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LETTERS TO THE EDITOR

Hirschsprungs disease in Western Saudi Arabia

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Hirschsprungs disease in Western Saudi Arabia

Sir,

Hirschsprungs disease (aganglionic megacolon) demonstrates a predilection for males. Its incidence, reported in the west, is one in 5000 newborns.1 Approximately 90% of patients presented in infancy, usually with constipation and abdominal distension, but diarrhea may occur. The anus is typically normal, but the anal canal and rectum are usually small and devoid of stool. In classic cases, these physical findings are confirmed with barium enema, the contrast material flows into an unexpanded distal segment, then passes through a cone-shaped area, and finally into the dilated proximal bowel. The pathologic change is, of course, aganglionosis. The narrowed distal segment shows complete absence of ganglion cells from both the submucosal and myenteric plexi, usually accompanied by hypertrophy of the muscularis mucosae and increased in numbers and size of nerves in the submucosa and between the muscle layers of the muscularis propria. In the tapered or cone-shaped region, the number of ganglion cells may be decreased.1 The frequent disease, which is rarely reported, lead us to analyze the 27 consecutive cases of endoscopic colonic biopsies and surgical colectomy specimens of both male and female cases presented with this disease in the Department of Surgical Pathology, King Abdul Aziz University Hospital, Jeddah, Kingdom of Saudi Arabia. Data on all colonic biopsies and colectomies carried out between January 1996 and December 2000 and was retrieved from the records of the laboratory keeping track of

age, histopathological diagnosis and frozen section diagnosis. Histopathologically, these cases were analyzed and classified, and the slides were also reviewed if the pathology records were not sufficient for precise diagnosis. We received 27 cases for investigation of ganglion cells for the Hirshsprung disease and related disorders (9.7% of total colonic biopsies) and 22 (81%) of these had gone through intraoperative frozen section examination for on time and rapid diagnosis of the length of aganglionic segment of the colon. The mean age of presentation was 3.33 (number (N)=27, mean age=3.33, standard error=0.50, median=2, mode=1.5, standard deviation (SD)=2.62, variances=6.85) with a prominent predilection for males. Out of 27 cases 18 (66.7%) were males and 9 (11.4%) were females. The average length of the aganglionic segment in these cases was 10.5 cm (range 4-20cm). The site of the lesion was mostly limited to the rectum with 15 (55.6%) cases. Sigmoid colon 3 (11.1%) cases, traverse colon one (3.7%) case, descending colon 3 (11.1%) cases and unspecified colostomy site 5 (18.%) cases. All the 27 (100%) patients presented with constipation, 15 (55.5%) patients also had abdominal distension and no patient presented with diarrhea. Hirschsprungs disease is a frequent congenital disorder (one in 5000 newborns) that results from lack of coordinated propulsive movement of the distal portion of the large bowel resulting from loss of intrinsic inhibitory innervation.2 This is caused by the absence of parasympathetic ganglion cells in the intramural and submucosal plexuses, which in turn may be caused by either failure of migration from the neural crest or by immunemediated neuronal necrosis. It is diagnozed during the first year of life in most patients, but it may present later, occasionally even in adulthood.3 Approximately 80% of the patients are male; 10% have Downs Syndrome and another 5% have other serious neurologic abnormalities. Cases have also been associated with intestinal atresia and anorectal malformations.2 Familial cases have been described, and a candidate gene for the disease has been located in chromosome 10.4 No such risk factor was analyzed in our present study but our intention is also to throw light on these genetic factors that could be more extensively analyzed and correlated in our population.2 The symptoms usually begin shortly after birth, with abdominal gaseous distention, delayed meconium passage, and tight anus.2 In our study, only 3 symptoms were looked for and all the 27 (100%) patients presented with constipation, 15 (55.5%) patients had abdominal distension and no patient presented with diarrhea. With the passage of time, the large bowel proximal to the lesion undergoes dilatation of the lumen and the hypertrophy of the muscular wall, whereas the diseased segment appears grossly normal. The most important complications are

the acute intestinal obstruction and entercolitis.2 Microscopically, the hallmark of the disease is the absence of ganglion cells (aganglionosis) in both plexuses of a segment of bowel. This is associated with the presence of hypertrophied, disorganized, non-myelinated nerve fibers of both adrenergic and non-adrenergic type in the aganglionic segment, which apparently tail to properly innervative muscle layers of the bowel.2 There may also be fibromuscular dysplasis in the arteries located in the transitional zone between the aganglionic and the dilated segment and hyperplasia of lymphoglandular complexes (the latter as an expression of secondary diversion colitis).2 The traditional approach to morphologic documentation of Hirschsprungs disease is the biopsy procedure described by Swenson et al,5 in which a full-thickness segment of the muscular wall of the rectum is exercised and examined for the presence of ganglion cells in the myenteric plexus. Since ganglion cells are normally scanty near the internal anal sphincter, the standard guideline is that the biopsy should be taken at a point 2 cm above the anal valve in infants and 3 cm in the older children. Most cases are examined in a frozen section. Many pathologists prefer to examined a frozen-section slide stained for acetylcholinesterase in addition to standard Hematoxylin and Eosin (H&E) sections. In Hirscsprungs disease, the acetylcholinesterase stain demonstrates a marked increase in the acetylcholinesterase-positive nerve fibers in the lamina propria and muscularis mucosae. The utility of this technique as an adjunct to diagnosis is debated. False-positive and false-negative reactions have been reported, and its use is a matter of personal preference.6 Occasionally,ganglion cells may be difficult to identify using light microscopy alone, especially in the neonate. In such cases, a positive immunocyto-chemical reaction for neurospecific enolase can be invaluable in documenting ganglion cells.6 A wider population study for the genetic and other risk factors of this frequent congenital disease is highly recommended in our population. The proper aid of immuno-histochemistry for the accurate diagnosis of this disease is also recommended.

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