

A 52-hour-old girl in the newborn nursery develops vomiting and abdominal distension. She was delivered at 40 weeks gestation to a 30-year-old woman by cesarean section for failure to progress. Apgar scores were 8 and 9 at 1 and 5 minutes, respectively. The patient has been voiding appropriately but has not yet passed meconium. Today, she has spit up all feeds and now has bilious emesis. Her abdomen is tense and distended. Rectal examination shows no stool in the rectal vault and normal tone. Feeds are held and a nasogastric tube is placed for decompression. Upright abdominal radiograph shows multiple dilated loops of small bowel with paucity of air in the large intestine and rectum. Left lateral decubitus view shows free air above the liver, and an emergency laparotomy is performed. Viscous meconium is irrigated and evacuated, primarily from the ileum with some from the colon. The colon is found to be diffusely narrow. Which comorbidity of her underlying condition will this patient most likely develop?

- ☐ A. Chronic rhinosinusitis
- ☐ B. Early-onset Alzheimer disease
- ☐ C. Hypothyroidism
- ☐ D. Infertility
- ☐ E. Sensorineural hearing loss

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- ✓

☒

A. Chronic rhinosinusitis [41%]
- ☐

B. Early-onset Alzheimer disease [26%]
- ☐

C. Hypothyroidism [5%]
- ☐

D. Infertility [23%]
- ☐

E. Sensorineural hearing loss [6%]

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Explanation:

User Id:

Differentiating features of Hirschsprung disease and meconium ileus		
	Hirschsprung disease	Meconium ileus
Associated disorder	Down syndrome	Cystic fibrosis
Typical level of obstruction	Rectosigmoid	Ileum
Meconium consistency	Normal	Inspissated



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User Id: [REDACTED]

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"Squirt sign"	Positive	Negative

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**Meconium ileus** and Hirschsprung disease (congenital aganglionic megacolon) should be considered in any neonate with **delayed passage of meconium** as 99% of healthy, full-term infants pass stool within 48 hours of birth. These 2 conditions have overlapping clinical features but can usually be differentiated by the level of intestinal obstruction and meconium consistency.

This patient's presentation is most consistent with meconium ileus, the earliest life-threatening manifestation of **cystic fibrosis (CF)**. Meconium ileus is virtually diagnostic for CF. Although only 20% of patients with CF develop meconium ileus, almost all newborns with meconium ileus have CF. A mutation in the CF transmembrane conductance regulator gene results in abnormal chloride and sodium transport and thick, viscous secretions in multiple organs. **Thick, inspissated meconium** is difficult to propel, resulting in obstruction at the level of the ileum and a narrow, underdeveloped colon (**microcolon**). Although contrast enema is typically performed to assess level of bowel obstruction in stable patients, the **free air** (red arrow) above the liver in this patient's radiograph was indicative of intestinal perforation and required emergency surgery.

Nearly all patients with CF develop **sinopulmonary disease**. **Opacification of all sinuses** can be seen as early as age 8 months, and patients often require surgical debridement of their sinuses.



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**(Choices B and C)** Hirschsprung disease is associated with Down syndrome, which in turn is associated with an increased risk of Alzheimer disease and hypothyroidism. It typically presents with increased rectal tone, "squirt sign" (forceful expulsion of stool after rectal examination), and obstruction at the rectosigmoid region with a transition zone between the aganglionic rectum and **proximal dilated colon**. Patients with CF are no more likely to develop Alzheimer disease or hypothyroidism than individuals in the general population.

**(Choice D)** Although men with CF are typically infertile due to congenital absence of the vas deferens, only 20% of women with CF have fertility problems. Infertility is due to the combination of secondary amenorrhea from malnutrition and thick cervical mucus obstructing sperm entry.

**(Choice E)** Approximately 20% of patients with CF develop sensorineural hearing loss due to frequent treatment with aminoglycosides for gram-negative infections (eg, *Pseudomonas aeruginosa*).

#### Educational objective:

Meconium ileus is virtually diagnostic for cystic fibrosis. Inspissated (viscous) meconium is responsible for life-threatening obstruction at the level of the ileum compared to more



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#### Educational objective:

Meconium ileus is virtually diagnostic for cystic fibrosis. Inspissated (viscous) meconium is responsible for life-threatening obstruction at the level of the ileum compared to more typical rectosigmoid obstruction seen in infants with Hirschsprung disease.

#### References:

1. **Rhinosinusitis in cystic fibrosis: not a simple story.**
2. **Gastrointestinal manifestations of cystic fibrosis.**



Media Exhibit

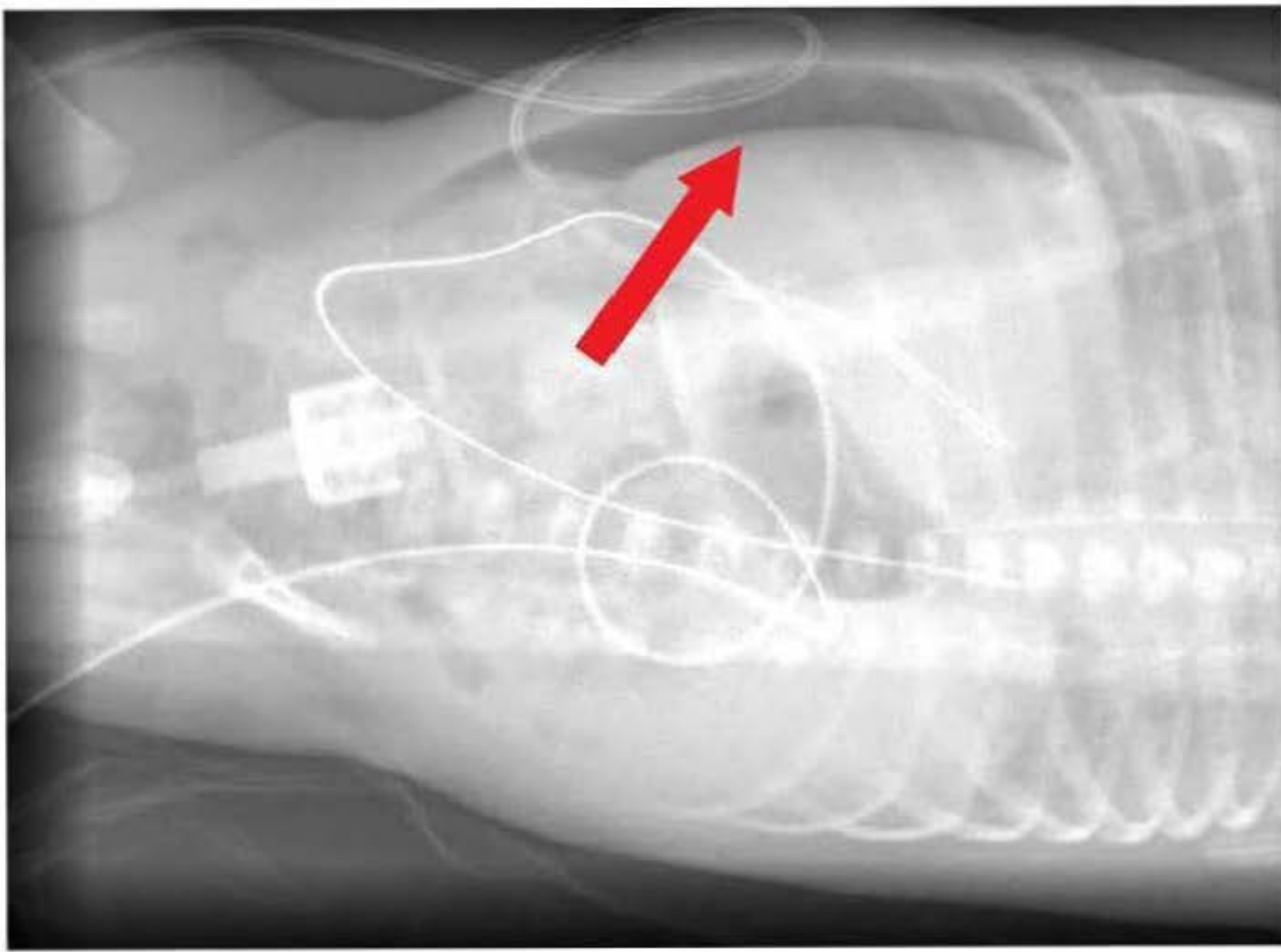
m ileus



Contrast enema  
demonstrating  
microcolon

Media Exhibit

peritoneal Air





Media Exhibit

maxillary sinusitis





Media Exhibit

lung disease

