Diyala Journal of Medicine

ORIGINAL RESEARCH
Published: 23 June 2022
Doi: https://doi:10.26505/DJM.22026360206

Pattern of Major Congenital Anomalies of the Gastrointestinal Tracts among Live Neonates at Al-Mukalla,Yemen

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Abstract

Background: Major Congenital anomalies of the gastrointestinal tract(GIT) is a life threating condidition usually diagnosed in the neonatal period.

Objective: To evaluate the prevalence of major congenital anomalies among live births at Al-Mukalla,Yemen.

Patients and Methods: A hospital-based, descriptive cross sectional study was conducted during 2 years period, from January 1st 2019 to December 31th 2020 and include live neonates aged ≤ 4 weeks who were admitted with major congenital gastrointestinal anomalies.

Results: The study include 62 neonates with major congenital GIT anomalies.The most common was imperforate anus in 41.9% followed by esophageal atresia with or without TEF in 24.2%, then hirschprung's disease in 19.4%. Males are more common (61.3%) than females (38.7%).The incidence is 7.7 per 10.000 newborn. Isolated major GIT anomalies occurs in 64.5%, while associated extra -GIT anomalies occurs in 35.5% , the most common was congenital heart diseases that occurs in 17.7% of cases .The majority neonates were term in 90.3%, with birth weight ≥ 2.5 kg in 88.7%. Most mothers were < 35 years in 67.7% ,with positive parental consanguinity occurs in 37% of cases.

Conclusion: High incidence of major Malformation were observed.

Keywords: Congenital, anomalies, Major, gastrointestinal tracts ,Neonates , Yemen.

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Received: 6 February 2022
Accepted: 30 March 2022
Published: 23 June 2022

Introduction

Congenital anomalies (CAs) are also known as birth defects, congenital disorders, or congenital malformations,congenital abnormalities, defined as structural or functional anomalies that occur during intrauterine life which can be detected prenatally, at birth, or in later life stages [1-3].Congenital anomalies are usually subdivided into minor CAs and major anomalies. A minor anomaly is defined as structural abnormality present at birth, having minimal effect on clinical function, while major CAs are conditions that are severe enough to reduce life expectancy or

ORIGINAL RESEARCH
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compromise normal function may lead to difficulty to survive or results in stillbirth/infant death . Major congenital malformations are divided into isolated anomalies (only one system involved) or complex anomalies (two or more Systems are involved)[4,5].Major CAs of the Gastrointestinal Tract(GIT) usually manifest in the neonatal period, with symptoms and signs of gastrointestinal tract obstruction and they can be life-threatening [6].

Major congenital gastrointestinal anomalies are classified and coded by World Health Organization (WHO) as part of the International Classification of Diseases-10 (ICD10) [7].

The risks of congenital malformations are various like maternal exposure to radiation, teratogenic drugs, maternal infectious diseases such as syphilis and rubella,genetic inheritance may have a role with autosomal recessive disorders are more common in males , most anomalies remaining of unknown etiology .Critical exposure periods to potentially teratogenic agents during pregnancy can vary by organ system or type of anomaly.However, first trimester is generally considered the highest risk period [8].

There are few studies identifying congenital GIT anomalies and to our best knowledge there is no published report on the prevalence and pattern of congenital malformations of the gastrointestinal tract in Yemen, The aim of this study was to assess the prevalence of major congenital gastrointestinal anomalies among live neonates at Al-Mukalla,Yemen.

Patients and Methods

A hospital-based, descriptive cross sectional study was conducted during 2 years period, from January 1st 2019 to December 31th 2020 at 3 hospitals the Modern Hadhramout hospital , University Hospital for Mother and Child Health and Al- Mukalla Maternity and Children Hospital .The first hospital is a private hospital while the last two hospitals is a governmental hospital. All of them are tertiary hospitals and have Neonatal intensive care units (NICU) .Most of cases are referred to the my co-author pediatric surgeon. The operation for the cases of major congenital gastrointestinal anomalies were done in the first two hospitals by a qualified pediatric surgeon . All these hospitals are located at al-Mukalla city a capital of Hadhramout governorate Yemen .

We use in this study the International Classification of Diseases-10 (ICD10), a classification of diseases by the World Health Organization (WHO) [7]For classification and coding of congenital gastrointestinal anomalies, this classification was adopted by several literatures [6,9,10].

Inclusion criteria

Live neonates aged ≤ 4 weeks who were admitted with major congenital gastrointestinal anomalies.

Exclusion criteria

1.Neonates with minor GIT anomalies .

2.Neonates with malrotation is a condition that includes a wide spectrum of disorders of intestinal fixation (rather than an actual malformation of the GI tract) [9].

3.Neonates with congenital diaphragmatic hernia , gastrosxis and omphalocele because these anomalies were due to diaphragm and abdominal wall defects.

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4.Neonates with congenital hypertophic pyloric stenosis (it is mentioned in the International Classification of Diseases-10 (ICD10 with code Q 40.0) it is not considered as a major GIT anomaly, because it is not severe enough to reduce life expectancy [7].

5.Neonates with incomplete or missed data.

All information among neonates with congenital gastrointestinal anomalies including history and physical examination were performed . Neonates with multiple congenital anomalies were grouped depending on whether those anomalies qualified as a specific syndrome or not.Diagnosis was confirmed by radiographic studies and surgical findings ,Echocardiography, X-ray imaging, cranial and abdominal ultrasonography were performed when required.

Statistical Analysis

The data were processed and analyzed by using Statistical Package for Social Sciences version 24 (SPSS Inc., Chicago, IL, USA) and the data was analyzed by proportion and percentage.

Results

The study include 62 neonate with major congenital GIT anomalies during the study period.Males are more common 38(61.3%) than females 24(38.7%).The most common major congenital GIT anomalies was imperforate anus in, 26 (41.9%) followed by esophageal atresia with or without TEF in, 15 (24.2%), then hirschprung's disease in , 12 (19.4%).The rest of findings were shown in Table (1) and Figure (1).

Table (1): Frequency and Sex distribution of major congenital gastrointestinal anomalies among neonates at Al-Mukalla, Yemen according to ICD 10 Classification (n= 62)

ICD 10 Classification	Gender		Total
	Male	Female	
Congenital malformation of the esophagus (Q39): Esophageal atresia with or without TEF*** (Q39.0/39.1)	9	6	15 (24.2%)
Congenital absence, atresia, and stenosis of small intestine (Q41) Duodenal Atresia (Q41.0) Jejunal Atresia (Q41.1)	4 1	3 -	7 (11.3%) 1(1.6%)
Congenital absence, atresia, and stenosis of large intestine (Q42) Imperforate Anus (Q 42.3)	16	10	26 (41.9%)
Other congenital malformations of the intestine (Q43) Hirschprung's disease (Q 43.1)	8	4	12 (19.4%)
Others Double rectum as part of teratoma	-	1	1(1.6%)
Total	38(61.3%)	24(38.7%)	62(100%)

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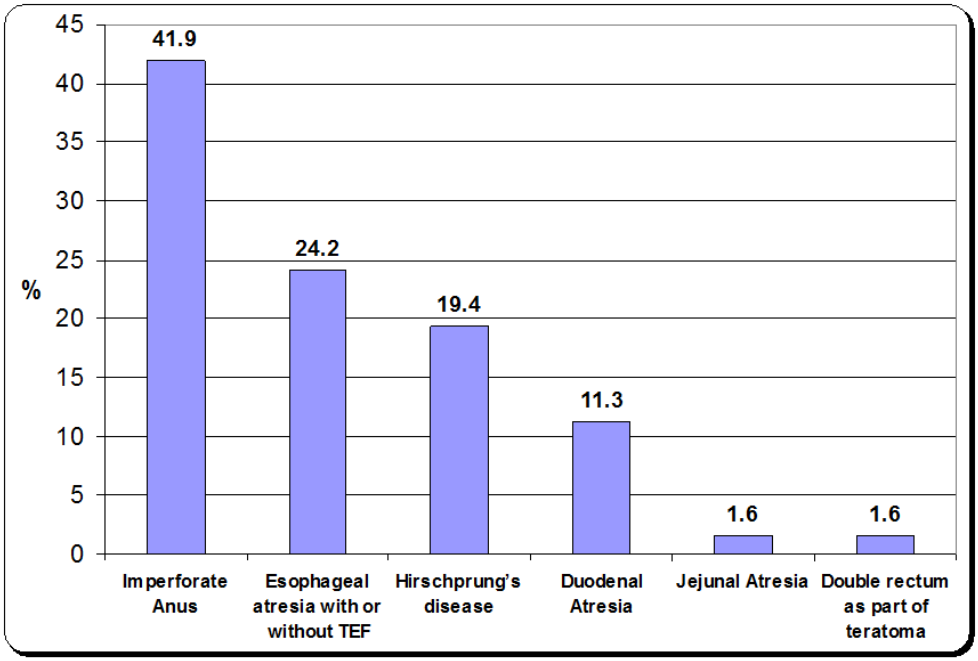

Figure (1): Frequency of major congenital gastrointestinal anomalies among neonates at Al-Mukalla, Yemen (n= 62)

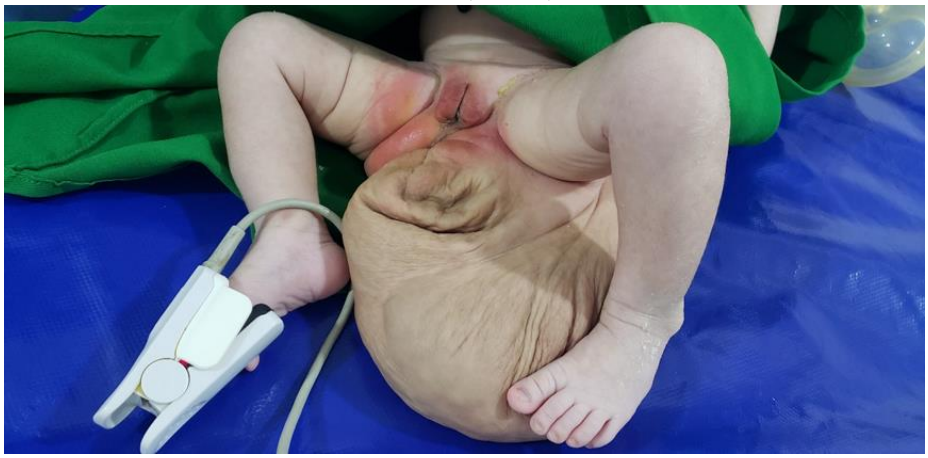

Figure (2): Neonate with double rectum as part of teratoma

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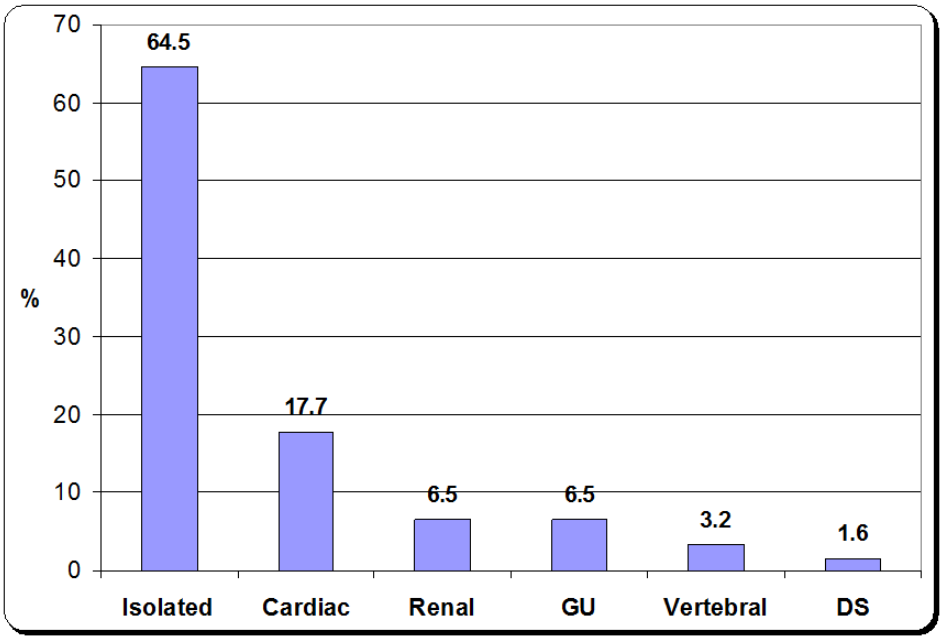

Figure (3): Frequency of associated extra- GIT major congenital anomalies among neonates at Al-Mukalla, Yemen (n= 62)

*GU: genitourinary, DS: Down syndrome

The majority neonates were term in 90.3%, with birth weight ≥ 2.5 kg in (88.7%) .And the majority of them (91.9 %) had vaginal delivery with cephalic presentations. While the rest of findings are shown in Table (2).

Table (2): Characteristics of neonates with of major congenital anomalies at Al-Mukalla, Yemen (n= 62)

Parameter	Category	Frequency	%
Gestational age	Preterm	6	9.7
	At term	56	90.3
Gender	Male	38	61.3
	Female	24	38.7
Birth weight	< 2.5 kg (SGA)	7	11.3
	≥ 2.5 kg (AGA)	55	88.7
Fetal presentation	Cephalic	57	91.9
	Breech	5	8.1
Mode of delivery	Vaginal delivery	50	80.6
	Caesarean delivery	12	17.4

Most mothers were < 35 years in (67.7% (of cases, and most families resides in urban areas in (74.2%).Most mothers were primigravida in (67.7%), positive parental consanguinity occurred in (37%) of cases.The rest of findings were shown in Table (3).

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Published: 23 June 2022
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Table (3): Parental characteristics of neonates with of major congenital anomalies at Al-Mukalla, Yemen (n= 62)

Parameter		Frequency	%
Maternal age	< 35 years	42	67.7
	≥ 35 years	20	32.3
Paternal age	< 45	55	88.7
	≥ 45	7	11.3
Gravida	Primigravida	42	67.7
	Multigravida	20	32.3
Antenatal visits	< 3 visits	48	77.4
	≥ 3 visits	14	22.6
Parental consanguinity	Yes	23	37
	No	39	63
Family residency	Urban	46	74.2
	Rural	16	25.8
Maternal occupation	Housewife	56	90.3
	Employed	6	9.7
Father occupation	Free worker	53	85.5
	Employed	9	14.5
History of abortion or still birth	Yes	8	13
	No	54	87

Discussion

Congenital malformations are one of the major causes of still-birth, neonatal death, and physical defects and disabilities around the world [11]. Al-Mukalla city is the capital of Hadhramout governorate and resides in it the tertiary hospitals where medical care of malformed neonate including surgical correction and cases were referred from Hadhramout and neighboring governorates as Shabowa and A- Mahra to Al-Mukalla city.

In this study there is 62 cases of major congenital GIT out of an estimated annual 40000 delivery at Hadhramout governorate during period of 2 years at Al-Mukalla district , accounting for an incidence of 7.7 per 10.000 newborn , this were in similar pattern with incidence from reported prevalence from the countries in the Middle East [12,13] and those from the Europe [14,15,16] varies between 10 and 15 per 10,000 live births. However, exact interpretations of prevalence are often difficult due to the differences in case ascertainment and study design.

In this study Males are more affected than females (61.3%) versus (38.7%) in every type of GIT anomaly in this study .The male preponderance are similar to several studies where congenital anomalies in general including GIT is more common in males than females [6,12,17,18-20].The explanation may be due to the fact that autosomal recessive disorders are more common in males.

The most common major congenital GIT anomalies was imperforate anus in 26 (41.9%) followed by esophageal atresia with or without TEF in (24.2%), Hirschprung's disease in 19.4%, duodenal atresia in (11.3%), while there is one case for both jejunal atresia and double rectum. A similar

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finding was observed in a previous study at Saudi Arabia [12] where imperforate anus was the most common in (44.8%) of cases followed by esophageal atresia with or without fistula in (24.1%), also a study in Ethiopia found the most common GIT anomalies was imperforate anus [2].

Similar findings is observed in other studies [21].in contradiction a study conducted at English Caribbean Countries found the most common GIT anomalies was esophageal atresia [6].

Isolated major GIT anomalies occurs in (64.5%), while associated extra -GIT anomalies occurs in (35.5%).The most common was congenital heart diseases that occurs in 17.7% of Cases. Similar finding was observed in other study were associated extra-GIT anomalies occurs in (22.7%)of Cases .The most common of it was genitourinary (GU) anomalies in (17.9%) followed by cardiac anomalies in(15.4%) [12].

A study in India , Ethiopia and Iraq found that the most common extra-GIT anomalies was craniospinal system anomalies [17,19,22] while a study in Nijeria found that the most common extr-GIT anomalies was GU anomalies [23,24].

In this study ,the majority of neonates were term in 90.3%,with prematures constitute (9.7%) .Birth weight was normal (≥ 2.5 kg) in 88.7%. Similar finding was observed in a study in Morocco where the majority of neonates with congenital anomalies were full term and with normal body weight where a prematures constitute 17.4% [8] a study at Nijeria recorded only 1.9% preterms [25].The observation that congenital anomalies including GIT were more common among full term and normal body weight is also observed in other studies [6,25-28].

The explanation for this finding is that affected premature neonates may have not survived to reach the tertiary hospital. Opposite to this a surprising finding in study at India found that majority of malformed neonates were pretem in 76.1% of cases with 68 % of malformed babies had birth weight <2.5 kg [17].

In this study, the age of most mothers were < 35 years in (67.7%) of cases, where the finding that congenital anomalies including GIT is more common among young mothers were observed in a similar studies [6 ,8,17,22]. The explanation for this may be due to the high number of structural, non-chromosomal anomalies that occurs at young age in contrast to the chromosomal anomalies in which commonly increases with increasing maternal age. Opposite to this finding in our study, other studies had reported a higher incidence of malformation in the newborn to mothers with maternal age > 35 years [29-31].

In this study most mothers were primigravida in (67.7%) where similar finding was reported with other studies [8],while other studies had reported a higher prevalence among multipara women [6,27,28] this may be partially explained due to variable genetic or ethnic differences.

In this study majority of the neonates (91.9 %) had vaginal delivery with cephalic presentations where similar finding was observed in other studies [2,8,28].This may be partially explained due to the fact that congenital GIT anomalies did not cause fetal distress or macrosomia and has no presentation in intrauterine life ,therefore

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most mothers can deliver as normal vaginal delivery.

Positive consanguinity was found in 37% of cases in a study at Iraq [22]they found that (21.98%) of families with congenital anomalies. There are contradicting studies whether positive consanguinity had a role in occurrence of congenital anomalies. Some studies showed that the consanguinity for malformed patients was high with no significant relationship [32-34] while positive consanguinity showed a significant relationship to congenital anomalies in other studies [28,35].

Due to lack of genetic investigations at Al-Mukalla city, genetic and chromosomal screening could not be performed in this study.

Genetic counselling and establishment of congenital malformation registry in Yemen is needed.

Conclusions

The most common major congenital GIT anomalies was imperforate anus in 41.9% followed by esophageal atresia with or without TEF in 24.2%.Males are more common to be affected than females 61.3% vs 38.7%.Further studies needed to clarify all types of congenital anomalies.

Limitations of the study

Some newborns whose congenital lesions present at birth may be missed due to poor facilities to reach to tertiary hospitals in Al-Mukalla either because of poverty or delay resulting in early neonatal death.

Silent congenital GIT anomalies may not discovered.

Recommendations

Genetic counseling and establishment of congenital malformation registry.

Source of funding: The current study was funded by our charges with no any other funding sources elsewhere.

Ethical clearance: The study protocol was conducted according to principles of the Declaration of Helsinki, as well as reviewed and approved by the Ethical Research Committee at Hadhramout University /College of Medicine.

Conflict of interest: Nill

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Diyala Journal of Medicine

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نمط انتشار التشوهات الخلقية الكبرى في الجهاز الهضمي في الأطفال حديثي الولادة الاحياء في مدينة المكلا – اليمن

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الملخص

خلفية الدراسة: التشوهات الخلقية الكبرى في الجهاز الهضمي تظهر الاعراض في الأطفال حديثي الولادة كعلامات انسداد الامعاء التي يمكن أن تكون مهددة للحياة.

اهداف الدراسة: لتحديد نمط انتشار التشوهات الخلقية الكبرى في الجهاز الهضمي في الأطفال حديثي الولادة الاحياء العناية المكثفة للأطفال في مدينة المكلا -اليمن .

المرضى والطرائق: هي دراسة مبنيه على بحث مستقبلي في المستشفى شمل فترة عامان من اول يناير عام ٢٠١٩ م الى ٣١ ديسمبر عام ٢٠٢٠ م ، و شمل كل الاطفال حديثي الولادة أقل أو يساوي ٤ أسابيع المرقدين في مستشفيات مدينة المكلا ، اليمن المصابين بتشوهات خلقيه كبرى في الجهاز الهضمي.

النتائج: الدراسة شملت ٦٢ من الاطفال حديثي الولادة المصابين بالتشوهات الخلقية الكبرى في الجهاز الهضمي ، و كان عددهم ٣٨ ذكر و ٢٤ أنثى و كان عدد الذكور أكثر من الاناث بنسبة ٦١،٣% . أغلب الحالات شيوعا كانت بسبب انسداد فتحة الشرج الخلقي بنسبة ٤١،٩ %، و بعدها أتت انسداد قناة المرء الخلقي بنسبة ٢٤،٢% ثم مرض هشبرنج الخلقي بنسبة ١٩،٤ %. كانت نسبة حدوث تشوهات الجهاز الهضمي الكبرى هي ٧،٧ حالة لكل ١٠،١٠٠٠ مولود. حصلت حالات تشوهات الجهاز الهضمي لوحدها بنسبة ٦٤،٥ % وكانت مشتركة مع تشوهات أخرى بنسبة ٣٥،٥ %. أغلب حالات الأطفال حديثي الولادة كانوا مكتملي النمو بنسبة ٩٠،٣ % و كان وزنهم طبيعي في ٨٨،٧ % من الحالات.أغلب الأمهات كان عمرهم أقل من ٣٥ سنة بنسبة ٦٧،٧ %. كانت هناك علاقة قرابة بنسبة ٣٧ % بين الأسر المصابين

الاستنتاجات: هناك حالات أصابة كبيرة بتشوهات الخلقية الكبرى في الجهاز الهضمي . يجب تفعيل نظام الأستشارة الأسرية ما قبل الزواج مع تفعيل برنامج أحصائي بحالات التشوهات الخلقية في اليمن

الكلمات المفتاحية: تشوهات الخلقية، التشوهات الخلقية الكبرى في الجهاز الهضمي ، اليمن

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تاريخ استلام البحث: ٦ شباط ٢٠٢٢

تاريخ قبول البحث: ١٥ آذار ٢٠٢٢

[٢،١] كلية الطب - جامعة حضرموت – المكلا - اليمن

[٤،٣]مستشفى أبن سيناء و مستشفى حضرموت الحديث - حضرموت - اليمن