The gastrointestinal (GI) tract is a hollow tube extending from the oral cavity to the anus that consists of anatomically distinct segments, including the esophagus, stomach, small intestine, colon, rectum, and anus. Each of these segments has unique, complementary, and highly integrated functions, which together serve to regulate the intake, processing, and absorption of ingested nutrients and the disposal of waste products. The regional variations in structure and function are reflected in diseases of the GI tract, which often affect one or another segment preferentially. Accordingly, following consideration of several important congenital abnormalities, the discussion is organized anatomically. Disorders affecting more than one segment of the GI tract, such as Crohn disease, are discussed with the region that is involved most frequently.

CONGENITAL ABNORMALITIES

Depending on both the nature and timing of the insult during gestation, a variety of developmental anomalies can affect the GI tract. Importantly, because many organs develop simultaneously during embryogenesis, the presence of congenital GI disorders should prompt evaluation of other organs. Some defects are commonly associated with GI lesions.

Atresia, Fistulae, and Duplications

Atresia, fistulae, and duplications may occur in any part of the GI tract. When present within the esophagus they are discovered shortly after birth, usually due to regurgitation during feeding. Without prompt surgical repair, these lesions are incompatible with life. Absence, or *agenesis*, of the esophagus is extremely rare, but *atresia*, in which development is incomplete, is more common. In esophageal atresia a thin, noncanalized cord replaces a segment of esophagus, causing a mechanical obstruction (Fig. 17-1*A*). Atresia occurs most commonly at or near the tracheal bifurcation and is usually associated with a *fistula* connecting the upper or lower esophageal pouches to a bronchus or the trachea (17-1*B*). In other cases, a fistula can be present without atresia (Fig. 17-1*B, C*). Either form of fistula can lead to aspiration, suffocation, pneumonia, and severe fluid and electrolyte imbalances. Developmental abnormalities of the esophagus are associated with congenital heart defects, genitourinary malformations, and neurologic disease. Intestinal atresia is less common than esophageal atresia but frequently involves the duodenum. *Imperforate anus*, the most common form of congenital intestinal atresia, is due to a failure of the cloacal diaphragm to involute.

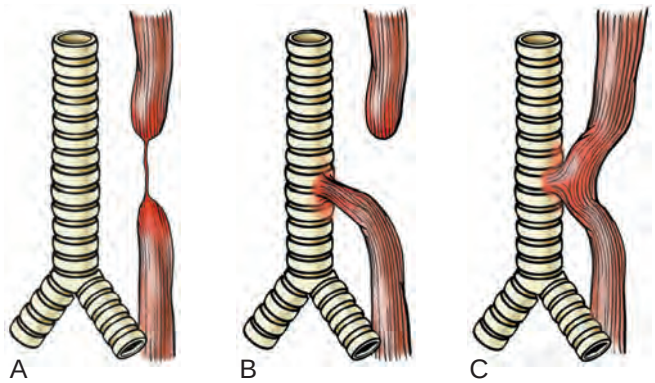

Figure 17-1 Esophageal atresia and tracheoesophageal fistula. **A,** Blind upper and lower esophagus with thin cord of connective tissue linking the two segments. **B,** Blind upper segment with fistula between lower segment and trachea. **C,** Fistula (without atresia) between patent esophagus and trachea. The developmental anomaly shown in **B** is the most common. (Adapted from Morson BC, Dawson IMP, eds: Gastrointestinal Pathology. Oxford, Blackwell Scientific Publications, 1972, p 8.)

Stenosis is an incomplete form of atresia in which the lumen is markedly reduced in caliber as a result of fibrous thickening of the wall. This results in either partial or complete obstruction. In addition to congenital forms, stenosis can be acquired as a consequence of inflammatory scarring, such as that caused by chronic gastroesophageal reflux, irradiation, systemic sclerosis, or caustic injury. Stenosis can involve any part of the GI tract, but the esophagus and small intestine are affected most often.

Diaphragmatic Hernia, Omphalocele, and Gastroschisis

Diaphragmatic hernia occurs when incomplete formation of the diaphragm allows the abdominal viscera to herniate into the thoracic cavity. When severe, the space-filling effect of the displaced viscera can cause pulmonary hypoplasia that is incompatible with life. *Omphalocele* occurs when closure of the abdominal musculature is incomplete and the abdominal viscera herniate into a ventral membranous sac. This may be repaired surgically, but as many as 40% of infants with an omphalocele have other birth defects. *Gastroschisis* is similar to omphalocele except that it involves all of the layers of the abdominal wall, from the peritoneum to the skin.

Ectopia

Ectopic tissues (developmental rests) are common in the GI tract. The most frequent site of *ectopic gastric mucosa* is the upper third of the esophagus, where it is referred to as an *inlet patch*. While generally asymptomatic, acid released by gastric mucosa within the esophagus can result in dysphagia, esophagitis, Barrett esophagus, or, rarely, adenocarcinoma. *Ectopic pancreatic tissue* occurs less frequently and can be found in the esophagus or stomach. Like inlet patches, these nodules are most often asymptomatic but they produce damage and local inflammation in some cases. When ectopic pancreatic tissue is

www.PTools.ir