East African Scholars Journal of Medicine and Surgery
Abbreviated Key Title: EAS J Med Surg
ISSN: 2663-1857 (Print) & ISSN: 2663-7332 (Online)
Published By East African Scholars Publisher, Kenya

OPEN ACCESS

Volume-7 | Issue-9 | Sep-2025 | DOI: https://doi.org/10.36349/easjms.2025.v07i09.003

Original Research Article

Clinical Characteristics and Risk Factors for Hirschsprung's Disease among Hirschsprung's Disease Patients at a Tertiary Hospital, Dar es Salaam, Tanzania

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Article History
Received: 02.07.2025
Accepted: 08.09.2025
Published: 12.09.2025

Journal homepage:
https://www.easpublisher.com

Quick Response Code

Abstract: ***Background*:** Hirschsprung's disease (HD) is the most common surgical condition in pediatrics posing challenges to pediatricians and pediatric surgeons mainly practicing in resource-limited countries. Clinical features of the disease are variable and the risk factors in our setting are not well studied. ***Objectives*:** This study aimed at describing the clinical characteristics and determining the risk factors for Hirschsprung's disease among Hirschsprung's disease patients at Muhimbili National Hospital. ***Method***: A case-control study was carried out whereby patients with histologically confirmed Hirschsprung's disease and their caretakers admitted to the ward or attending regular clinics were selected, and verbally interviewed using a structured questionnaire and their findings were compared with corresponding controls without HD. ***Results*:** The study enrolled 225 cases and 476 controls. The majority of participants were male accounting for 156(69.4%) and 286(63.4%) for both cases and controls respectively. The mean age of the participants was 58 months, mainly coming from the coastal region where the hospital (MNH) is located. The most common age at first clinical presentation was neonatal age but the most age at confirmatory diagnosis was 25-60 months. Chronic c**o**nstipation was the most common clinical presentation accounting for 223(99.1%) of cases followed by abdominal distension 207(92.0%) and soiling. Failure to pass meconium within 24 hours was common in cases (37.6%) compared to control (8.6%). %). Prematurity, Down's syndrome, history of HD, and congenital anomalies in the family were found to be associated risk factors for Hirschsprung's disease. ***Conclusion***: The most common clinical presentations were found to be chronic constipation, abdominal distension, failure to pass meconium within 24-48 hours of life, and male predominance. Prematurity, Down's syndrome, history of HD, and congenital anomalies in the family were more likely to be associated with Hirschsprung's disease with the late diagnosis mainly at 25 to 60 months.

Keywords: Hirschsprung's Disease, Constipation, Down's syndrome.

BACKGROUND

Hirschsprung's Disease (HD) is a congenital disorder characterized by the absence of enteric ganglia along a variable length of the large intestine, resulting in a functional obstruction with various clinical manifestations [1–3].

The incidence of the disease ranges from 1 per 4400 to 1 per 7000 live births with most cases diagnosed in infancy in developed countries compared to developing countries [1, 2].

Clinical features of the disease and even its diagnostic approaches and treatment are various [2]. The clinical presentation ranges from neonatal intestinal

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obstruction to chronic progressive constipation in older children. Approximately 80 percent of patients present in the first few months of life with difficult bowel movements, poor feeding, and progressive abdominal distention [1]. Some patients may present with mild or severe constipation at different ages, failure to pass meconium 24-48 hours after birth, prominent abdominal distension, even perforation of the proximal colon, and peritonitis during the first few days after birth [1].

Some cases of the disease have been seen with the associated anomalies that account for 25% of familial cases and 10% of non-familial cases [1, 2]. There is also a male predominance of about 4 to 1 in HD [1-4]. There are also few studies on the risk factors where the disease is linked with maternal obesity, Downs syndrome, and prematurity [3-5].

In Tanzania, the majority of patients present very late when the disease becomes complicated contributing significantly to high morbidity and mortality [1, 2]. This is mainly due to atypical presentation that hinders the early detection of the disease. There are also few studies on typical presentation and risk factors in our setting leading to little knowledge about the disease.

The study aims to establish the clinical characteristics and risk factors for Hirschsprung's disease in our setting to increase the index of suspicion for early diagnosis, intervention, and plan for future preventive measures.

METHODS

The study design was an unmatched 1:2 case-control study conducted for one year at Muhimbili National Hospital. The sample size was calculated using sample size calculation for case-control studies. A convenient sampling technique was used to recruit participants for the study. The cases included any patient with histologically confirmed HD aged less than 18 years whose parent or caretaker has consented to participation in the study while controls included any patient with normal bowel movement less than 18 years of age with parental or caretaker's consent. Participants were recruited from admitted patients in surgical pediatric wards and those seen as an outpatient during the surgical pediatric clinic. A patient whose parent or caretaker could not retrieve at least 50% of the required information was excluded.

Data was collected by filling the structured questionnaires through interviews with caretakers of the patients in the ward or at the clinic. Data that was collected included demographic data, clinical presentation, risk factors, and associated congenital anomalies.

Data extracted from the structured questionnaires were analyzed using the Statistical Package for Social Sciences (SPSS) version 27. Two by two tables involving case and control were constructed. The chi-square and T-test were used to find the p-values for categorical and continuous variables. Fisher's Test was only used to find P-values for categorical variables with values less than five. P Values less than 0.05 were considered significant for available to be used for calculating the Odd ratio (OR) in logistic regression analysis.

RESULTS

The study aimed at finding the clinical characteristics and risk factors associated with HD whereby 225 cases and 476 controls treated at MNH as an inpatient or outpatient were studied. The majority of participants were male accounting for 156(69.4%) and 286(63.4%) for both cases and controls respectively. The mean age of the participants was 58 months mainly coming from the coastal region (Dar es Salaam, Morogoro, Pwani, and Linda) where the hospital (MNH) is located **(**Table 1A).

The commonest age at the time of initial clinical presentation was neonatal age (0-1 month) accounting for 92(40.9%) of all cases while that at confirmatory diagnosis was preschool age (25-60 months) accounting for 81(36%) of all the cases. (Fg. 1A and 1B).

Chronic **co**nstipation was found to be the commonest clinical presentation accounting for 223(99.1%) of cases followed by gaseous abdominal distension in 207(92.0%). Failure to pass meconium within 24hour was common in cases (37.6%) compared to control (8.6%). Table 1B while Soiling accounted for 46% of all other clinical presentations assessed among the cases. (Fig. 2)

Gestation age less than 37 weeks (prematurity) and Down's syndrome were associated with Hirschsprung's Disease. (p=0.001, OR=6.11 95% CI=3.01-12.32) and (p=0.001, OR=2.53, 95% CI=1.71-3.74) respectively Table 2A. Family history of HD and congenital anomalies were also associated with having a child with HD. (p=0.001, OR=9.78, 95% CI=2.27-34.53) and (p=0.002, OR=3.54, 95% CI=1.157.99) respectively. Table 2B

On the other hand, most of the cases 196(87.1%) and controls 422(93.6%) had no congenital anomalies clinically. However among the few anomalies identified, CNS and Sinonasal anomalies were found to be more in cases 6(2.7%) and 5(2.2%) compared to control 4(0.9%) and 0(0%) respectively (fig.3) though there was no statistical significance on logistic regression. Table 3

Table 1A: Socio-demographic characteristics of participants

Demographic characteristic Variables	Type of Participant			P-Value
	Control	Case	Total	
Sex	**N(%)**	**N(%)**	**N(%)**	
Male	286(63.4)	156(69.3)	442(65.4)	
Female	165(36.6)	69(30.7)	234(34.6)	0.127
Total	451(100)	225(100)	676(100)	
Age				
1-12	64(14.2)	7(3.1)	71(10.5)	
13-24	118(26.2)	14(6.2)	132(19.5)	
25-60	142(31.5)	82(36.4)	224(33.1)	
61-144	105(23.3)	103(45.8)	208(30.8)	
>144	22(4.9)	19(8.4)	41(6.1)	0.001
Total	**451(100)**	**225(100)**	**676(100)**	
Residence				
Central zone	12(2.7)	28(12.4	40(5.9)	
Coastal zone	368(81.6)	144(64.0)	512(75.7)	
Lake zone	5(1.1)	4(1.8)	9(1.3)	
Northern zone	20(4.4)	12(5.3)	32(4.7)	
Southern H. zone	23(5.1)	16(7.1)	39(5.7)	
Southern zone	14(3.1)	11(4.9)	25(3.7)	
Western zone	3(0.7)	2(0.9)	5(0.7)	
Zanzibar	6(1.3)	8(3.6)	14(2.1)	0.001
Total	**451(100)**	**225(100)**	**676(100)**	

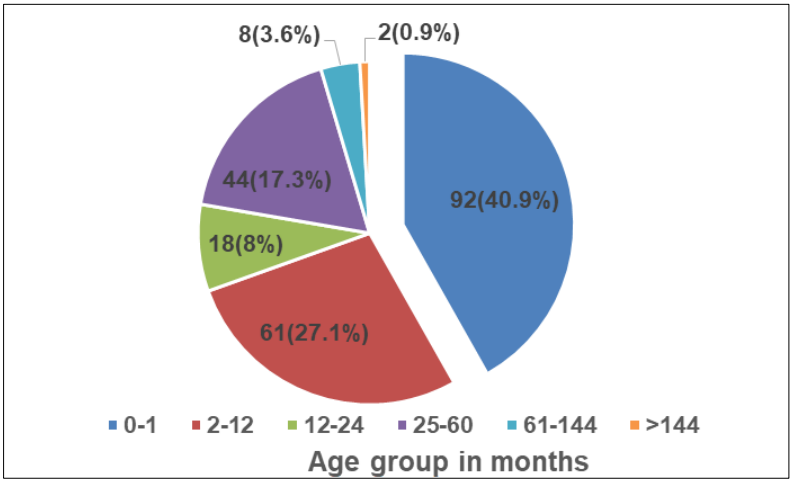

Figure 1A: Age distribution among the case at the time of initial clinical presentation

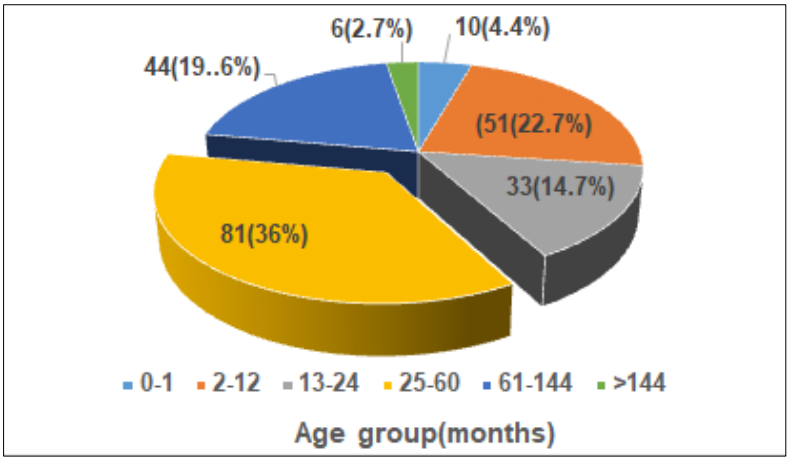

Figure 1B: Age distribution among the case at the time of confirmatory diagnosis

Table 1B: Child Clinical characteristics

Clinical Characteristic variable	Type of participant			
	Control	Case	Total	P-value
Chronic Constipation	N(%)	N(%)	N(%)	
Yes	0(0.0)	223(99.1)	223(33.0)	
No	451(100)	2(0.9)	453(67.0)	0.001
Total	451(100)	225(100	676(100)	
Abdominal distension				
Yes	0(0.0)	207(92.0)	207(30.6)	
No	451(100)	18(8.0)	469(69.4)	0.001
Total	451(100)	225(100	676(100)	
Meconium				
Yes	408(90.5)	126(56.0)	534(79.0)	
No	39(8.6)	84(37.3)	123(18.2)	
Not sure	4(0.9)	15(6.7)	19(2.8)	0.001
Total	451(100)	225(100	676(100)	
Vomiting				
Yes	0(0.0)	43(19.1)	43(6.4)	
No	451(100)	182(80.9)	633(93.6)	
Total	451(100)	225(100	676(100)	0.001
Fever				
Yes	0(0.0)	47(20.9)	47(6.9)	
No	451(100)	178(79.1)	629(93.1)	0.001
Total	451(100)	225(100	676(100)	

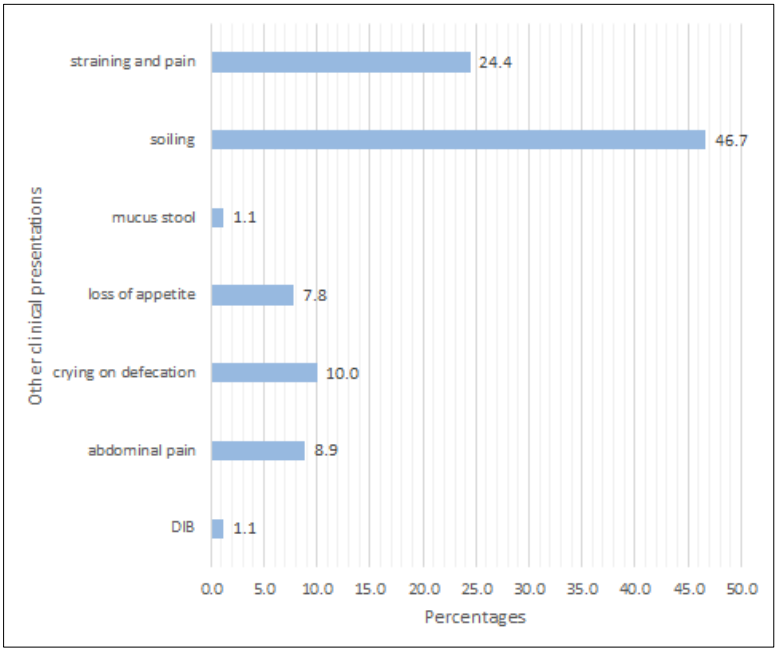

Figure 2: Other Clinical presentations

Table 2A: Logistic regression of maternal and child characteristics factors versus the type of participants

	Crude Odds Ratio			Adjusted Odds Ration		
	OR	95%CI	P-Value	OR	95%CI	P-Value
Maternal variables						
Birth weight						
Normal	Ref					
Low	1.67	1.03-2.71	0.036*	0.87	0.47-1.58	0.642
Gestation age						
>=37	Ref					
<37	6.36	3.42-11.80	0.001**	6.11	3.01-12.38	0.001

Mother body mass index (BMI)						
Normal	Ref					
Underweight	6.14	1.25-30.18	0.025*	4.94	0.94-25.73	0.061
Overweight	1.23	0.84-1.79	0.274	1.27	0.85-1.88	0.113
Obesity	0.42	0.27-0.65	0.001*	0.51	0.32-0.81	0.004
Maternal diseases						
No	Ref					
Yes	2.24	1.57-3.21	0.001*	7.25	0.82-63.50	0.073
Down syndrome						
No	Ref					
Yes	10.23	1.19-88.07	0.034*	2.53	1.71-3.74	0.001
Gravidity						
One	Ref					
Two	0.71	0.46-1.10	0.130	0.68	0.43-1.09	0.111
Three	1.02	0.67-1.56	0.906	1.07	0.67-1.70	0.765
Four	0.72	0.44-1.13	0.154	0.68	0.41-1.14	0.148
History of medication use						
No	Ref					
Yes	1.03	0.69-1.52	0.883	0.93	0.62-1.40	0.734

Table 3B: Logistic regression for familial factors and type of participants

Family History Variables	**Crude Odds Ratio**			**Adjusted Odds Ration**		
	OR	**95%CI**	**P-Value**	**OR**	**95%CI**	**P-Value**
Congenital diseases in the family						
No	Ref					
Yes	3.83	1.73-8.45	0.001	3.54	1.57-7.99	0.002
Family history of HD						
No	Ref					
Yes	10.67	3.05-37.24	0.001	9.78	2.77-34.53	0.001

Table 3: logistic regression for congenital anomalies and type of participants

	Crude odds Ratio	**Adjusted Odds Ratio**		
	OR	**OR**	**95%CI**	**P-Value**
Congenital anomalies				
Cardiovascular	0.90	0.48	0.31-2.58	0.84
Cleft palate	3.23	2.10	0.90-11.57	0.07
CNS	2.15	1.53	0.53-8.69	0.28
Gastrointestinal	2.15	2.16	0.30-15.39	0.45
Hematological	6.46	7.48	0.67-62.49	0.11
Musculoskeletal	2.15	3..05	0.13-34.60	0.59
Sino nasal	2.15	3.05	0.13-34.61	0..59

DISCUSSION

The study revealed that the majority of patients 91(40.9%) presented with clinical symptoms during neonatal age but there was a delay in confirming the diagnosis whereas the majority 81(36%) were confirmed between the age of 2-5years. This is similar to other studies done in Africa which showed that few cases less than 30% were diagnosed in the neonatal period compared to developed countries [1-3]. This is might be due to less understanding of the disease, especially in primary health facilities where these patients are initially treated as patients with other gastrointestinal problems before realizing that it is a quite different disease.

The study also revealed male predisposition among patients with HD equivalent to 2:1. This is similar to most studies done in America, Europe, Asia, and Africa that also showed male predisposition among patients diagnosed to have Hirschsprung's disease. However, there was variation in sex ratio with some studies in Asia showing 2:1 while studies in Africa going up 4:1 male to female sex ratio [1–3]**.** The male predisposition is probably due to sex-linked genes present in males.

The commonest clinical presentations noted were chronic constipation 223(99.1%), gross gaseous abdominal distension 207(92.0%), soiling 105(46.7%), and failure to pass meconium within 24 hours after birth 84(37.3%). The other clinical manifestations were straining and pain on defecation 55(24.4%), fever 47(20.9%), and vomiting 43(19.0%). This is similar to

most studies done in Africa, Asia, and other developed countries which showed that chronic constipation, abdominal distension, and delay in passing meconium were the commonest presentations except for soiling which was almost not mentioned in these studies [1–8]. The similarity is mainly to the common pathophysiology of the disease. Soiling was found to be common in this study compared to other studies because of the late diagnosis of the disease where parents could pick it up easily when the child has grown up to the age of controlling continence.

Prematurity, having a family history of congenital anomalies, and HD as well as features of Down's syndrome were associated with developing HD. This is quite similar to some studies done in developed countries and other parts of Africa that showed that these factors were related to HD [5–10]. However other factors mentioned in these studies such as maternal age, obesity, and prenatal use of medication linked with HD were not statistically significant in this study. The reason for similarity may be due to the immature nervous system in premature babies and the genetic link of the disease in family history. The difference is probably due to biological variations between the regions.

The family history of HD was noted to be common in cases 15(6.7%) compared to control 3(0.7%) suggesting genetic predisposing of the disease in the family. This is quite similar to the fact noted in some studies that HD is known to be a multifactorial disease caused by both genetic and environmental factors [11, 12]. It is also noted that there is a familiar tendency with risk to a relative being higher than the incidence in the general population [31–16).

Central nervous system anomalies were seen more in case 6(2.7%) followed by sinonasal anomalies 5(2.2%) and Down's Syndrome 5(2.2%) compared to 4(0.9%), 1(0.2%) and 0(0%) in control respectively. This is similar to the study done in Turkey which showed that CNS and special senses were common anomalies in 28.92% of HD patients. Also, several studies in Africa, Asia, America, and Europe have mentioned Down's syndrome to be a commonly associated congenital disease in Hirschsprung's patients [5–18]**.** The similarity is mainly due to the pathogenesis behind the development of the disease that involves the migration of neurocrest cells.

CONCLUSION

The most common clinical presentations were found to be chronic constipation, abdominal distension, failure to pass meconium within 24-48 hours of life, soiling, and male predominance with the late diagnosis mainly at 25 to 60 months. Prematurity, Down's syndrome, history of HD, and congenital anomalies in the family were more likely to be associated with Hirschsprung's disease.

Recommendation

Clinical presentations and risk factors identified in the study should raise high suspicions of HD that need evaluation for early diagnosis and intervention, especially at primary health facilities.

Abbreviations

CI: Confidence interval, CNS: Central Nervous System, HAEC: Hirschsprung Associated Enterocolitis, HD: Hirschsprung's Disease, MNH: Muhimbili National Hospital, MUHAS: Muhimbili University of Health and Allied Sciences.

Declarations

Ethics Approval and Consent to Participate

The study was approved by our MUHAS and MNH IRB committee (MUHAS-REC-06-2021-684 and MNH/TRCU/Perm/2021/196). Verbal consent was obtained from all parents or guardians who participated in the study.

Consent for Publication

Consent for publication was obtained from Muhimbili University of Health and Allied Science (MUHAS), Director of Postgraduate Studies and Research Publication.

Availability of Data and Materials: The data and material are available for review.

Competing Interests: There are no competing interests.

Funding

The funding for this study was brought by the Tanzania Ministry of Health, Community Development, Gender, Elderly and Children (MoHCDDEC), and there was no conflict of interest.

Authors' Contributions

MM participated in data analysis, interpretation, and manuscript editing.TT, MN, SM, EM and I.K assisted on study design, data collection, analysis, interpretation and manuscript writing. All authors have read and approved the final manuscript.

Acknowledgments

To the department of General Surgery for coordinating this study and manuscript preparation.

To Tanzania Ministry of Health, Community Development, Gender, Elderly and Children (MoHCDDEC) for funding this study.

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Cite This Article: Laetus Mwombeki, Sidney Yongolo, Petronilla Ngiloi, Mungeni Misidae, Timon Theophil, Museleta Nyakiroto, Sophia Mosha, Erick Mwijage, Immaculate Kalungi (2025). Clinical characteristics and Risk Factors for Hirschsprung's Disease among Hirschsprung's Disease Patients at a Tertiary Hospital, Dar es Salaam, Tanzania. *East African Scholars J Med Surg, 7*(9), 264-270.