Congenital malformations of gastrointestinal tract in children aged less than 5 years of age: A retrospective and prospective study

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Abstract

Introduction: The spectrum and prevalence of congenital abnormalities of gastrointestinal tract vary in different populations depending upon the environment influences, genetic predisposition and prevalent socioeconomic factors. The aim of this study is to review and analyse the cases registered in the last five years in our centre and study their clinical presentation, spectrum, prevalence and also the histopathological picture. Methods: This cross-sectional descriptive study was taken up in Niloufer Hospital, Hyderabad. This was a 3 year retrospective and 2 year prospective study. All Pediatric surgical specimens and biopsies of children aged less than 5 years received during a period of 5 years were included in the study. Results: During the study period, total 2842 pathological specimens were reviewed, out of which 161 (5.67%) cases were related to GI tract Anomalies. Hirschsprung’s disease was the commonest accounting for about 47.2% of all the anomalies. Together Hirschsprung’s disease, Omphalo mesenteric duct remnants, intestinal atresia’s, duplication cysts and meconium ileus contributed to greater than 75% (¾) of the cases. Most of the cases were less than 1 year of age (71%). Male predominance was noted with a male to female ratio of 2.74:1. Conclusion: Congenital and developmental abnormalities of gastro intestinal tract are not very uncommon in the Indian sub continent and these are a cause of significant mortality and morbidity in pediatric age group. This study has highlighted the prevalence and types of congenital anomalies seen in our Geographical area.

Keywords: Congenital anomaly, Prevalence, Types, GIT anomalies, Children less than five years of age, Histopathological examination.

Material and Methods

Study Design- This Cross sectional descriptive study was taken up in Niloufer Hospital, Hyderabad. This is a
3 year retrospective and 2 year prospective observational study.

**Setting** - The present study was done to evaluate the prevalence and pattern of congenital malformations of the gastrointestinal tract based upon the pathological specimen sent. It was taken up in Niloufer Hospital which is a pediatric referral centre for medical and surgical diseases.

**Inclusion Criteria** - Specimens of children less than 5 years only were included in the study.

**Participants** - The cases that presented with various causes of constipation and intestinal obstruction such as Hirschsprung’s disease, Omphalo mesenteric duct abnormalities, Intestinal atresia, Gut malrotation, Intussusceptions, webs/stenosis and various enteric cysts were examined by surgical biopsies and specimens. Several sections were taken from these cases for the demonstration of H & E.

**Variables**: Data sheet was completed for each patient detailing the Age, Sex, Clinical presentation, Pertinent Investigations, Biopsy site and Histological diagnosis.

**Data Source**: Congenital malformations of the gastrointestinal tract reported in 3 years from August 2006 to July 2009 were retrieved from the pathology register and the histopathology slides were studied. This constituted the retrospective study. The clinical details including the radiological and intra-operative findings were retrieved from the hospital registers.

The prospective study constituted study of pathological specimens diagnosed as congenital gastrointestinal lesions received in the department of pathology of Niloufer hospital during the period from August 2009 to July 2011.

All the clinical details including radiological findings as well as intra-operative findings were recorded. Several sections were taken from these cases for the demonstration of H & E. The blocks were routinely fixed and processed.

**Study Size** - This study included all gastrointestinal tracts pathological specimens of children aged less than 5 years, received in the Department of Pathology, Niloufer Hospitals, Hyderabad.

**Quantitative Variables** - The various congenital anomalies were analysed and grouped according the age, sex and biopsy sites and further analysis was done.

**Statistical methods** - Data was entered into excel data sheet and appropriate statistical analysis was performed.

**Results**

From August 2006 to July 2011, the total number of Pediatric surgical specimens and biopsies received at pathology department of Niloufer hospital, Hyderabad for a period of 5 years were 2842.

Out of these 161 (5.67%) cases were related to congenital and developmental anomalies of gastrointestinal tract in children aged less than 5 years.

Among the GIT anomalies, Hirschsprung’s disease was the commonest accounting for about 47.2% of all the anomalies. This was followed by Omphalo mesenteric duct remnants which were accounting for about 13.7% of the cases.

Intestinal atresias were seen in about 8% of the cases and Duplication cysts were responsible for another 6.5% of cases.

Most of the cases diagnosed to have GIT anomalies were less than 1 year of age (71%). The age group ranged from 1 day-5 years with 36% presenting before the age of one month. In the present study, male predominance was noted with a male to female ratio of 2.74:1.

Among those diagnosed to have Hirschsprung’s disease, male predominance was noted with a male to female ratio of 2.8:1, which was similar to the overall sex ratio.

More than 2/3 of the cases diagnosed were younger than 1 year of age, with around 41% between the ages of 1 month-1 year.
Table-1: Distribution of GIT anomalies.

| S.No | Congenital Anomaly                  | Age Range  | Male | Female | Total | %  |
|------|------------------------------------|------------|------|--------|-------|----|
| 1    | Hirschsprung’s Disease             | 0-5 yrs    | 56   | 20     | 76    | 47.2|
| 2    | Omphalomesenteric duct remnants    | 0-5 yrs    | 17   | 5      | 22    | 13.7|
| 3    | Intestinal Atresias               | 0-1 year   | 8    | 5      | 13    | 8.1 |
| 4    | Meconium Ileus                    | 0-1 month  | 2    | 1      | 3     | 1.9 |
| 5    | Duplication Cysts                 | 0-5 yrs    | 6    | 4      | 10    | 6.2 |
| 6    | Mesenteric Cysts                  | 0-5 yrs    | 2    | 1      | 3     | 1.9 |
| 7    | Web/Stenosis                      | 0-5 yrs    | 3    | 1      | 4     | 2.5 |
| 8    | Malrotation                       | 0-1 month  | 1    | 0      | 1     | .6  |
| 9    | Ileocecal intussusceptions        | 1 month-5 yrs | 2   | 3      | 5     | 3.1 |
| 10   | Polyps                             | 2-5 years  | 3    | 0      | 3     | 1.9 |
| 11   | Omphalocele/gastrochisis          | 0-1 month  | 2    | 0      | 2     | 1.2 |
| 12   | Congenital diaphragmatic hernia   | 0-1 month  | 0    | 1      | 1     | .6  |
| 13   | Normal bowel segments             | 0-5 years  | 10   | 0      | 10    | 6.2 |
| 14   | Inadequate biopsy                 | 0-5 years  | 4    | 2      | 6     | 3.7 |
| 15   | Others                             | 0-5 years  | 2    | 0      | 2     | 1.2 |

Among the patients with Intestinal atresia (13), the commonest location was Ileal atresia.

Table-2: Regional and sex distribution of Intestinal atresia.

| Type of Atresia                     | Males | Females | Total |
|-------------------------------------|-------|---------|-------|
| Ileal Atresia                       | 5     | 2       | 7     |
| Jejunal Atresia                     | 2     | 3       | 5     |
| Colonic Atresia                     | 1     | 0       | 1     |
| **Total**                           | **8** | **5**   | **13**|

Among the 22 patients diagnosed to have Omphalomesenteric duct remnant, 20 of them were diagnosed to have Meckels diverticulum with its ensuing complications. One each was diagnosed to have a Persistent vitellointestinal duct and Omphalomesenteric duct/cyst. Compared to the overall group, significant male predominance was noted here (3.4:1) except for Omphalomesenteric duct/cyst.

Most of the cases of omphalomesenteric duct remnants were diagnosed beyond the age of one month (> 90%), with around 45% diagnosed at greater than one year of age. Those who were diagnosed after 1 year of age were exclusively male children.

Table-3: Age and sex distribution of Omphalomesenteric duct remnants.

| Age            | Males | Females | Total |
|----------------|-------|---------|-------|
| 0-1 Months     | 1     | 1       | 2     |
| >1-12 Months   | 6     | 4       | 10    |
| >1-5 Years     | 10    | 0       | 10    |
| **Total**      | **17**| **5**   | **22**|
Similar to the patients with Omphalomesenteric duct remnants, enteric duplication cysts were more common in children aged greater than one month of age (70%), with 40% greater than one year of age. Male predominance was also noted in this group except for the patients presenting before age of one month, where female predominance was noted. Ileum (5 (50%)) was the commonest site of enteric duplication cysts followed by gastric and jejunum.

Table-4: Age and sex distribution of enteric duplication cysts.

| Age         | Males | Females | Total |
|-------------|-------|---------|-------|
| 0-1 Months  | 1     | 2       | 3     |
| >1-12 Months| 2     | 1       | 3     |
| >1-5 Years  | 3     | 1       | 4     |
| Total       | 6     | 4       | 10    |

Discussion

The incidence of 5.67% noted in the present study shows that Congenital and developmental abnormalities of gastrointestinal tract are not very uncommon in the Indian sub continent. In a study by Malla BK et al at Maternity Hospital, Khatmandu medical college, the incidence of congenital malformations among newborn was 0. 36 per 100 live births. 17.34% of total anomalies were related to the Gastrointestinal Tract [4].

Table-5: Distribution of anomalies in various studies.

|                  | Asindi et al[5]                          | Ahn et al[6]                          | Present Study          |
|------------------|------------------------------------------|---------------------------------------|------------------------|
| 1                | Imperforate anus (44.8%)                 | Congenital megacolon (20.0%)          | Hirschsprung’s Disease (47.2%) |
| 2                | Tracheosophageal fistula/atresia (24.1%) | Imperforate anus (18.0%),             | Omphalomesenteric duct remnants (13.7%) |
| 3                | Intestinal atresia (21.3%)               | Congenital hypertrophic pyloric stenosis (16.4%) | Intestinal Atresias (8.1%) |
| 4                | Hirschsprung’s disease (8%)              | Congenital anomalies of intestine (11.6%), | Duplication Cysts(6.2%) |
| 5                | Stenosis (1.7%)                          | Congenital anomalies of hepato-biliary system(10.0%) | Ileocecal intussusception (3.1%) |
| 6                | -                                        | Meckel's diverticulum (7.2%),         | Web/Stenosis (2.5%)     |
| 7                | -                                        | Situs inversus (4.4%)                 | Mesenteric Cysts (1.9%) |
| 8                | -                                        | Congenital diaphragmatic hernia (2.4%), | Meconium Ileus (1.9%) |
| 9                | -                                        | Congenital anomalies of cecum (2.4%)  | Polyps (1.9%)           |
| 10               | -                                        | Esophageal diverticulum (2.4%),       | Omphalocoele/gastrochisis (1.25) |

In our study Hirschsprung’s disease was the commonest accounting for about 47.2% of all the anomalies, unlike other studies. Probably the Geographical, Ethnic and Genetic differences were the reasons for the differences noted in the distribution of congenital anomalies. In our study more than 2/3 of the cases of Hirschsprung disease diagnosed were younger than 1 year of age, with around 41% of them between the ages of 1 month - 1 year. 29% were diagnosed between ages of 1-5 years. Male predominance was noted with a male to female ratio of 2.8: 1. This was similar to the available literature [7,8].
Table-6: Prevalence of Intestinal Atresias in various studies.

| Study          | Study Group                                      | Incidence/Prevalence       |
|----------------|--------------------------------------------------|----------------------------|
| Asindi et al[5]| Infants admitted to NICU, Aseer hospital, Saudi Arabia | 21.3%                      |
| Francannet et al[9] | Central eastern French Registry                     | 2.25 cases per 10,000 live births |
| Martinez-Frias et al[10] | Latin America & Spain Registry                      | 1.13 cases per 10,000 live births |
| Present study  | Pediatric Inpatients, Niloufer hospital Hyderabad, India | 8.1%                      |

In Intestinal atresia, the commonest location was Ileum followed by jejunum and colon. In most studies, jejunoileal atresias seem to be more common than duodenal atresias and colonic atresias accounted for least number of cases [9,10,11].

In the present study Omphalomesenteric duct remnants were seen in 13.7% (22 cases) of the patients. Among the omphalomesenteric duct remnants, Meckels diverticulum is the commonest anomaly and in the present study it was seen in 12.47% of the cases (20 cases). Although no sex-based difference was reported in studies that evaluated this condition as an incidental finding during operations or autopsies, males are as much as 3-4 times more prone to complications than females [6].

In the present study compared to the overall group, significant male predominance was noted here (3.4:1). Most of the cases of omphalomesenteric duct remnants in the present study were diagnosed beyond the age of one month (> 90%), with around 45% diagnosed at greater than one year of age.

In the present study Duplication cysts were seen in 6.2% (10 cases). Gastrointestinal duplications are observed in 1 of every 4500 autopsies, predominantly in white males.

Table-7: Distribution of intestinal duplication cysts in various studies.

| Location         | Present study | Nagar et al[12] | Iyer CP et al[13] | Abdur-Rahman et al[14] |
|------------------|---------------|------------------|-------------------|------------------------|
| Oesophago-Gastric| 2             | 1                | 3                 | 0                      |
| Duodenal         | 1             | 0                | 0                 | 0                      |
| Jejunum- ileum   | 7             | 6                | 12                | 4                      |
| Caecum-Colon     | 0             | 2                | 7                 | 3                      |
| Rectum           | 0             | 0                | 5                 | 0                      |
| Total            | 10            | 9                | 27                | 7                      |

Similar to the available literature, Ileum was the most common site of enteric duplication cysts [12,13,14]. Enteric duplication cysts were more common in children aged greater than one month of age (70%), with 40% greater than one year of age. Male predominance was also noted in this group except for the patients presenting before the age of one month, where female predominance was noted.

In the present study Intussusception was seen in 5 cases (3.1%). Its estimated incidence is approximately 1 case per 2000 live births [7]. In the present study the male to female ratio was 1.5:1, In the literature, the male-to-female ratio is approximately 3:1[7,15,16].

With advancing age, gender difference becomes marked; in patients older than 4 years, the male-to-female ratio is 8:1[7]. Intussusception is the most common cause of intestinal obstruction in infants between 6 and 36 months of age. Intussusception is less common before three months and after six years of age. Approximately 60% of children are younger than one year of age, and 80% are younger than two years of age [15]. In the present study 80% of the children presenting with Intussusceptions were younger than 1 year of age.
In the present study, Colorectal polyps were seen in 3 cases corresponding to 1.9% of the patients. All the children who presented with polyps were greater than 2 years of age and all of them were male. In study by Latt TT et al, the mean age of presentation was 5.6 years and about two thirds of the patients were under the age of 5 years [17]. Similar in another study by Prakash mandhan et al the mean age at presentation was 5.2 years, with a male predominance [18]. In the present study, all those diagnosed to have polyps were male. Similarly Prakash mandhan et al and Wajeehuddin et al in their studies also documented a significant male predominance (4:1)[18,19].

Mesenteric cysts are rare; the incidence is about 1 per 140,000 general hospital admissions and about 1 per 20,000 pediatric hospital admissions [20,21]. In a study from Egleston Children’s Hospital at Emory University from 1965-1994, showed a prevalence of about 1 case per 11,250 admissions [22]. In the present study mesenteric cysts were seen in 3 cases which contributed for about 1.9% of all cases.

The age range in present study varied between 0-5 years. In a study by Hebra et al 75% of cases occurred in children younger than 5 years of age [23]. In another study by from Bliss et al approximately one third of cases occur in children younger than 15 years of age [24].

Limitations- As it is a tertiary care hospital or referral centre, prevalence calculated may be higher than the general population in this hospital-based study. Hence, the data cannot be projected to the general population, for which population-based studies are necessary.

Conclusions

Congenital and developmental abnormalities of gastrointestinal tract are not very uncommon in the Indian subcontinent and these are a cause of significant mortality and morbidity in pediatric age group. This study has highlighted the prevalence and types of congenital anomalies seen in our locality. Probably the Geographical, Ethnic and Genetic differences were the reason for the differences noted in the distribution of congenital anomalies.

Though radiodiagnosis is the preliminary diagnostic modality, but gross and histopathologic examination are essential to confirm the diagnosis. Especially in the case of Hirschsprung’s disease histopathological examination is the diagnostic modality to ensure early surgical management.

Funding: Nil, Conflict of interest: None initiated, Permission from IRB: Yes

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How to cite this article?

Reddy V. B, Ramnani M, Reddy J. B. Congenital malformations of gastrointestinal tract in children aged less than 5 years of age: A retrospective and prospective study. Trop J Path Micro 2017;3(1):55-61.doi: 10.17511/jopm.2017.i1.10.