Improving the accuracy of contrast enema in diagnosis of Hirschsprung disease

A THESIS SUBMITTED FOR PARTIAL FULFILMENT OF MASTER'S DEGREE IN RADIOLOGY

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List of abbreviations

AXR	Abdominal X-ray
CE	Contrast enema
DCBE	Double contrast barium enema
ENS	Enteric nervous system
GI	Gastrointestinal tract
HAEC	Hirschsprung's associated enterocolitis
HD	Hirschsprung's disease
RSI	Rectosigmoid index
TCA	Total colonic aganglionosis
TZ	Transitional zone
WSCE	Water soluble contrast enema

Introduction

Hirschsprung's Disease is a developmental disorder caused by premature arrest of the migration of the neural crest cells along the hind gut leading to absence of ganglionic cells in the myenteric and submucosal plexuses of the distal intestine resulting in functional obstruction. The usual presentation of HD is a full-term neonate with delayed passage of meconium, distended abdomen, feeding intolerance with bilious aspirate or bilious vomiting. (**Singh and Baruah**, **2016**)

It has incidence of 1:5000 live birth, with male: female ratio of 4:1. HD was reported to be the etiology in up to 13.85% of pediatric intestinal obstruction cases.(**PEYVASTEH et al.**, **2016**)

HD patients may be asymptomatic in childhood and are diagnosed in adolescence and puberty with nearly 15% are diagnosed after the age of 5. The gold standard for diagnosis is pathological analysis that reveals absence of ganglion cells in the sub mucosa and the myenteric plexus which lead to aperistalsis in the affected region resulting in functional intestinal obstruction.(**Alehossein et al.**, **2015**)

Pathological analysis sample is acquired either through suction biopsy or full thickness biopsy. 13% of suction biopsy

acquired samples yield false negative result due to inadequate biopsy cases and full thickness biopsy result in severe complications. Moreover only 12-17% of chronic constipation patients who underwent rectal biopsy were HD Patients. Concluding that up to 80% of patients were exposed to unnecessary risk of biopsy complications. (**Guo et al.**, **2006**)

Although full thickness biopsy is the gold standard to diagnose HD, the associated risks related to it as perforation, scarring, stricture and bleeding in addition to anesthesia- related adverse effects leads to the rise of importance of non-invasive methods such as CE and anorectal manometry to diagnose HD. Anorectal manometry is not available in many institutions with CE widely available in many centers without the need of a pediatric surgeon. CE is not just helpful in diagnosing the disease but also in pre-operative planning by demonstrating the level of agangliosis which is most reliably evaluated by the transitional zone, some studies fail to show matching result of aganglionosis by histopathology.(**Alehossein et al.**, **2015**; **PEYVASTEH et al.**, **2016**)

Aim of work

A scoring system using a checklist of radiologic and clinical signs was developed and used for a more accurate diagnosis of Hirschsprung disease using contrast enema, and the sensitivity and specificity were determined in comparison with biopsy, as a gold standard method.

Embryology and anatomy of the large intestine

Embryology of the large intestine

During embryonal development, mitotic cell division produces three germ layers - ectoderm, mesoderm, and endoderm, with the embryo being a trilaminar disc. The surface facing the yolk sac becomes the endoderm, the surface facing the amniotic sac becomes the ectoderm, and the middle layer is the mesoderm. At 4 weeks gestation by process of folding the flat endodermal layer forms a tube which is the primitive gut. **(Figure 1)** (**Bass and Wershil**, **2016**)

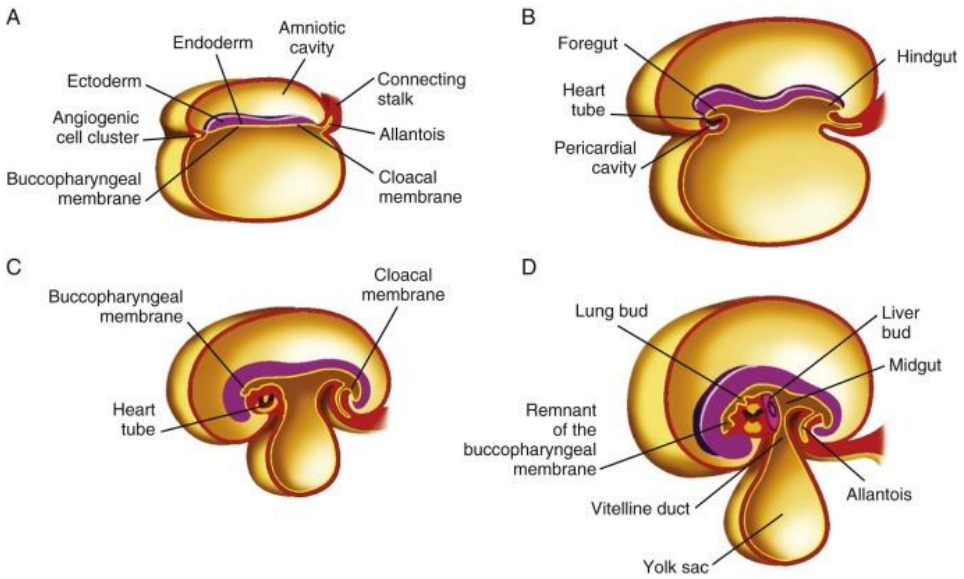

Figure 1 Formation of foregut, midgut, and hindgut **(Bass and Wershil, 2016)**

The primitive gut is divided into three parts, the cranial part is the foregut, the caudal part is the hindgut and the midgut in between. (**Figure 2)** The foregut will give rise to the pharynx, esophagus, stomach and proximal part of the duodenum. The midgut gives rise to distal duodenum, jejunum, ileum, and proximal portions of the colon (caecum, ascending and proximal two thirds of the transverse colon). The distal third of the transverse colon, the descending colon and sigmoid, the rectum, and the upper part of the anal canal originate from the hindgut. (**DAUVÉ and MCLIN**, **2016**)

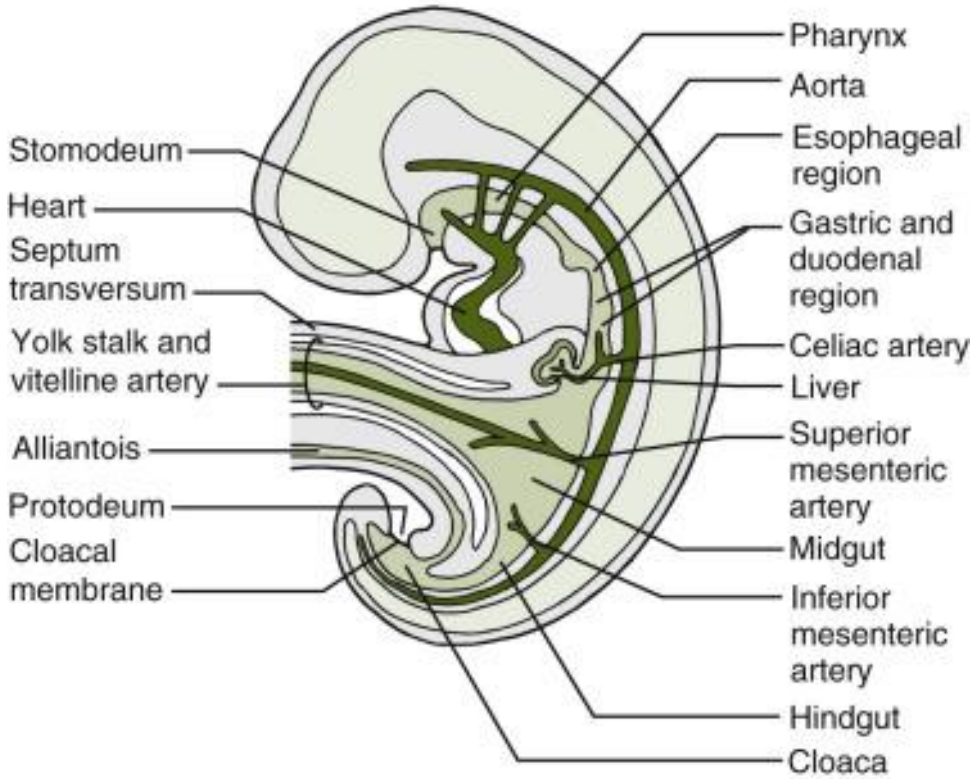

Figure 2 Median section of 4-week-old embryo showing the early digestive system and its blood supply. **Quoted from: (Fiorino and Nurko, 2016)**

The fetal colon develops over 30 weeks in 3 stages. Primitive stratified epithelium similar to that in the small intestine appears between 8 and 10 weeks. Conversion to villus architecture with developing crypts occurs at 12 to 14 weeks. Remodeling to the adult-type crypt epithelium with loss of the villi occurs at 30 weeks. At first the urinary, genital and rectal tracts open in a common opening called the cloaca, which then become separated into the anterior urogenital sinus and a posterior intestinal tract by the caudal descent of the urorectal septum. (**Bass and Wershil**, **2016**)

The hindgut enters the posterior aspect of the cloaca – the primitive anorectal canal – with a membrane forming a boundary between the cloacal ectoderm and the hindgut endodermal lining. This membrane ruptures at 7th week. This membrane also marks the junction between distal and proximal portion of the anal canal. With both portions having different embryonic origin, they have different blood supply and most importantly different innervation. The inferior mesenteric ganglia and the pelvic splanchnic nerves innervate the superior portion of the anal canal. The inferior rectal nerve supplies the inferior rectal canal. **(Bass and Wershil, 2016)**

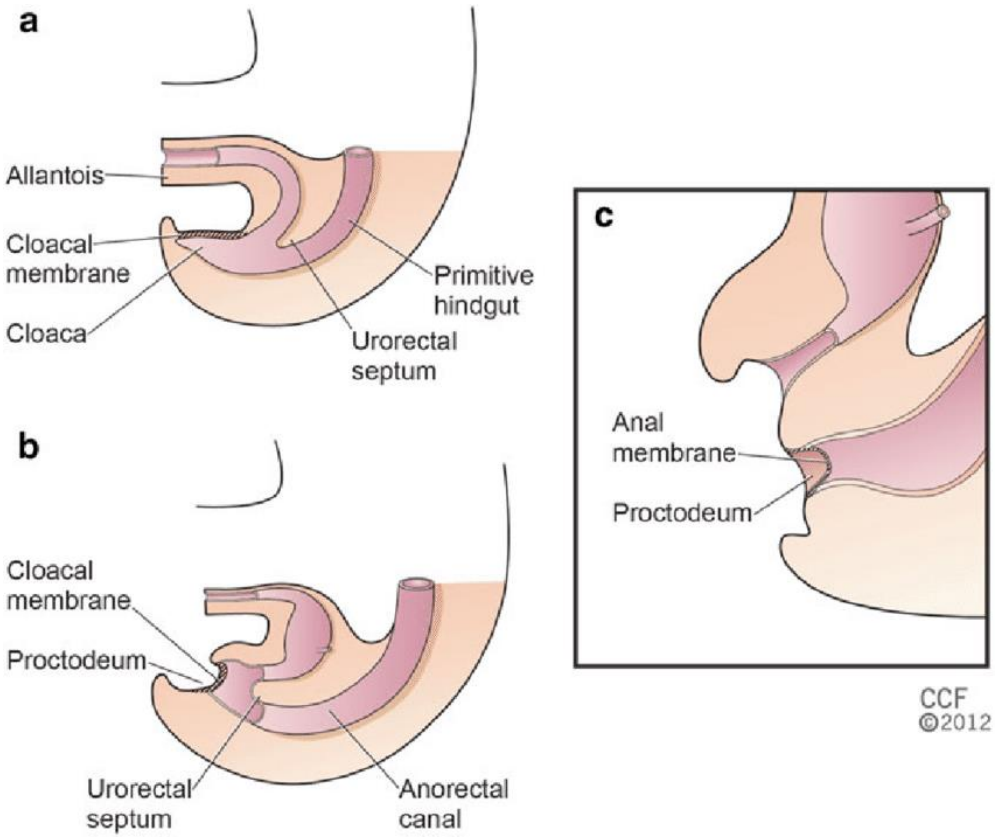

Figure 3 Embryology of the anorectum. (a) The cloaca—the fusion of the hindgut with the allantois and mesonephric ducts—is partitioned by the urorectal septum, creating (b) the urogenital sinus anteriorly and the anorectum posteriorly. The once common chamber terminates blindly at the cloacal membrane, which similarly is divided into the anterior urogenital membrane and posterior anal membrane. (c) The ectodermal layer of the anal membrane gives rise to the surrounding protuberances—the anal folds—that create a central depression, the proctodeum, which ultimately develops into the distal anal canal. **Quoted from** (**Inkster et al.**, **2016**)

The mature intestine develops from all three germ layers: endoderm, mesoderm, and ectoderm. The endoderm gives rise to the simple columnar epithelial cell lining of the surface of the small and large intestines. Cells of the lamina propria and muscularis layers derive from the embryonic mesoderm. While