https://doi.org/10.29309/TPMJ/2022.29.08.7040

The Professional Medical Journal
www.theprofesional.com
2022, Volume, 29 Issue, 08

ORIGINAL ARTICLE

Frequency of various types of gastrointestinal abnormalities in patients with Down's syndrome.

Sumera Makhdoom[1], Shazia Mahar[2], Zubair Khoso[3], Roshia Parveen[4], Versha Rani Rai[5], Shazia Kulsoom[6]

Article Citation: Makhdoom S, Mahar S, Khoso Z, Parveen R, Rai VR, Kulsoom S. Frequency of various types of gastrointestinal abnormalities in patients with Down's syndrome. Professional Med J 2022; 29(8):1218-1222. https://doi.org/10.29309/TPMJ/2022.29.08.7040

ABSTRACT… Objective: To assess the frequency of various types of gastrointestinal abnormalities in patients with Down's syndrome. **Study Design:** Cross Sectional Study. **Setting:** National Institute of Child Health, Karachi. **Period:** January 2019 June 2019. **Material & Methods:** All children with age 1 month to 16 years with Down's syndrome of either gender presented with gastrointestinal signs and symptoms were enrolled. History along with demographic information was obtained from parent and GI examination was done. The outcome variables like duodenal stenosis, gastrophageal reflux, imperforate anus and Hischsprung's disease were noted. **Results:** The mean age of the children was 8.53+4.07 years. The mean terminal age at the time of delivery was 30.70+3.60 years. The mean number of siblings was 1.08+0.91. Duodenal stenosis was found in 18(6.40%) patients, gastroesophageal reflux was found in 10(3.50%) patients, imperforate anus in 9(3.10%) and Hischsprung's disease was found in 7(2.40%) children. **Conclusion:** Duodenal stenosis was the most common gastrointestinal (GI) abnormalities found in patients with down's syndrome.

Key words: Down's Syndrome, Gastrointestinal Abnormalities, Duodenal Stenosis, Hirschsprung's Disease, Imperforate Anus.

INTRODUCTION

Down's syndrome, or trisomy 21, the common inherited genetic syndrome and occurs in 1 in 800 - 100 live births.[1] Down syndrome patients can now reach their full mental and physical potential due to improvements in prenatal diagnosis, fetal and infant surgery, early intervention program.[2,3] It is essential to document growth, monitor developmental milestones, offer vaccination, or test for acute illness in Down syndrome children, just as it does with other children. Guidelines for people with Down syndrome in terms of Healthcare providers must anticipate healthcare problems and screen aggressively to help this growing population of patients receive quality and efficient care.[4,5]

Advanced maternal age, which increases beyond the age of 35.0 years; previous children with Down syndrome or other chromosomal abnormality; paternal stable translocation and one parent with chromosomal diseases are all risk factors for Down syndrome.[6,7] Hormones, medications, Vitamin defeciency, or viruses have not been identified as causes of Down syndrome. Parental screening can be done during the 1st or 2nd trimester and usually includes serum and ultrasound tests. Hypotension, lymphedema, thickening of nuchal cord, epicanthi folds and Simian crease are the striking features of Down syndrome at birth.[8] It is imperative to screen the life-threatening complications of Down syndrome before discharge after birth as it accompanies various complications including cardiovascular, pulmonary, gastrointestinal and ENT etc…. among all of the above complications there's increased frequency of gastrointestinal problems.[9]

A12 year retrospective review, 187.0 patients with Down's syndrome admitted to the Columbia

1. MBBS, Senior Registrar Pediatric Medicine, NICH.
2. FCPS (Pediatric), Senior Registrar Pediatric Medicine, NICH.
3. MBBS, FCPS, Assistant Professor Pediatric, NICH.
4. FCPS (Pediatric), Senior Registrar Pediatric Medicine, NICH.
5. FCPS (Pediatric), Senior Registrar Pediatric Medicine, NICH.
6. MBBS, FCPS, Assistant Professor Pediatric, NICH.

Correspondence Address:
Dr. Shazia Mahar
Department of Pediatric Medicine
NICH.
shazia.mahar@hotmail.com

Article received on: 24/02/2022
Accepted for publication: 27/04/2022

Medical center were identified. 27(14.4%) had major GI disorders, the common duodenal stenosis (DS) 9(4.8%), gastroesophageal reflux (GER) 5(2.67%), imperforate anus 5(2.67%) and Hirschsprung's disease 4(2.1%). The overall death rate was 20(11%) of the patients.[10]

The data regarding gastrointestinal complications in Down's syndrome is very scarce and no data is available locally. The international study done is also very old, the current study is designed to assess the current and local status of gastrointestinal complications in patients with Down's syndrome, thereby strategies could be developed to screen these cases at the earliest and prompt treatment may decrease the morbidity.

MATERIAL & METHODS

This cross sectional study was conducted form National Institute of Child Health, Karachi. Two hundred seventy eight (278) sample size was calculated with 80 power of test and 5% level of significance by taking proportion of Hirschsprung's 2.13 of Down' syndrome children.

Children with Down's syndrome of either gender presenting with gastrointestinal signs and symptoms with age 1 month to 16 years were included from the study. Children with neural tube defect and non-consenting parents were excluded from the study.

History along with demographic information was obtained from parents and GI examinations was done. The outcome variables like duodenal stenosis, gastroesophageal reflux, imperforate anus and Hirschsprung's disease was labelled. These findings along with the demographic were entered in the questionnaire attached.

Data analyses was done on SPSS 20. Type of GI abnormalities, economic status, consanguineous marriage of parents, maternal age at delivery, number of siblings, family history of Down's syndrome and gender were presented as frequency and percentage. Age at delivery was presented as mean and SD. Stratification was done with regard to age, economic status, consanguineous marriage of parents, maternal age at delivery, number of siblings, family history of Down's syndrome and gender to see the effect with Hirschprung's disease. Chi square test was applied and p value < 0.05 was taken as significant.

RESULTS

Total 278 children were included in this study. The mean age was 8.53+4.07 years. There were 201(70.4%) children with <10 years. Mean maternal age at time of delivery was 30.70+3.60 years. Almost half of the patients 170 (59.2%) had maternal age at the time of delivery of > 30 years. Mean number of siblings was 1.08+0.91. Majority of the patients 269(93.7%) had <2 number of siblings.

Female predominance was found to be higher 191(66.60%) as compared to males 96(33.40%). Poor economic status was found in 184(64.10%) children, middle in 61(21.30%) and higher 42(14.60%) patients. Table-I

Consanguineous marriage of parents was found in 207(72.10%), while family history of Down's syndrome was found in 217(75.60%) children. Duodenal stenosis was found in 18(6.30%) patients, gastroesophageal reflux was found in 10(3.50%) patients, imperforate anus in 9(3.10%) and Hischsprung's disease was found in 7(2.40%) children. Table-II

Stratification was done with respect to age, gender, maternal age at delivery, number of siblings, economic status, consanguineous marriage of parents and family history of Down's syndrome with the outcome. Chi-square test was applied, there were no significant relationship with Hirschprung's disease. Table-III

DISCUSSION

Down's syndrome is by far the common and best known chromosomal disorder in human or the most common cause of intellectual disability. Hypotension, microcephaly, nuchal cord thickening, lymphedema, epicanthal folds and simian creases are the striking signs of Down's syndrome at birth.

		Frequency (%)
Age	Mean+ SD	8.53+4.07
	<10	201(70.4%)
	>10	85(29.6%)
Maternal Age	Mean+ SD	30.70+ 3.60
	<30	117(40.8%)
	>30	170(59.2%)
Number of Siblings	Mean+ SD	1.08+0.91
	<2	269(93.7%)
	>2	18(6.3%)
Gender	Male	191(66.60%)
	Female	96(33.40%)

Table-I. Distribution of age, maternal age, number of siblings & gender

	Yes	No
Consanguineous marriage of parents	207(72.10%)	80(27.90%)
Family history of Down's syndrome	217(75.60%)	70(24.45%)
Duodenal stenosis	18(6.30%)	269(93.70%)
Gastroesophageal Reflux	10(3.50%)	277(96.50%)
Imperforate Anus	9(3.10%)	278(96.90%)
Hirschsprug's Disease	7(2.40%)	280(97.60%)

Table-II. Distribution of consanguineous marriage of parents,

		Hirschsprug's Disease		
		Yes	No	P-Value
Age	<10	5	197	1.00
	>10	2	83	
Gender	Male	4	92	0.228
	Female	3	188	
Maternal age at time of delivery	<30	1	117	0.247
	>30	6	170	
Economic status	Poor	5	184	0.894
	Middle	1	61	
	Higher	1	42	
Consanguineous marriage of parents	Yes	6	207	0.678
	No	1	80	
Family history of syndrome	Yes	7	217	0.201
	No	0	70	

Table-III. Comparison of Hirschsprung's disease with general characteristics

Midface hypoplasia, a curved digit, clinodactyly and brachydactylic are further phenotypic signs of Down's syndrome.[11] Because Down's syndrome is associated with a variety of problems, including cardiovascular, CNS, pulmonary, ENT, and gastrointestinal, it is critical to check for life-threatening abnormalities before discharge after delivery. There is a higher incidence of gastrointestinal disorders among all complications.

In this study, Duodenal stenosis was found in 18(6.30%) patients gastroesophageal reflux was found in 10(3.50%) patients, imperforate anus in 9(3.10%) and Hischsprung's disease was found in 7(2.40%) children. Similar results were found in another study in which, out of 187 patients with Down's syndrome 27(14.44%) had main GI disorder, Duodenal stenosis (DS) is the common (4.8 %), gastroesophageal reflux (GER) 5(2.67%), imperforate anus 5(2.67%) and Hischsprung's disease 4(2.13%). The death rate was particularly high in a small sample of individuals with duodenal stenosis (56%).[12]

There was no relationship between mother age, race, newborn sex, or chromosomal error origin when all GI abnormalities were considered combined. Even if the number of abnormalities is minimal, it is generally best to evaluate each one separately since their embryological origins varies. For example, At 4-5 weeks of development, abnormalities in mesenchyme growth and portioning of the esophagus and trachea cause esophageal atresia[13], failure to recanalize the intestinal lumen at 8 weeks is the most common cause of duodenal atresia, Between the 5th or 12th week, neural crest cells fail to migrate and colonized the submucosal and myenteric plexuses of the enteric nervous system, resulting in Hirschsprung disease.[14]

Hirschsprung's disease is responsible for around one-fifth of all newborn intestinal occlusions.[15] Aganglionosis only affects the rectum and sigmoid in around 3-quarters of such cases.[16] Long segment disease affects a piece of the colon proximal to the sigmoid, whereas complete aganglionosis coli affects the whole colon as well as a portion of the terminal ileum.[17] Total colonic aganglionosis and ultra-short segment disease is uncommon. The number of patients show up within the first six weeks of their life. Failure to pass meconium, stomach distention, vomiting, or enterocolitis are

common in newborns. Hirschsprung's illness is characterized by enterocolitis, which is the major cause of mortality.[18] Long segment disease is more likely to cause enterocolitis and rupture. The ascending colon or the appendix are the most prevalent sites for perforation.[19]

CONCLUSION

Duodenal stenosis was the most common Gastrointestinal abnormalities found in patients with Down' syndrome.

REFERENCES

1. Kurabayashi N, Nguyen MD, Sanada K. **Triple play of DYRK1A kinase in cortical progenitor cells of Trisomy 21.** Neuroscience Research. 2019; 138:19-25.
2. Antonarakis SE, Skotko BG, Rafii MS, Strydom A, Pape SE, Bianchi DW, et al. **Down syndrome.** Nature Reviews Disease Primers. 2020; 6(1):1-20.
3. Bull MJ. **Down syndrome.** New England Journal of Medicine. 2020; 382(24):2344-52.
4. Yi H, Siu QK, Ngan OM, Chan DF. **Parents' experiences of screening, diagnosis, and intervention for children with autism spectrum disorder.** American Journal of Orthopsychiatry. 2020; 90(3):297.
5. Capone GT, Chicoine B, Bulova P, Stephens M, Hart S, Crissman B, et al. **Co□occurring medical conditions in adults with Down syndrome: A systematic review toward the development of health care guidelines.** American Journal of Medical Genetics Part A. 2018; 176(1):116-33.
6. Jones JT, Smith C, Talib N. **Brief musculoskeletal screen and patient education for down syndrome-associated arthritis.** Global Pediatric Health. 2021;8:2333794X211045562.
7. Oyenusi EE, Ajayi EO, Akeredolu FD, Oduwole AO. **Pattern of thyroid disorders in children and adolescents seen at the Lagos University Teaching Hospital, Nigeria, over a 10-year period.** Nigerian Medical Journal: Journal of the Nigeria Medical Association. 2017; 58(3):101.
8. Innes AM, Lynch DC. **Fifty years of recognizable patterns of human malformation: Insights and opportunities.** American Journal of Medical Genetics Part A. 2021; 185(9):2653-69.
9. Schuchman EH, Desnick RJ. **Types a and B Niemann-pick disease.** Molecular genetics and metabolism. 2017; 120(1-2):27-33.
10. Bermudez BE, de Oliveira CM, de Lima Cat MN, Magdalena NI, Celli A. **Gastrointestinal disorders in Down syndrome.** American Journal of Medical Genetics Part A. 2019; 179(8):1426-31.
11. Graham Jr JM, Sanchez-Lara PA. **Smith's recognizable patterns of human deformation:** Elsevier Health Sciences; 2015.
12. Buchin PJ, Levy JS, Schullinger JN. **Down's syndrome and the gastrointestinal tract.** Journal of clinical gastroenterology. 1986; 8(2):111-4.
13. Beasley SW. **Oesophageal atresia and tracheo-oesophageal fistula.** Surgery (Oxford). 2019; 37(11):623-7.
14. Puri P, Holschneider AM. **Hirschsprung's disease and allied disorders:** Springer; 2019.
15. Lau PE, Cruz S, Cassady CI, Mehollin-Ray AR, Ruano R, Keswani S, et al. **Prenatal diagnosis and outcome of fetal gastrointestinal obstruction.** Journal of pediatric surgery. 2017; 52(5):722-5.
16. Zani A, Eaton S, Morini F, Puri P, Rintala R, Van Heurn E, et al. **European Paediatric Surgeons' Association survey on the management of Hirschsprung disease.** European Journal of Pediatric Surgery. 2017; 27(01):096-101.
17. Romano C, Oliva S, Martellossi S, Miele E, Arrigo S, Graziani MG, et al. **Pediatric gastrointestinal bleeding: Perspectives from the Italian Society of Pediatric Gastroenterology.** World journal of gastroenterology. 2017; 23(8):1328.
18. Mungnirandr A. **Hirschsprung's disease.** Siriraj Medical Journal. 2017; 69(4):223-7.
19. Subramaniam V, Shah MSM, Faiziah WARW. **Perforated neonatal appendicitis: A rare presentation of Hirschsprung disease.** Journal of Pediatric Surgery Case Reports. 2021; 71:101935.

AUTHORSHIP AND CONTRIBUTION DECLARATION

No.	Author(s) Full Name	Contribution to the paper	Author(s) Signature
1	Sumera Makhdoom	Concept of study.	
2	Shazia Mahar	Paper writing and study design.	
3	Zubair Khoso	Conduction of study and preparation of mansucirpt.	
4	Roshia Parveen	Final draft & manuscript.	
5	Versha Rani Rai	Data collection.	
6	Shazia Kulsoom	Read and approved the final manuscirpt.	