GIT anomalies[13].

In the study second most common GIT anomaly in the study was persistent vitelline duct found in 17 % of cases followed by intestinal atresia and intestinal stenosis in 11%and 14% respectively. Shamim et al has reported as intestinal atresia /stenosis as second most common GIT anomaly after anorectal malformation[12] .Agarwal et al has reported as neonatal intestinal obstruction as 15% of total congenital anomalies and Ochaga et al has reported jejuna atresia as 9.5% of GIT anomalies[5,13] .

Other common CMGIT were exomphalos also called as omphalocele and hirshsprung disease. Ochaga et al has reported after anorectal malformation and intestinal atresia other CMGIT were gastroschisis, omphalocele[13]. Another study from India has reported as Gastroschisis, Imperforated anus, Ileocecal atresia and Tracheoesophageal fistula as common causes of CMGIT[14] .

A study from Nigeria has reported omphalocele as 50% case of CMGIT[15,16].

Other Studies from different parts of the World have reported as GIT anomalies constitute 20.39% of total congenital malformations and among which important are Tracheo-oesophageal fistula, Diaphragmatic hernia, Tongue-tie, Cleft lip, Cleft palate ,Imperforate anus ,Gastrochisis, Omphalocele, and Duodenal atresia[17,18].

We found persistent vitelline duct as second most common cause of MCMGIT, in contradictory to other studies where it has been reported second most important CMGIT as intestinal atresia or trachea-esophageal fistula/ esophageal atresia[1,5,12,17,18].

This study emphasizes burden of CMGIT. Though there is rise in burden of congenital anomalies, still there is lack of appropriate preventive measures and antenatal diagnostic measures in developing country like in Pakistan.

There is need for good antenatal care and antenatal diagnosis so that treatment plan may be made before delivery and appropriate intervention may be made immediately after delivery to reduce morbidity and complications of surgery which may be increased if surgery is delayed, like development of perforation or sepsis.

WHO has emphasized as congenital anomalies burden is rising due to improved health care facilities and focus has to be made on early recognition and improvement in surveillance grants

to improve understanding of risk factors and improvement of outcome.

CONCLUSION

Anorectal malformations were most common anomaly followed by persistent Vitelline duct. Antenatal follow up and anomaly scan of all neonates should be done for efficient treatment plan to improve outcome of such neonates. Nationwide surveillance is needed to recognize burden of CMGIT.

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