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# Gastrointestinal congenital malformations: a review of 230 cases at Albert Royer National Children's Hospital Center in Senegal

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## Abstract

**Background** Congenital malformations are the third cause of mortality in children under five. We aimed to report sociodemographic and diagnostic aspects of gastrointestinal ones and their outcomes.

**Methods** We conducted a descriptive cross-sectional study of patients admitted from January 2018 to January 2021 to our department at Albert Royer National Children's Hospital Center in Dakar, Senegal. A total of 230 were included.

**Results** The frequency of these malformations was 6.18%. The mean age was 1.9 years, with neonates representing 43.48% and males 59.56%. 64.35% of patients came from the Dakar area. Parental consanguinity was reported in 11.73%, prenatal diagnosis in 5.56% of 36 cases, and prematurity in 28.84% of 52 patients. The most typical reasons for referral or symptoms were constipation (23.91%), imperforated anus (23.91%), and vomiting (23.48%). In 93.91%, the malformation was isolated, of which Hirschsprung's disease accounted for 30.56% and anorectal malformation for 30.09%. Esophageal atresia and anorectal malformations had more associated anomalies with 28.57% each. The VACTER-L association represented 21.42% of associated anomalies. Mortality was 27.83%, and lethality was 100% for intestinal atresia, 87.5% for esophageal atresia, and 85.71% for polymalformation. Causes of mortality were reported in 21%, with septic shock and respiratory distress in all esophageal atresia patients (Manama, Contribution à l'étude des malformations congénitales : à propos de 188 cas du service de néonatalogie de l'Hôpital Aristide Le Dantec de Dakar, 1983) and hypovolemic shock in all patients with duodenal atresia (Wright et al., Lancet 398:325–39, 2021).

**Conclusion** Congenital malformations of the gastrointestinal tract are still lately diagnosed in our environment, resulting in higher mortality. Further studies should analyze delayed presentation and mortality, and their risk factors in our settings.

**Keywords** Congenital malformations, Gastrointestinal tract, Delayed presentation, Mortality, Senegal

## Background

Congenital anomalies or birth defects occur in 1/5000 live births, and 94% happen in low- and middle-income countries (LMICs) [1]. They comprise functional and structural anomalies, representing two-thirds [2]. Their place in the mortality of children under 5 years is increasing, as they represent the third cause of mortality in under-five children [2]. Structural anomalies are congenital malformations (CMs), among which digestive ones are the fifth most commonest [3].

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In Sub-Saharan Africa, antenatal diagnosis of these defects is scarce, with many of them being lately diagnosed, with much-increased mortality compared to high-income countries (HICs) [4].

Congenital malformations of the gastrointestinal tract (GIT) have been separately studied in Senegal for many decades [5–9], reporting the local experience. This study aimed to comprehensively review sociodemographic elements, diagnosis, and outcomes of these diseases at Albert Royer National Children's Hospital Center in Dakar, Senegal.

## Methods

We conducted a descriptive cross-sectional study in the Pediatric Surgery Department of Albert Royer National Children's Hospital Center (ARNCHC) located in Dakar, the capital city of Senegal. It is a public-funded hospital, a subsidiary for patients. It is one of the country's three tertiary hospitals providing pediatric surgical care. All three tertiary hospitals are located in the Dakar area. The study period was 3 years, from January 2018 to January 2021.

Patients' medical records were retrieved from patients' medical files, operating room registers, and the Hospital's Informatics Services.

We included patients admitted for at least a single congenital malformation of the GIT from the esophagus to the anus. Patients whose medical records could not be retrieved or referred to another hospital were excluded.

Collected data were sociodemographic (age at diagnosis, sex, maternal age, geographic origin), diagnostic (prenatal ultrasound, pregnancy term, main complaint on admission, requested investigations, type of congenital malformation of the GIT, associated malformations), and outcomes (postoperative complications and mortality). Mortality is defined as the frequency of death in the population study, while lethality is the frequency of death specific to each disease. There were first registered on a pre-designed printed sheet before being encoded using an Excel spreadsheet (Microsoft™ 2019) and analyzed with SPSS version 26.0 (IBM Software). For qualitative variables, the percentage was calculated when mean and standard deviation (SD) were calculated for quantitative ones.

This study was approved by the Board of Albert Royer National Children's Hospital Centre. Due to its retrospective aspect, the consent to participate was waived.

## Results

Over the study period, 4762 patients were admitted to our department, while 242 had a congenital malformation of the GIT, which frequency is 5.08% in our department. Among the 242 patients, 12 were excluded, as 230 constituted our population study.

At diagnosis, the mean age was 1.9 years (1 day–15 years). Patients' age by the groups is represented in Table 1. There were 137 males, with a sex ratio of 1.47/1. Maternal age was reported in 61 cases, of which those aged from 25 to 30 years represented 29.50%. The data related to age and sex are summarized in Table 1.

Patients originated from 13 of the 14 regions of Senegal, with 148 (64.35%) coming from Dakar. The number of patients by area is reported in Fig. 1, a map of Senegal.

Parental consanguinity was reported in 27 cases (11.73%). Reports of prenatal US were found in 36 cases, of which 26 (72.22%) were normal and 10 (27.78%) abnormal. Two of the latter (5.56%) prenatally diagnosed the malformation (intestinal atresia and anorectal malformation), and eight (22.22%) suspected a problem but did not diagnose the malformation. The pregnancy term was reported in 52 cases, among whom prematurity was found in 15 (28.84%) and prolonged pregnancy in 4 (7.69%).

The symptoms or reasons for referral are represented in Fig. 2. Complementary investigations were reported in 114 cases, among which the most requested was plain abdominal X-ray (37.71%), followed by abdominal ultrasound (36.84%), plain thoracic X-ray (35.96%), upper gastrointestinal (UGI) series (22.80%), cardiac ultrasound (14.04%), rectal biopsy (7.89%), and UGI endoscopy (6.14%).

In 216 of the 230 patients, the congenital malformation of the GIT was isolated. The malformations were ARMs and Hirschsprung's disease (HD), found in 66 and 65 of the 216 patients with isolated gastrointestinal congenital malformations. In the 14 remaining patients, the GIT malformation was associated with other malformations. The VACTER-L association was reported in three patients, among whom the first had esophageal atresia (EA) associated with ARM and a limb malformation (digital agenesis), the second had EA associated with ARM and congenital heart disease (CHD), and the third had ARM associated with CHD and a single kidney. Different

**Table 1** Age and sex of included patients

Variable	Number (n = 230)	Percentage
<b>Age</b>		
0–28 days	100	43.48
29 days–30 months	87	37.83
31 months–5 years	15	6.52
5–15 years	28	12.17
<b>Sex</b>		
Males	137	59.56
Females	93	40.44

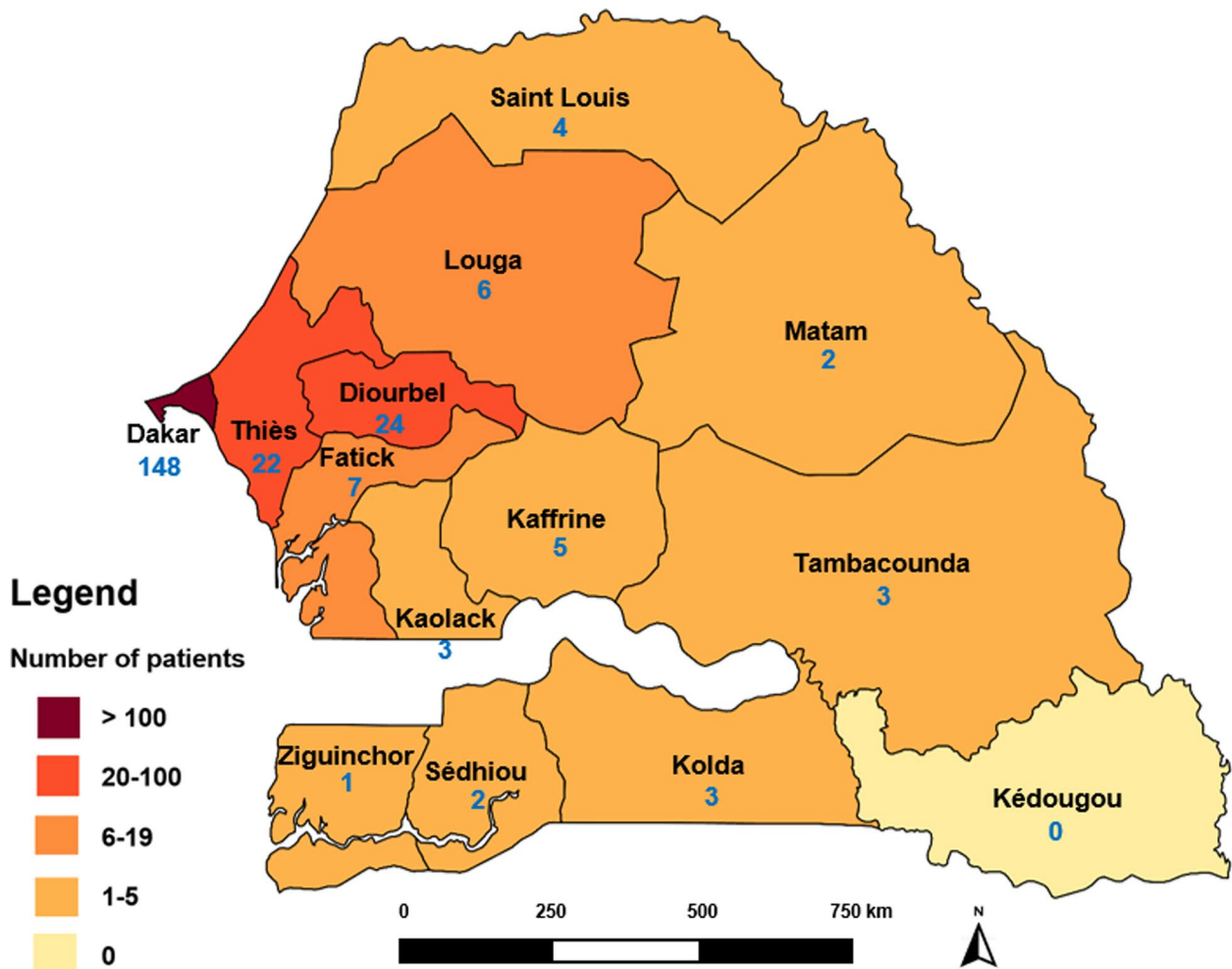


Fig. 1 Origin of patients included in our study

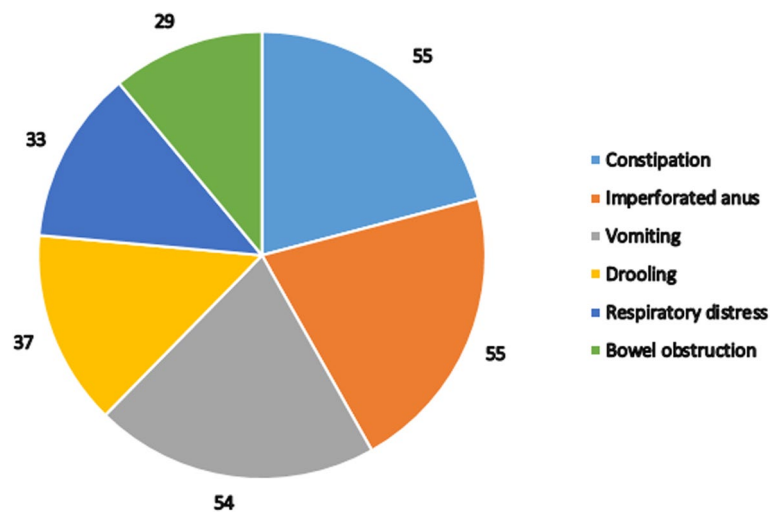


Fig. 2 Representation of symptoms and reasons or referral

digestive malformations found in our study are listed in Table 2, with their associated anomalies.

The mean duration of hospitalization was 9.52 days. The global mortality was 27.83%. Among the 230

**Table 2** List of reported malformations of the GIT: isolated and with associations

Gastrointestinal congenital malformations	Number	Percentage
<b>Isolated</b>	216	
Hirschsprung's disease	66	30.56
Anorectal malformation	65	30.09
Esophageal atresia	32	14.82
Malrotation	28	12.96
Duodenal atresia	15	06.94
Intestinal atresia	05	02.32
Congenital esophageal stenosis	04	01.85
Colonic atresia	01	00.46
<b>With associated malformations</b>	14	
VACTER-L syndrome	03	21.42
Esophageal atresia	04	28.57
+ ARM	01	07.14
+ Choanal atresia	01	07.14
+ CHD	01	07.14
+ Duodenal atresia	01	07.14
Duodenal atresia	01	07.14
+ Hydrocephalus	01	07.14
Malrotation	02	14.28
+ Umbilical hernia	01	07.14
+ Omphalocele	01	07.14
ARM	04	28.57
+ CHD	01	07.14
+ CHD and DSD	01	07.14
+ Dawn syndrome	01	07.14
+ DSD	01	07.14

ARM Anorectal malformation, CHD Congenital heart disease, DSD Disorder of sexual development, GIT Gastrointestinal tract, VACTER-L Vertebral, anorectal, cardiac, tracheoesophageal, renal, limbs malformations

patients, 184 underwent surgical intervention, 29 were waiting for a surgical procedure, and 17 died before any surgery, which gives a preoperative mortality rate of 7.39%. Among the 184 patients who benefited from a surgical procedure, 47 deaths were recorded, with 25.54% postoperative mortality. Table 3 reports the mortality and lethality of each congenital malformation of the GIT, with polymalformation as the leading etiology preoperatively (35.29%), while EA had the most postoperative deaths (48.93%) and the most considerable global mortality (43.75%). Lethality of malformation was 100% for intestinal atresia, 87.50% for EA, 85.71% for polymalformation, and 53.33% for duodenal atresia. The causes of mortality were reported in 14 patients (21.87%), of whom four deaths were preoperative and ten postoperative, as summarized in Table 4. In patients with preoperative mortality, the time from admission to mortality was reported in 16 of 17. The mean duration was 8.28 days, ranging from 1 to 25 days. For those with postoperative mortality, the time from surgery to death was reported in 19 of 47 patients. The mean duration was 9.47 days, ranging from 0 to 36 days.

## Discussion

Congenital malformations occur in 197.21 of 10,000 live births [10], with digestive ones occurring in 16.17 of 10,000 live births. In LMICs, population-based studies are scarce due to the lack of national registers of congenital malformations [11, 12]. Many studies in LMICs only report the in-hospital frequency of these defects, which in our review was approximately 5% for congenital malformations of the GIT.

The neonates represented almost half of our population of the study, differently from the findings of a global review on these malformations, which reported 90% of neonates among 3849 patients [13]. Many malformations of the GIT present early in the neonatal period. However, in LMICs, delayed diagnosis is

**Table 3** Mortality and lethality of malformations

Malformations	Preoperative mortality (n = 17)		Postoperative mortality (n = 47)		Mortality (n = 64)		Lethality	
	Number	Percentage	Number	Percentage	Number	Percentage	Number	Percentage
EA	5	29.42	23	48.93	28	43.75	28/32	87.50
Polymalformation	6	35.29	06	12.77	12	18.75	12/14	85.71
Duodenal atresia	4	23.53	04	08.51	08	12.50	08/15	53.33
ARM	1	05.88	05	10.64	06	09.32	06/65	09.23
Intestinal atresia	–	–	05	10.64	05	07.81	05/05	100
Malrotation	1	05.88	01	02.13	02	03.13	02/28	07.14
HD	–	–	01	02.13	01	01.56	01/66	01.52

ARM Anorectal malformation, EA Esophageal atresia, HD Hirschsprung's disease

**Table 4** Mortality causes

Diagnosis (number)	Timing of death (number)	Cause of death (number)
Esophageal atresia [5]	Postoperative: 5	Septic shock (2), respiratory distress (2), mediastinitis (1)
Duodenal atresia [4]	Preoperative: 3 Postoperative: 1	Hypovolemic shock (3) Hypovolemic shock (1)
Intestinal atresia [2]	Postoperative: 2	Septic shock (2)
Malrotation [1]	Postoperative: 1	Respiratory distress (1)
Hirschsprüng's disease [1]	Preoperative: 1	Denutrition (1)
Anorectal malformation [1]	Postoperative: 1	Enterocolitis (1)

common in non-fatal conditions such as HD or ARMs, for which the mean age reported in Senegalese studies is higher than those of HICs [14–16]. The causes of delayed diagnosis of CMs include cultural beliefs of parental society, parental level of education, kind and anatomic location of the CM, availability of medical workforce and structures, and difficulties in making the diagnosis [17].

Depending on the culture, consanguineous marriage is encouraged in some societies. In Senegal, parental consanguinity is not rare, as shown in our study by its finding in 11.73% of cases, similar to the frequency reported by other authors [18, 19]. Since some digestive malformations (ARMs, EA, HD) have a genetic component, consanguinity would logically increase their occurrence [20]. However, no specific counseling was reported on patients' files. Some authors demonstrated an increased frequency of CMs in children issued from consanguineous marriages [21]. Nevertheless, there was no statistically significant association between consanguineous marriage and digestive malformations [20, 22].

More than half of the patients recruited in our study originated from the Dakar area, followed by neighborhood regions, as fewer patients were referred from far regions. This may be due to the proximity of our hospital for patients living in the capital city and the fact that the Dakar area has more population than other areas, thus having more birth and, in turn, more CMs [23]. Nevertheless, a consistent number of patients came outside Dakar, representing up to a third of our population study. This may be due to many reasons: (a) our service is part of the National Children's Hospital Centre, the principal center for pediatric surgical care in the whole country, which receives patients from the whole Senegal and neighboring countries, such as Gambia and Guinea; (b) the lack of trained pediatric surgeons in some regions; and (c) the lack of dedicated structure to manage some pediatric surgical conditions, such as EA, ARMs, HD, or polymalformation. The

Global PaedSurg Research Collaboration's review highlighted that to reach tertiary structures, patients further traveled in low-income countries (LICs), compared to middle-income countries (MICs) and HICs [13].

Prenatal diagnosis of congenital anomalies is rare in Sub-Saharan Africa (SSA) [24]. Our study found prenatal ultrasound (US) reports in approximately 10% of patients. This highlights the need for systematic data collection in pediatric surgical services, which would help better analyze prenatal diagnosis through its different components: frequency of US realization, detection of anomalies, prenatal diagnosis, and management. Among the 36 patients, approximately 5% were diagnosed with a malformation of the GIT. In a fifth of prenatal US, an anomaly was seen but not diagnosed. This shows the need for projects to favor access to the prenatal US and improve the accuracy of prenatal diagnosis in LMICs. A regional or national antenatal surveillance program would act at two levels: (a) improve prenatal US skills among prenatal US performers working at district hospitals and (b) organize the transfer of pregnant women with abnormal prenatal US to tertiary centers, where the congenital anomaly has more chance to be diagnosed and subsequent management, initiated. However, prenatal diagnosis of these conditions is lower in LICs than in high-income ones [13]. Unlike HICs, where the prenatal US does not improve the outcome, it is postulated that it would improve outcomes in LMICs [24–26].

In our study, almost a quarter of patients reported premature pregnancy terms. In digestive malformations, various conditions can lead to total or partial obstruction of the GIT (EA, duodenal or intestinal stenosis/atresia, and colonic atresia/stenosis). This would lead to polyhydramnios, which can be responsible for premature birth. However, in our review, no cause of prematurity was reported to allow a better analysis of its potential association with these malformations. A 20-year review in English Caribbean countries reported prematurity in one-third of patients with digestive

malformations [27]. However, polyhydramnios is not specific to all malformations of the GIT.

Our review identified approximately 90% of isolated digestive malformations, among which HD and ARMs were the most frequent, with almost a third of the population study for each. Other studies in LMICs reported the same trend [11, 28, 29]. Even though some authors suggested that ARMs were more frequent in SSA [30, 31], there is still no sufficient data to confirm that hypothesis [32]. The higher frequency of these two malformations can be linked to the fact that pediatric surgical care in SSA is available only in some tertiary centers or children's hospitals, which leads to their higher referral to these centers. As reported in a recent review, digestive malformations with early fatality, such as EA, are less reported in LICs than in MICs and HICs [13]. This can be related to hidden mortality, such as in congenital diaphragmatic hernias, with newborns passing away before the proper diagnosis due to a lack of pediatric surgeons in the district hospitals or long before referral to a tertiary center. We reported almost 5% of congenital malformations of the GIT associated with other congenital malformations, much lower than data reported in the literature, with frequency ranging from 15.07 to 20% [28, 33]. This can be due to mortality before referral at a tertiary level and expenses of investigations, leading to underdiagnosis of associated malformations.

The mortality was approximately 30% in our study, similar to findings of other African studies, ranging from 20.8 to 39.8% [13, 34]. This is higher than the findings of HICs, where mortality of these conditions is approximately 5.6% [13]. Recently, it has been shown that there is a significant difference in digestive malformations mortality between HICs and LMICs [13]. In LMICs, many factors are contributive, such as the lack of prenatal diagnosis, anesthesiologist, surgical safety checklist, lack of ventilation, and unavailability of parenteral nutrition (PN) [13]. Other possible factors are delayed referral to tertiary hospitals, unappropriated medical transfer of patients, and inadequate resuscitation during the transfer and on admission. Additionally, some conditions had very high mortality in our context: such as intestinal atresia, EA, and cases of polymalformation. The latter was associated with mortality when IA and EA were not [13]. However, in African studies, EA lethality ranges from 45.5 to 90% [11, 13, 29, 35], while the one of IA ranges from 52.3 to 60.0% [13, 29, 35]. In our patients with EA where the mortality cause was reported, all died from infectious complications or respiratory distress. The delay in diagnosis and occurrence of aspiration pneumonia before surgery leads to higher mortality of EA in our environment, as previously reported in a local study [6]. Reported cause of death in our patients with DA was a hypovolemic shock in all four,

with three who died preoperatively. This should be linked to delayed diagnosis leading to progressive patient dehydration since oral hydration is inefficient, as the duodenal obstruction persists. Improving prenatal diagnosis would make such patients be born in tertiary centers, where they could rapidly be managed. Better medical transportation of patients during referral, with correct intravenous hydration, would also avoid critical dehydration [4]. The high mortality of IA was due to postoperative septic shock. Whether the latter was linked to preoperative or postoperative infection was not reported in medical files. Studies with a larger sample than ours (5 patients) would allow a better understanding of mortality in this particular group. The unavailability of PN would also play a role in mortality of such patients.

### Limitations

In our study, a total of 12 cases were not included. This was mainly linked to the lack of important information on their medical records. Including a more significant number of patients would be better for a better representation of rare digestive malformations. Secondly, results of prenatal US were retrieved only in 36 of the 230 included cases. This is a crucial point whose impact on outcomes should be analyzed [13]. The same applies to pregnancy terms, as prematurity is linked with higher mortality in these malformations. The causes of mortality were reported only in a few patients, denying better analysis of high mortality in our patients. A systematic collection of data, with the in-hospital register of congenital malformations, would help to provide better registration of data and then allow additional prospective studies on factors associated with mortality in these malformations, including deep analysis of patients, surgeons, and system factors.

### Conclusion

Our study on congenital malformations of the GIT highlighted that prenatal diagnosis was rare in our environment. The majority of patients were diagnosed beyond the neonatal period. The mortality in our study was very high, particularly in intestinal atresia, esophageal atresia, and polymalformation. Further studies, preferably prospective, on the analysis of mortality and its risk factors are required as they would provide a better understanding of this high mortality and then allow actions to improve outcomes of these malformations in our environment.

### Abbreviations

ARM(s)	Anorectal malformation(s)
ARNCHC	Albert Royer National Children's Hospital Center
CHD	Congenital heart disease

CM(s)	Congenital malformation(s)
DSD	Disorder of sexual development
EA	Esophageal atresia
GIT	Gastrointestinal tract
HD	Hirschsprung's disease
HICs	High-income countries
IA	Intestinal atresia
LICs	Low-income countries
LMICs	Low- and middle-income countries
MICs	Middle-income countries
PN	Parenteral nutrition
SD	Standard deviation
SSA	Sub-Saharan Africa
US	Ultrasound
VACTER-L	Vertebral, anorectal, cardiac, tracheo-esophageal, renal, limbs

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### Authors' contributions

FTAZ analyzed the interpreted findings and wrote the manuscript. PAM participated in data acquisition and was a major contributor in writing the manuscript. DG interpreted the data and proceeded to a critical review of the manuscript. RN acquired data, analyzed, and interpreted them. IBW acquired data, analyzed, and interpreted them. NFS participated in the study design, acquired data, and contributed to writing the manuscript. MF analyzed the data and interpreted them. NAN participated in the conception of the study, interpreted the data, and proceeded to critical revision of the manuscript. AS participated in the study design and in the critical revision of the manuscript. ON participated in the conception of the study and made critical revisions to the manuscript. GN participated in the conception and design of the study, interpreted the data, and made a significant contribution by critically revising the manuscript. He is the guarantor of the present work. All authors read and approved the final manuscript.

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### Availability of data and materials

The datasets used and analyzed during the current study are available from the corresponding author upon reasonable request.

### Declarations

#### Ethics approval and consent to participate

Our review received approval from the Ethics Committee of ARNCHC. Seeing its retrospective aspect, consent to participate was waived by the committee.

#### Consent for publication

Not applicable.

#### Competing interests

The authors declare that they have no competing interests.

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