Congenital malformation of the gastrointestinal tract in Aseer region, Saudi Arabia

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ABSTRACT

Objective: The aim of this prospective study was to evaluate the prevalence and pattern of congenital malformations of the gastrointestinal tract among the Saudi newborn population in Aseer region, Kingdom of Saudi Arabia.

Methods: Every consecutive newborn admitted to the neonatal intensive care unit of Aseer Central Hospital, Kingdom of Saudi Arabia with features of gastrointestinal tract anomaly during the period January 1995 to December 2000 had relevant data obtained and entered into a program form.

Results: During the 6 year period, a total of 1386 Saudi infants were admitted into the neonatal intensive care unit of Aseer Central Hospital. Of these, 12.4% were confirmed to have congenital malformation of the gastrointestinal tract; male/female ratio of 1.7:1. The total number of live births by Saudi mothers in Aseer region during the period was 128,093, giving an incidence rate of 1.3 per 1000 live births. The 172 newborns presented with 174 anomalies of the gastrointestinal tract. The leading malformations were imperforate anus (78 cases or 44.8%), tracheosophageal fistula/atresia (42 cases or 24.1%) and intestinal atresia (37 cases or 21.3%). Other lesions included Hirschsprung's disease (14 cases or 8%) and stenosis (2 pyloric and one duodenal) (1.7%). Some patients had more than one defects within the tract (1%) and multisystemic defects (23%). The overall fatality rate was (12%), due largely to post-operative infection (75% of cases) and multiple anomalies (25% of cases).

Conclusion: The prevalence of congenital defect of the gastrointestinal tract in Aseer region appears to be high. The incidence of associated multisystemic anomalies is also high. Fatality incidence is influenced by post-operative sepsis and associated multiple defects. A high incidence of consanguineous marriage in the region may be the underlying etiological factor hence genetic counseling may be helpful.

Keywords: Gastrointestinal tract malformations, incidence.

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To our knowledge, a comprehensive account focusing specifically on congenital anomalies of the alimentary tract in the Kingdom of Saudi Arabia (KSA) has not yet been documented. In a previous general survey on congenital malformations in Aseer region of the Kingdom, the alimentary system was found to be the system most commonly affected (43%).[1] The alimentary tract therefore constitutes an important site of malformation in Aseer province and perhaps in the entire KSA. The aim of this survey, therefore, is to determine the prevalence and the pattern of congenital lesions of the gastrointestinal tract (GIT) among Saudi infants in the Southwest region of Saudi Arabia. This report represents our experience with consecutive admissions of Saudi newborns admitted for congenital deformities of the

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GIT in the neonatal intensive care unit (NICU) of Aseer Central Hospital (ACH), KSA, over a period of 6 years. For genetic and racial homogeneity, the study is confined to infants of Saudi nationality. For the purpose of this study, the terms, gut, alimentary tract and gastrointestinal tract, are used interchangeably. This communication deals with congenital anomalies involving the esophagus contiguously down to the anus. Lesions in the mouth, pharynx and extra-GIT organs such as the liver, pancreas and gall bladder were not considered. We apply the term multiple or complex where malformations occur in more than 2 body systems.

Methods. This is a prospective study which involves all Saudi newborns admitted and treated in ACH with confirmed diagnosis of congenital malformation of the GIT. The cases were conjointly managed by the pediatric surgery and neonatology teams. Every consecutive newborn admitted with features of GIT anomalies during the period January 1995 through December 2000 had relevant data obtained and entered into a program form. The data of interest included date of birth, age of admission, sex, place of birth, nationality and clinical features. The pre-operative and intra-operative findings and other defects outside the GIT and number of days in the hospital, were also recorded. The diagnosis of GIT anomaly was derived from the history, radiographic studies and surgical findings. No autopsy examination was performed on any of the infants who succumbed. Aseer Central Hospital is a referral, tertiary care institution with a dedicated pediatric and neonatal surgical service. The NICU of the hospital is purely referral since ACH has no obstetric service of its own. Sick newborns in Aseer Region (population 2 million) requiring specialized investigations and care are referred to the NICU from 18 general hospitals, 238 primary health care centers and private hospitals. Indeed, ACH is the only institution in Aseer region with facilities for pediatric and neonatal surgery. The NICU in ACH has facilities to provide level III service. It delivers mechanical ventilation, total parenteral nutrition and other basic requirements. There are facilities for plain radiography, ultrasound, computerized tomography, magnetic resonance imaging and contrast studies of the GIT among others. Infants admitted with congenital malformation of the GIT are routinely investigated to exclude deformities in the vertebral column, cardiovascular systems and genitourinary tract and other systems as indicated. Management of the cases involved routine surgical correction of the defect, intravenous infusion of dextrose saline, total parenteral nutrition (TPN), blood transfusion and antibiotics, if necessary. Other modalities of management were as indicated. Figures of deliveries in the population were obtained from the Ministry of Health.

Results. During the 6-year period of study, 1386 Saudi infants were admitted into the NICU of ACH. Of this number, 172 or 12.4% were confirmed to have congenital malformation of the alimentary tract. The total number of live births by Saudi mothers in Aseer region during the 6-year period was 128,093, giving an incidence rate of 1.3 per 1000 live births. **Table 1** shows birth incidence and the yearly admission of Saudi infants with GIT malformation as a proportion of the total yearly admissions into the NICU. These were made up of 109 males and 63 females, a male/female ratio of 1.7:1 **(Table 2)**. All the cases were referred within 24-48 hours of birth. Patients with imperforate anus had colostomy or anoplasty carried out within 6-12 hours of admission and if their post-operative course was satisfactory, they were subsequently transferred back within 2-3 days to the referring hospital with appointment for follow-up or final definitive surgery. Patients with tracheosophageal fistula (TOF), intestinal atresia and Hirschsprung's disease were hospitalized for periods ranging from 13-26 days post-operatively. The 172 newborns presented with 174 anomalies of the GIT; some patients had more than one lesion within the tract. The leading malformations were imperforate anus (78 cases or 44.8%), TOF (42 cases or 24.1%) intestinal atresia (37 cases or 21.3%). Other lesions included Hirschsprung's disease (14 cases or 8%) and stenosis (2 pyloric and one duodenal) (1.7%).

Imperforate anus. The 78 newborns with imperforate anus were consisted of 56 males and 22 females. One of the cases died before the anatomical type could be determined. The others were 35 low, 34 high and 8 intermediate types. Seven of the males had recto-urethral fistula and 7 of the females had recto-vestibular fistula. Associated anomalies in **(Table 3)** this group consisted of Down's syndrome (8 of cases), ambiguous genitalia 5 (3 males and 2 females), polycystic kidneys and cleft lip (one case each). Two of the infants had associated ileal atresia and TOF.

Tracheosophageal fistula/atresia. Forty-two infants (25 males and 17 females) presented with tracheosophageal anomaly. These consisted of proximal esophageal atresia with distal fistula (41 or 97.6%) infants and one case with proximal and distal atresia. Associated anomalies in this group of patients included cardiac defect, hydronephrosis, hypospadias (2 cases each), VATER association, situs inversus, cleft palate (one case each).

Intestinal atresia/stenosis. The intestinal lesions encountered 2 patients with hypertrophic pyloric stenosis, one duodenal stenosis, 8 duodenal atresia, 10 jejunal atresia, 15 ileal atresia and 4 with colonic atresia. One patient had a duplication cyst of the ileum. Annular pancreas was involved in one case of duodenal stenosis and 4 of duodenal atresia. The associated anomalies included cardiac malformation (3 cases), Down syndrome, renal, vertebral and multiple anomalies (one case each).

Table 1 - Number of infants admitted per year and those with GIT malformation in the years 1995-2000.

Year	n admitted	n with GIT anomalies	%
1994	214	52	24.3
1995	223	21	9.4
1996	288	17	5.9
1997	219	25	11.4
1998	211	29	13.7
1999	231	28	12.1
Total	**1386**	**172**	**12.4**

n- number, GIT - gastrointestinal tract

Table 2 - Sex distribution and incidence of GIT malformations seen in 172 neonates admitted in Aseer Central Hospital (1995-2000).

Type of malformations	n of males	n of females	Total (%) n	Birth incidence
Imperforate anus	56	22	78 (44.8)	0.6/1000
TOF	25	17	42 (24.1)	0.3/1000
Atresia	20	17	37 (21.3)	0.29/1000
Hirschsprung disease	7	7	14 (8)	0.1/1000
Stenosis†	2	1	3 (1.7)	0.02/1000
Total	**110**	**64**	**174* (100)**	

* 2 patients with imperforate anus also had ileal atresia and TOF respectively, † - 2 pyloric one doudenal, n - number, TOF - tracheosophageal fistula

Hirschsprung's disease. In the 14 patients with Hirschsprung's disease the lesion was sited in the recto-sigmoid in each of the cases. Three of the infants (21.4%) had Down's syndrome (the ages of their mothers were 40, 41 and 45 years).

Outcome. Twenty (11.6%) of the 172 patients died. Of the 20 fatalities, 15 (75%) was related to septicemia, and the other 5 (25%) to co-existing multiple/complex anomalies.

Discussion. This study is limited to infants with defects that were clinically symptomatic during the newborn age bracket. It undoubtedly misses out some newborns whose congenital lesions which, though present at birth, were silent or could have been misdiagnosed during the neonatal period. For example, H-type TOF, ultra-short-segment Hirschsprung's disease and intestinal stenosis, are some of the lesions that may not be symptomatically obvious in the early days of life. Such cases might not have been transferred to our purely referral NICU. Taking all these factors into consideration, our figures on birth incidence and admission frequency are very likely to be an underestimation of the real situation. Nevertheless, since ACH is the only referral center for neonatal surgery in Aseer region, it can be assumed that the NICU had pooled the bulk of Saudi babies born with major GIT anomalies in the area. It is therefore hoped that the findings of this survey provides a clue as to the pattern of congenital GIT lesions in our environment and perhaps the entire KSA. The study has revealed that congenital malformation of the alimentary tract alone constitutes up to 12.4% of admission in our NICU. It gives an impression that more than one Aseer infant per 1000 will be born with defect(s) of the gut. This implies that alimentary tract malformation contributes significantly to morbidity and mortality in Aseer region of KSA. In descending order of frequency, the major lesions include anorectal malformation, TOF with atresia, intestinal

Table 3 - Extra-GIT anomalies associated with malformation of the alimentary tract in 172 newborns.

Type of malformations	Renal	GU	Cardiac	DS	Vertebral	Multiple	Total (%) n
Imperforate anus	1	5	0	8	0	7	21 (27)
TOF	2	2	3	0	0	1	8 (19)
Atresia	1	0	3	1	1	1	7 (18.4)
Hirschsprung disease	0	0	0	3	0	0	3 (21.4)
Stenosis	0	0	0	0	0	0	0 (0)
Total	**4**	**7**	**6**	**12**	**1**	**9**	**39 (22.7)**

n - number, GU - genitourinary, GIT - gastrointestinal tract, DS - Down's syndrome, TOF - tracheosophageal fistula

atresia and Hirschsprung's disease. Males are more affected than females in every type of anomaly except Hirschsprung's disease in which the sex ratio appears equal (**Table 2**). The male preponderance seen in this survey corroborates the observation of other researchers.[2,3,4] The birth incidence of anorectal malformation (0.6:1000 live births) observed in Aseer region is 3-fold higher than the global figure (1:5000 live births).[5] That imperforate anus was the dominant lesion in our series is in consonance with the observation made from a study in Al Qassim in the central region of KSA.[2] The incidence of associated anomalies in patients with this lesion can be as high as 50% with vertebral and genitourinary malformation being the most common.[6] In our study the frequency of associated anomaly was much less (27%); no vertebral anomaly was recorded, but the most prevalent accompaniments were Down's syndrome (10%), complex/multiple lesions (9%), and defects in the genitourinary tract (6.4%). The bulk (41/42 or 98%) of patients with esophageal anomaly presented with a combination of proximal atresia with distal TOF which is in keeping with the global experience.[7,8] The frequency of associated anomalies was 19% in our study. In intestinal atresia the most affected sites in order of frequency are jejunoileal, duodenal and colonic; 55% of intestinal atresias occur in the jejunum and ileum.[5] In the 21 cases reported in United Arab Emirate, there were 10 patients with duodenal atresias; 11 had jejunoileal atresias, 2 of them with multiple jejunal atresias and in one of the 2 there was an associated colonic atresia.[9] This observation is in consonance with ours in that the bulk (25/37 or 67.5%) of the atresia was located in the jejunoileal segment of the intestine. Failure of the gut to recanalize during the 8th to 10th week of gestation seems to be the most likely aetiology of duodenal atresia. In jejunum, ileum and colon, vascular compromise early in gestation may be responsible for the bowel atresia.[10] The incidence of associated anomalies and congenital duodenal obstruction is variable ranging from 13%[11] to as high as 78%.[12] The association of congenital duodenal obstruction and Down's syndrome is well known with an incidence of 30-50%[5] but a low incidence of 11% and a high incidence of 70%[12] have also been reported. In our series, only one case of Down's syndrome (12.5%) was recorded among the 8 patients with duodenal atresia, which appears to represent a low incidence. Globally, colonic atresia accounts for approximately 10% of intestinal atresia;[5] this is similar to the 10.8% derived from the present study. Hirschsprung's disease is diagnosed in 1:5000 live births and there is a male preponderance of 4:1.[13] This contrasts with the present study which reveals a much lower birth incidence (1:10,000 live births) and an equal sex incidence. Perhaps a lot of Hirschsprung babies in our population might not have been referred during the neonatal period. According to Landman,[14] only 15% of infants with Hirschsprung's disease are diagnosed within the first month of life, 64% by the 3rd month and 80% by one year; 8% may not be recognized until 3-12 years of age.[14] The diagnosis can be relatively delayed in patients with short-segment disease; 45% of these cases are not identified until 19 months to 7 years of age.[15] Approximately 81% of children with ultra-short segment Hirschsprung's disease (5 cm or less), are not diagnosed until 18 months to 14 years of age.[16] In view of all these possible aberrations, our figure on this defect should be interpreted with caution. Congenital GIT lesions can occur at multiple sites in one individual. For instance in the present survey, 2 infants were identified to have 2 lesions each (imperforate anus with ileal atresia; imperforate anus with esophageal atresia/fistula). This underscores the importance of searching for other anomalies along the tract, when one defect has been identified. Generally, the study reveals multi-systemic lesions among a significant number (23%) of infants (**Table 3**) thus reflecting the polymorphous nature of congenital malformation. This finding might have been utilized in formulating a syndromal diagnosis but the preliminary nature of this survey precluded a venture on this perspective. In the United Arab Emirate the major factors which contributed to a high mortality among neonates with congenital defects of the GIT were lack of parenteral nutrition, late presentation and associated multiple anomalies.[9] In contrast, our patients reported early, total parenteral nutrition was easily available but the high mortality frequency was attributed to associated complex anomalies and post-operative infection probably due to long period of hospitalization. This calls for a greater regard for aseptic approach in the handling of all newborns under our care. Consanguinity has been shown to be associated with increased prevalence of congenital anomalies.[17] Although the consanguinity among the parents was not part of the present survey, it is reasonable to hypothesize that the high consanguinity rate in Aseer region (54%)[18] may contribute to the high frequency of GIT anomalies in the population. It may therefore be wise to advocate the use of genetic counseling to reduce the high incidence of congenital gut lesions in this population.

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