Radiologic imaging of congenital gastrointestinal anomalies in infants

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Abstract

Background Congenital gastrointestinal anomalies may manifest signs or symptoms in the first few days of life, most commonly in the form of obstructions. Radiologic imaging plays an important role in diagnosis confirmation and surgical correction plans. Most cases may be diagnosed by plain radiographs alone, but CT scans and MRI may be needed to make accurate diagnoses, especially in difficult cases.

Objective To report radiologic imaging findings in infants with congenital gastrointestinal anomalies.

Methods For this retrospective, cross-sectional study we took secondary data from medical records of infants with congenital gastrointestinal anomalies in Dr. Kariadi Hospital, Semarang, Indonesia from January 2010 – June 2011. Diagnosis of congenital anomalies was confirmed by clinical manifestation and radiologic imaging. Radiologic findings were reviewed by a single radiologist on duty at that time. Data is presented in the form of frequency distribution.

Results Subjects consisted of 50 males and 23 females. The most common complaints were vomiting in 14 subjects (19%), abdominal distension in 31 subjects (43%), and fecal passage dysfunction in 28 subjects (38%). Radiologic imaging of subjects with congenital gastrointestinal anomalies revealed the following conditions: anal atresia in 28 subjects (38%), congenital megacolon in 21 subjects (29%), esophageal atresia in 14 subjects (19%), duodenal atresia in 9 subjects (12%), and pyloric atresia in 1 subject (2%).

Conclusion Using radiologic imaging of infants with congenital gastrointestinal anomalies, the most to least common conditions found were anal atresia, congenital megacolon, esophageal atresia, duodenal atresia, and pyloric atresia. [Paediatr Indones. 2012;52:341-5].

Keywords: congenital gastrointestinal anomalies, radiologic imaging, anal atresia, congenital

Congenital anomalies are one of the most common causes of disability in developing countries.\textsuperscript{1} A wide spectrum of congenital anomalies may affect the gastrointestinal tract, some of which manifest at early after birth while others may not present until late childhood or adulthood.\textsuperscript{2,3} Congenital gastrointestinal anomalies may affect the upper gastrointestinal tract, including anomalies of the esophagus (e.g., atresia, fistulas, webs, duplications, and vascular rings), the stomach (e.g., congenital gastric outlet obstruction and duplications), and the duodenum (e.g., atresia, annular pancreas, duplications and malrotation).\textsuperscript{3} Congenital abnormalities involving the small bowel or colon are detected in neonates only when they are direct cause of obstruction.\textsuperscript{4} Clinical signs and symptoms including abdominal distention, vomiting and constipation, lead the clinician to promptly consult to a radiologist to help determine the presence, location, and cause of the obstruction.\textsuperscript{4}

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In normal neonates, swallowing begins almost immediately after birth and gas should be present in the stomach within few minutes. Within 3 hours of birth, the entire small bowel usually contains gas while the sigmoid colon is seen only after 8–9 hours. Disruption of this common pattern is seen in obstruction or in the presence of underlying illness such as brain damage, septicemia or hypoglycemia.²

Plain radiograph is a useful, simple and inexpensive tool in the evaluation of the neonate with gastrointestinal (GI) obstruction. Ultrasound is useful in the evaluation of many congenital anomalies affecting pediatric gastrointestinal tract, especially hypertrophic pyloric stenosis, enteric duplication cysts, midgut malrotation, meconium ileus and meconium peritonitis. Moreover, computerized tomography (CT) and magnetic resonance imaging (MRI) have assumed a greater importance as these methods provide excellent anatomic details which may be necessary for correct diagnosis as well as treatment planning.²

The objective of this study was to determine the type and frequency of congenital gastrointestinal anomalies using radiologic imaging.

Methods

This cross-sectional, retrospective study included all infants with congenital gastrointestinal anomalies who were born or referred to Dr. Kariadi Hospital, Semarang, Indonesia between January 2010 to June 2011. We reviewed the medical records of all infants admitted during this period who were diagnosed with congenital gastrointestinal anomalies. We recorded subjects’ data, including date of birth, sex, weight, chief complaint, diagnosis of congenital gastrointestinal anomalies and radiologic imaging results. Diagnosis of congenital anomalies was reconfirmed to be consistent with the recorded clinical manifestations and radiologic imaging. Radiologic expertise was given by a single radiologist who was on duty at that time. Data is presented in the form of frequency distribution.

Results

Subjects were 73 infants with congenital gastrointestinal anomalies. The majority of subjects were males and the mean age was 5 (SD 5) days. Table 1 shows the characteristics of subjects.

Table 1. Characteristics of subjects

| Characteristics | n = 73 |
|-----------------|-------|
| Mean age (SD), days | 5 (5) |
| Sex             |       |
| Male, n (%)    | 50 (69)|
| Female, n (%)  | 23 (31)|
| Mean body weight (SD), grams | 2800 (481) |
| Clinical symptoms |       |
| Vomiting, n (%) | 14 (19)|
| Abdominal distension, n (%) | 31 (43)|
| Fecal passage dysfunction, n (%) | 28 (38)|

Clinical manifestations of the congenital gastrointestinal anomalies found in our subjects are shown in Table 2. The most common complaints in infants with congenital gastrointestinal anomalies were abdominal distension (31 cases, 43%), fecal passage dysfunction (28 cases, 38%) and vomiting (14 cases, 19%).

Table 2. Clinical symptoms and radiological imaging of congenital gastrointestinal anomalies

| Clinical symptoms             | Anal atresia | Congenital megacolon | Esophageal atresia | Duodenal atresia | Pyloric atresia | Total (n) |
|-------------------------------|--------------|----------------------|--------------------|------------------|----------------|----------|
| Abdominal distension         | 0            | 19                   | 2                  | 9                | 1              | 31       |
| Fecal passage dysfunction    | 28           | 0                    | 0                  | 0                | 0              | 28       |
| Vomiting                     | 0            | 2                    | 12                 | 0                | 0              | 14       |

The most common congenital gastrointestinal anomalies were anal atresia (38%), congenital megacolon (29%) and esophageal atresia (19%). The less common congenital gastrointestinal anomalies were duodenal atresia (12%) and pyloric atresia.
Discussion

Congenital malformations are physical defects present at birth, irrespective of whether they are due to genetic or non-genetic prenatal events. A major birth defect is an abnormality of an organ structure that results in physical disability, mental disability, or death. A minor defect does not produce significant health consequences. Congenital anomalies of the gastrointestinal tract are significant causes of morbidity in children and, less frequently, in adults. In our study, there were 73 cases of infants with congenital gastrointestinal anomalies during the period of study.

The mean age of our subjects was 5 (SD 5.0) days and most clinical manifestations were present in the first few days of life. Most congenital anomalies that affect the gastrointestinal tract manifest early after birth, while others may not present until late childhood or adulthood. Of our 73 subjects with congenital gastrointestinal anomalies, 69% were male (n=50) and 31% were female (n=23). Sun et al. reported that males comprised 65% of their total cases of intestinal and anorectal malformations. Similarly, Cho S et al. observed that the sex distribution of all 103 patients with anorectal malformations was 67% males and 33% females.

We found that the clinical manifestations in infants with congenital gastrointestinal anomalies

| Diagnoses              | n (%) |
|------------------------|-------|
| Anal atresia           | 28 (38) |
| Congenital megacolon   | 21 (29) |
| Esophageal atresia     | 14 (19) |
| Duodenal atresia       | 9 (12) |
| Pyloric atresia        | 1 (2) |

Table 3. Frequency of congenital gastrointestinal anomalies confirmed by radiologic imaging

Figure 1. Radiologic imaging of anal atresia. A, X-ray babygram shows there were no air in pelvic cavity. B, x-ray knee-chest position shows the distance between anal dimple and air of intestine is > 1.5 cm

Figure 2. Radiologic imaging of short segment congenital megacolon. A, X-ray babygram shows colon dilatation. B, Supine anteroposterior barium enema and C, Lateral barium enema shows cone shape transition at rectosigmoid and rectosigmoid index less than 1 cm

Figure 3. Radiologic imaging of anal atresia. A, X-ray babygram shows there were no air in pelvic cavity. B, x-ray knee-chest position shows the distance between anal dimple and air of intestine is > 1.5 cm
were abdominal distension 43% (n=31), fecal passage 38% (n=28) and vomiting 19% (n=14). The usual presentation of high intestinal obstruction is vomiting which may be bile-stained if the obstruction is distal to the ampulla of Vater. Failure to pass meconium in the first 24-48 hours of life may be due to low intestinal obstruction including ileal or colon atresia, anorectal malformations, Hirschprung’s disease, meconium plug syndrome or neonatal small left colon syndrome.

In our study, 38% of the congenital gastrointestinal anomalies were due to anal atresia. All 28 cases of anal atresia had fecal passage dysfunction. Anorectal malformations remain a significant birth defect with an accepted incidence of approximately 0.2 -1.2%. Failure to pass meconium, combined with progressive abdominal distension, refusal to feed and vomiting of bilious intestinal contents are the classic clinical signs of intestinal obstruction in neonates. Anal atresia may be characterized as “high” or “low”, depending on whether the rectum ends above the levator muscle or partially descends through this muscle. Perineal inspection reveals the absent anus. The easily available plain radiography is simple and is often the preliminary test used to evaluate these patients. Prone, cross-table lateral view with the infant in a genupectoral position is useful in determining the level of atresia. In our study, diagnoses of anal atresia were confirmed by x-ray babygram and the x-ray in the knee-chest position.

We found that 29% (n=21) of cases had congenital megacolon, of which 19 subjects had abdominal distension and 2 subjects had vomiting. Hirschprung’s disease (congenital megacolon) has an overall incidence of 1 in 5,000 live births. It accounts for 20-25% of neonatal bowel obstruction cases. Symptoms range from neonatal intestinal obstruction to chronic progressive constipation in older children. Approximately 80% of patients in the first few months of life present with difficult bowel movements, poor feeding, and progressive abdominal distension. Imaging may be used to diagnose Hirschprung’s disease, as a plain abdominal radiograph may show a dilated small bowel or proximal colon. Contrast enema radiographs of the colon are commonly normal in the first three months of life and indefinitely in patients with total colonic disease. Radiologic imaging used to confirm the diagnosis of congenital megacolon consisted of plain abdominal radiographs and barium enema.

Esophageal atresia is a relatively common congenital malformation occurring in 1 in 2,500-3,000 live births. From our 73 cases of congenital gastrointestinal anomalies, 19% (n=14) had esophageal atresia. Esophageal atresia and tracheoesophageal fistula are complex of congenital anomalies characterized by incomplete formation of the tubular esophagus or an abnormal communication between oesophagus and trachea. Infants with esophageal atresia are unable to swallow saliva and noted to have excessive salivation requiring repeated suctioning. In our study, we found 12 cases of esophageal atresia who presented with vomiting and 2 cases had abdominal distension. Radiologic diagnosis is based on findings of antero-posterior and lateral chest radiography, which reveals a blind pouch of the proximal esophagus that is distended with air. Radiographic evaluation should always include the abdomen to assess the presence of gas in the gastrointestinal tract. The absence of gas in the stomach or intestinal tract is suggestive of pure esophageal atresia without fistula, or esophageal atresia with proximal tracheoesophageal fistula.

We observed that 12% of subjects had duodenal atresia, and all were admitted with abdominal distension. Duodenal atresia occurs in approximately 1 in 4,000 live births. Dalla Vecchia et al. reported that duodenal obstruction was found in 50% of the neonates enrolled in their study. Their clinical presentations included bilious emesis or aspirates, upper abdominal distension and feeding intolerance. Total failure of recanalization resulting in obstruction accounts for most cases of duodenal obstruction, and partial failure of recanalization of the gut may result in duodenal stenosis, which is slightly more common than duodenal web. The degree of bowel obstruction determines the presence and severity of symptoms. Clinically bile-stained vomiting within the first 24 hours of life is the hallmark of severe stenosis. An atretic lesion may be proximal to the ampulla of Vater, such that the vomitus may not be bile-stained. Conventional radiographs may show the classic “double bubble” appearance.

In conclusion, the congenital gastrointestinal anomalies found in our study from most to least common were anal atresia, congenital megacolon, esophageal atresia, duodenal atresia, and pyloric anomalies.
atresia. We suggest a further population-based study with a larger sample size to determine the birth prevalence, type and distribution of congenital anomalies in the Indonesian population, as well as to look for any correlation between clinical manifestations and radiologic findings to confirm diagnoses of congenital gastrointestinal anomalies.

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