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Research Paper

# CONGENITAL GASTROINTESTINAL ANOMALIES AND THEIR ASSOCIATIONS TO GENETIC DISORDERS

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**ABSTRACT** **Congenital anomalies of the digestive tract are an important part of congenital anomalies as they constitute one of the main causes of mortality in children. The aim of this study was to evaluate the most common digestive anomaly associated with genetic disorders. We also sought to ascertain the most frequent genetic disorder in our patients and its connection to mortality, the average age of diagnosis, prenatal diagnosis, gestacional age and associations with other organ anomalies. The study was conducted in two phases and includes a total of 273 children diagnosed with Congenital anomalies of the digestive tract, presented at the Pediatric Intensive Care Unit in UHC "Mother Teresa". The first phase, retrospective, includes 137 patients during the period January 2006 – December 2010. The second phase of the study, prospective, includes 136 patients from January 2011 - March 2015. The most frequent digestive anomaly in patients with genetic disorders was Anal atresia (39.40%), followed by Omphalocele and Intestinal atresia (15.10%). Down syndrome was the most frequent genetic disorder,9 cases (3.30%). It is important to identify cases associated to genetic disordes because they condition patient's managment and prognosis. Anomalies that are often associated with other defects have shown early clinical symptoms and require more time and effort for correction.**

**KEYWORDS** :Congenital gastrointestinal anomalies, newborn babies, genetic

## INTRODUCTION

The gastrointestinal tract may be subject to a variety of congenital abnormalities (i.e. those present at birth) that arise during embryological development. Specific patterns of malformations of the gastrointestinal tract include abnormal lumenisation (stenoses and atresias), duplications, abnormal and fixation, abdominal defects and a variety of others associated with persistence of embryonic structures (e.g. Meckel's diverticulum), or abnormal formation of specific regions of the gastrointestinal tract (e.g. microgastria) or its cellular components (e.g. nerves in Hirschsprung's disease). These disorders primarily result in symptoms of intestinal obstruction, effects on surrounding structures or of associated anomalies. [1]

They frequently manifest with feeding difficulties, distention, and emesis at birth or within 1 or 2 days.[2] The clinical symptoms varies depending on the pathology, the age of diagnosis and associated anomalies. They may be from mild to severe with, recurrent abdominal pain, intestinal obstruction, dehydration, malnutrition, malabsorption/ diarrhea, peritonitis/septic shock, solid food intolerance, common bile duct obstruction, abdominal distention, and failure to thrive.[3] Some congenital GI malformations, such as malrotation, have a very good outcome, whereas others, such as congenital diaphragmatic hernia, have a poor outcome, with a relatively high mortality rate of 10 to 30%.[2]

Early clinical recognition of these disorders is essential to minimise complications and allow the early institution of appropriate therapies.

Although surgery is the commonest intervention early treatment must include adequate resuscitation and stabilisation of the child prior to definitive surgery. Many of these abnormalities are associated with other congenital Paper ID: ART20183796 anomalies or genetic syndromes and disease making the clinical symptoms and the management more complex and difficult. [1]

A genetic disorder is a disease caused in whole or in part by a change in the DNA sequence away from the normal sequence. Genetic disorders can be caused by a mutation in one gene (monogenic disorder), by mutations in multiple genes (multifactorial inheritance disorder), by a combination of gene mutations and environmental factors, or by damage to chromosomes, changes in the number or structure of entire chromosomes, the structures that carry genes (chromosome disorders). [4]

The incidence of disease syndromes and genetic defects in patients with malformations is much higher than in the general population.[5] Since genetic syndrome is seen to have an important impact in patient's life, in the U.S. and many other countries there are created healthcare programs for newborn genetic screening that identify treatable genetic disorders in newborn infants. Early intervention to treat these disorders can eliminate or reduce symptoms that might otherwise cause a lifetime of disability. [6]

Through our study we sought to evaluate the connection between genetic disorders and congenital gastrointestinal anomalies in newborns who were hospitalized in our Intensive Unit Care.

## 2. Material and methods

The study was conducted in two phases and includes a total of 273 children diagnosed with Congenital anomalies of the digestive tract, presented at the Pediatric Intensive Care Unit in UHC "Mother Teresa". The first phase of the study is case-control type or otherwise known as retrospective study.

It includes 137 patients, who were admitted to the Pediatric Intensive Care Unit during the period January 2006 – December 2010 with the main diagnose "Congenital gastrointestinal anomalities".

The second phase of the study is prospective type and includes cases presented from January 1, 2011 to March 1, 2015, where patients have been followed for at least 1 month.

The second phase of the study, prospective, includes 136 patients.

### Statistical analysis

Continuous data were presented in average value and in standard deviation. Discrete data were presented in absolute value and in percentage. The correlation between the two variables was analyzed by the Kendal's tau correlation coefficient.

Presentation of the data was done through simple and composite tables and graphs of different types.

The statistical analysis was carried out through the statistical package SPPS 19.0 (Statistical Package for Social Sciences Inc., Chicago II USA) and Microsoft Excel.

Significant values of p=0.01 were considered.

## 3. Results and discussion

In our study are included 273 patients with congenital digestive anomaly as above in table 1:

**Table 1: Congenital digestive anomaly frequency**

| *Congenital digestive anomaly.* | *No* | *(%)* |
|---|---|---|
| **Anal atresia** | 59 | 21.60% |
| **Omphalocele** | 38 | 13.90% |
| **Intestinal atresia** | 50 | 18.30% |
| **Meconuim ileus** | 3 | 1.10% |
| **Hirschsprung's disease** | 8 | 3.00% |
| **Duodenal atresia** | 20 | 7.30% |
| **Esophageal atresia** | 48 | 17.60% |
| **Biliary atresia** | 6 | 2.20% |
| **Diaphragmatic hernia** | 26 | 9.50% |
| **Intestinal malrotation** | 3 | 1.10% |
| **Common mesentery** | 6 | 2.20% |
| **Congenital megacolon** | 2 | 0.70% |
| **Hypertrophic pyloric stenosis** | 4 | 1.50% |
| **Total** | 273 | 100.00% |

The most frequent anomaly in our study is anal atresia (21.60%), followed by intestinal atresia (18.3%) and esophageal atresia (17.60%).

In a total of 273 cases diagnosed with Congenital gastrointestinal anomaly, 33 of them (12.10%) had also a genetic syndrome or disease. (Table 2)

**Table 2: Frequency of genetic disorders in our patients**

| *Genetic disorder* | *No.* | *(%)* |
|---|---|---|
| **Present** | 33 | 12.10% |
| **Not present** | 240 | 87.90% |
| **Total** | 273 | 100% |

From 33 cases presents with genetic disorder the most common digestive anomaly was found Anal atresia (39.4%).In our study biliary atresia, diaphragmatic hernia, intestinal malrotation, common mesentery, congenital megacolon and hypertrophic pyloric stenosis have not been associated with genetic disorders.

**Table 3: Frequency of digestive anomalies associated with**

| *Congenital digestive anomaly.* | *No* | *(%)* |
|---|---|---|
| **Anal atresia** | 13 | 39.40% |
| **Omphalocele** | 5 | 15.10% |
| **Intestinal atresia** | 5 | 15.10% |
| **Meconuim ileus** | 3 | 9.10% |
| **Hirschsprung's disease** | 3 | 9.10% |
| **Duodenal atresia** | 2 | 6.10% |
| **Esophageal atresia** | 2 | 6.10% |
| **Biliary atresia** | 0 | 0% |
| **Diaphragmatic hernia** | 0 | 0% |
| **Intestinal malrotation** | 0 | 0% |
| **Common mesentery** | 0 | 0% |
| **Congenital megacolon** | 0 | 0% |
| **Hypertrophic pyloric stenosis** | 0 | 0% |
| **Total** | 33 | 100.00% |

The most frequent genetic disorder seen in our study was Down syndrome, with 9 cases (3.30%), followed by Cystic fibrosis, 5 cases (1.80%). (Table 4)

**Table 4: Genetic dosorders by their frequency**

| *Congenital digestive anomaly.* | *No* | *(%)* |
|---|---|---|
| S.Down | 9 | 3.30% |
| Cystic fibrosis | 5 | 1.80% |
| VACTERL | 1 | 0.37% |
| OEIS Complex | 1 | 0.37% |
| Trizomy 18 | 1 | 0.37% |
| Noonan | 1 | 0.37% |
| Without genetic disorder | 240 | 87.90% |
| Others | 15 | 5.50% |
| Total | 273 | 100% |

In our study was found a significative relation (p<0.01) betweet genetic disorders and other associated anomalies in patients with congenital digestive anomalies. Most frequent associated anomalies were found heart (20.50%), pulmonary (7.70%), abdominal (5.50%) and renal diseases (4.0%). 23 (69.70%) of 33 cases with Congenital digestive anomaly and genetic disorder, also had another associated anomaly. (Table 5). In another study made in Hospital Infanta Cristina, SES, Spain was considered that it is important to identify any associated anomalies, especially heart disease, craniofacial anomalies and other gastrointestinal malformations, because they condition the patient's management. The knowledge of these patients is essential for correct treatment. [5]

**Table 5: Associated anomalies**

| **Genetic disorder** | **Associated anomalies** | **No other anomaly** | **Total** |
|---|---|---|---|
| **Present** | **23** | **10** | **33** |
| **69.70%** | **30.30%** | **100%** | |
| **Not present** | **97** | **143** | **240** |
| **40.40%** | **59.60%** | **100%** | |

Also data from the literature showed that children with congenital heart disease have a much higher incidence of intestinal malformations than those with normal heart and that they frequently present with multiple malformations (chromosome aberrations or multiple organ lesions). This multiple malformation complex is particularly common in anorectal malformations where the incidence of congenital heart diseases is 9 to 14%, with predominance of VSD and tetralogy of Fallot. [7]

Another significant connection (p< 0.01) was found between the

presence of genetic disorders and the diagnostic age of patients with congental digestive anomalies. The more associated anomalies, the more simptoms are presented, and this is reflected in the age of diagnosis. In cases with genetic disorder the average age of diagnosis was about two times smaller than in cases with isolated congenital digestive anomaly. (Table 6)

**Table 6: Connection with average age of diagnosis**

| Genetic disorder | Avarage diagnostic age |
|---|---|
| **Present** | 2.24 days |
| **Not present** | 4.3 days |

Between mortality rate, prenatal diagnosis, gestacional age, birth weight and genetic disorders was not fund a significant correlation ($p > 0.01$).

## CONCLUSION

Identification of patients with genetic syndrome or disease plays a foundamental role in management and prognosis of patients diagnosed with Congenital digestive anomalies. Coexistence with multiple anomalies makes the clinic presentation more severe and more complex to achieve a better management.

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