

A 10-hour-old boy in the newborn nursery has bilious emesis. He was born at 38 weeks gestation by vaginal delivery to a primigravid 39-year-old woman. The mother received no prenatal care until the third trimester, when prenatal ultrasound showed polyhydramnios. The boy has voided once but has not yet passed meconium. Physical examination shows a hypotonic neonate with a flat face, prominent tongue, low-set ears, and slanted palpebral fissures. His hands are short with incurved fifth fingers, and a large space is present between bilateral first and second toes. The abdomen is soft and without distension, guarding, or rigidity. Auscultation shows a loud holosystolic murmur most prominent at the left low sternal border with a precordial thrill. Abdominal x-ray is shown below.



What is the most likely diagnosis in this patient?

☐ A. Biliary atresia

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- ☐ A. Biliary atresia
- ☐ B. Congenital aganglionic megacolon
- ☐ C. Duodenal atresia
- ☐ D. Imperforate anus
- ☐ E. Meconium ileus
- ☐ F. Pyloric stenosis
- ☐ G. Sigmoid volvulus

Submit

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What is the most likely diagnosis in this patient?

☒ A. Biliary atresia [1%]

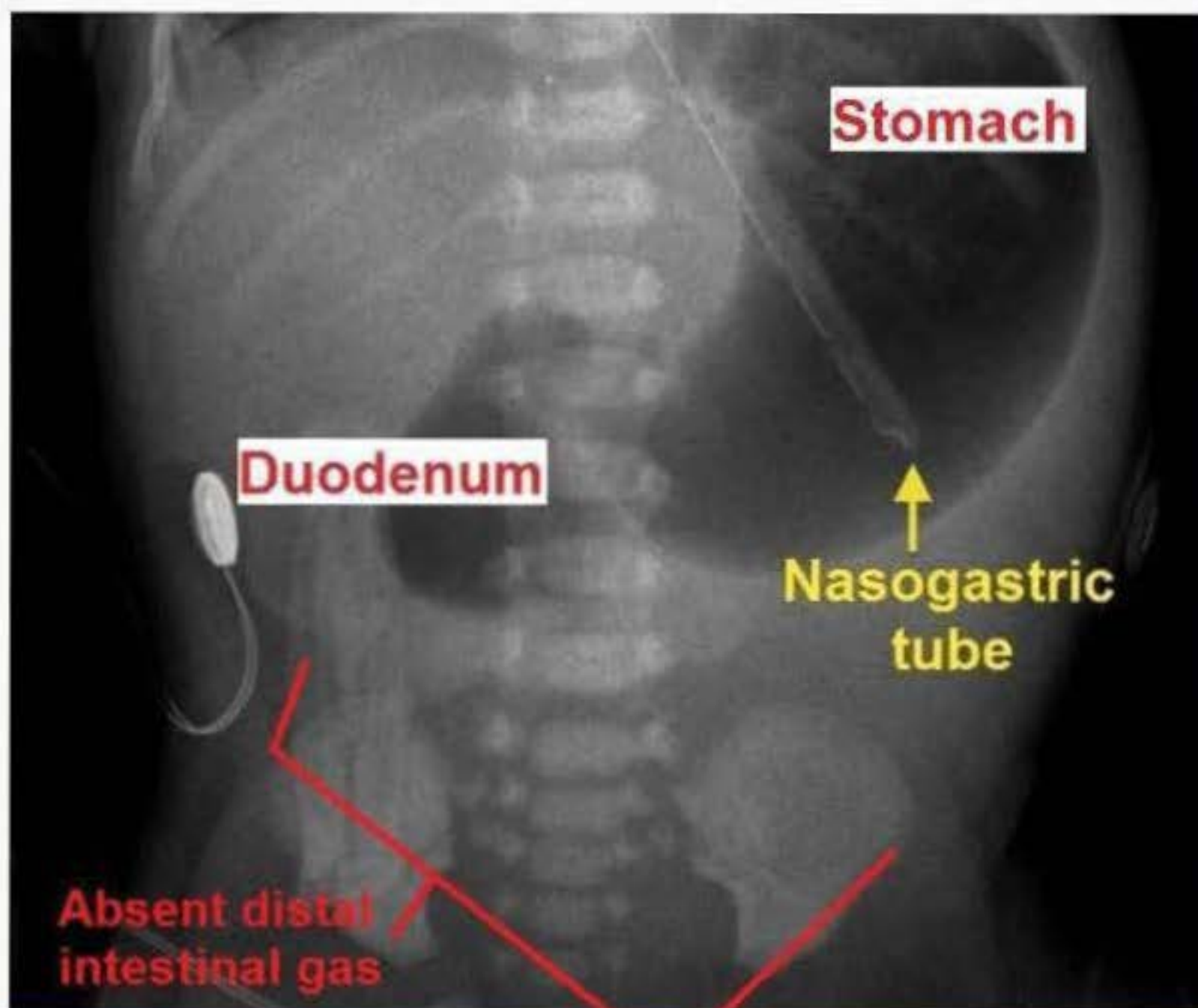
What is the most likely diagnosis in this patient?

- ☐ A. Biliary atresia [1%]
- ☐ B. Congenital aganglionic megacolon [2%]
- ☒ C. Duodenal atresia [93%]
- ☐ D. Imperforate anus [1%]
- ☐ E. Meconium ileus [1%]
- ☐ F. Pyloric stenosis [2%]
- ☐ G. Sigmoid volvulus [0%]

Proceed to Next Item

Explanation:

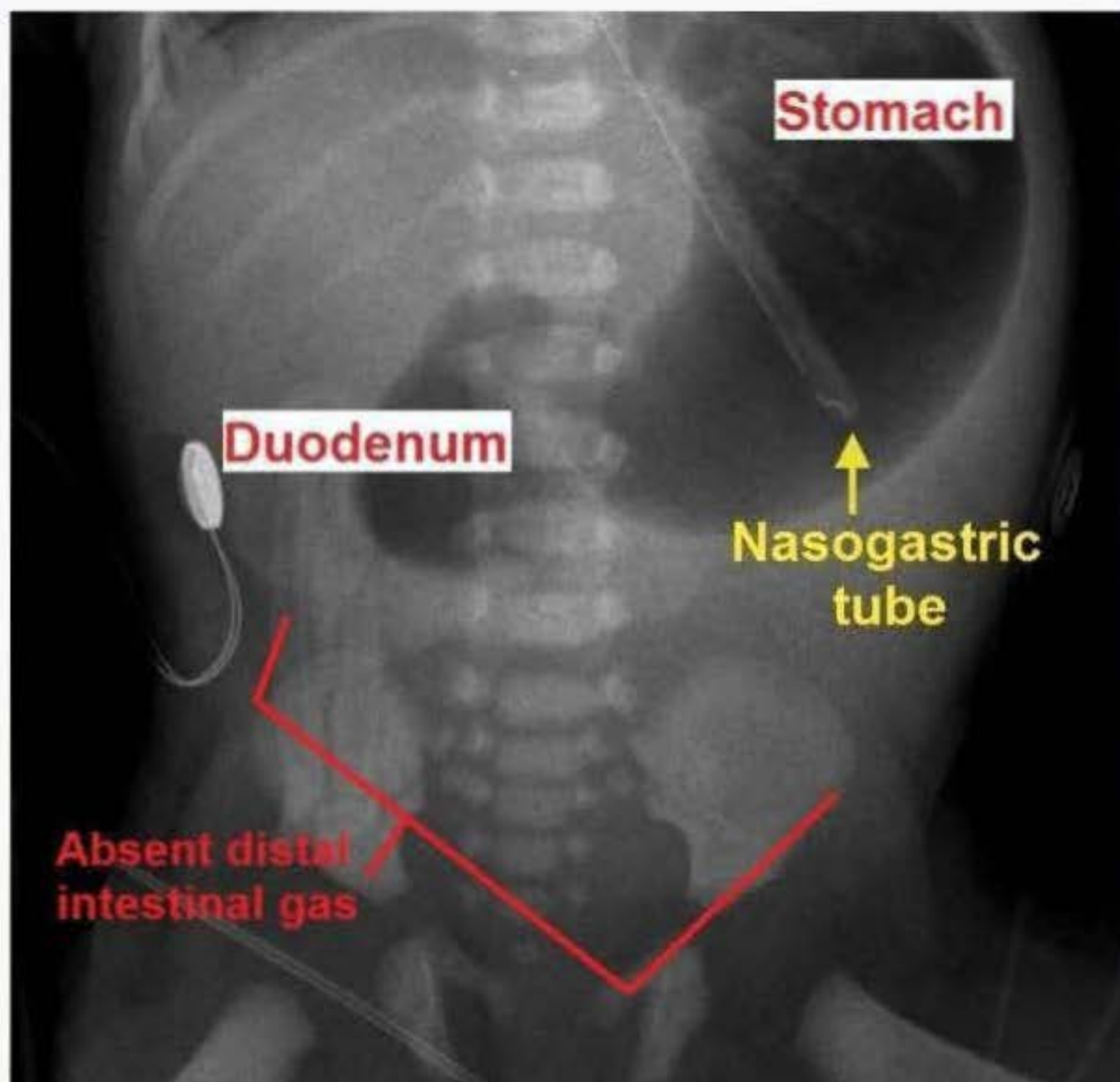
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Explanation:

User Id:



This neonate has a constellation of **dysmorphic features** that are characteristic of **Down syndrome (trisomy 21)**. Trisomy 21 is the most common chromosomal abnormality responsible for intellectual disability and congenital malformations. Up to 5% of patients have gastrointestinal anomalies, with duodenal atresia or stenosis occurring most often. Conversely, up to a third of patients with duodenal obstruction have Down syndrome.

Duodenal atresia classically presents with **bilious vomiting** within the first 2 days of life. The abdomen is not distended due to inability for gas to pass the duodenum.

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Duodenal atresia classically presents with **bilious vomiting within the first 2 days of life**. The abdomen is not distended due to inability for gas to pass the duodenum. Prenatal ultrasound shows **polyhydramnios** due to inability to swallow and remove amniotic fluid. X-ray shows air trapped in the stomach and the first portion of the duodenum ("**double bubble sign**") and no distal intestinal gas. Management includes holding enteral feeds, decompression with a nasogastric or orogastric tube placed to suction, and surgical repair.

A preoperative cardiac assessment is extremely important as 50% of patients with Down syndrome have congenital heart disease, especially **ventricular and/or atrial septal defects**. This patient has the characteristic holosystolic murmur of ventricular septal defect.

(Choice A) Most infants with biliary atresia are asymptomatic at birth and initially seem healthy. Jaundice and acholic (pale) stools typically develop by age 2 months, making this diagnosis unlikely.

(Choices B and D) Other gastrointestinal defects associated with Down syndrome include **congenital aganglionic megacolon (Hirschsprung disease)** and imperforate anus. These patients can have bilious emesis and delayed passage of meconium (age >48 hours). This infant is only 10 hours old and is not expected to have passed meconium yet. However, typical features of distal colonic obstruction include marked abdominal distension and **dilated loops of small and large bowel** on x-ray, findings not present in this case.

(Choice E) Meconium ileus is pathognomonic for cystic fibrosis and typically presents as delayed passage of meconium, marked abdominal distension, and sometimes bilious emesis. It is not associated with Down syndrome. The obstruction is typically at the level of the terminal ileum, and abdominal x-ray shows multiple dilated loops of small bowel.

(Choice F) Pyloric stenosis classically presents as postprandial nonbilious emesis at ages 3-6 weeks, making this diagnosis unlikely.

(Choice G) Sigmoid volvulus presents as abdominal pain, distension, and constipation due to torsion of the sigmoid colon. X-ray shows an inverted U-shaped appearance of the distended sigmoid loop ("**coffee bean sign**"). This typically occurs in elderly patients

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(Choice G) Sigmoid volvulus presents as abdominal pain, distension, and constipation due to torsion of the sigmoid colon. X-ray shows an inverted U-shaped appearance of the distended sigmoid loop ("**coffee bean sign**"). This typically occurs in elderly patients and is exceptionally rare in children.

Educational objective:

Bilious vomiting in the first 2 days of life and a "double bubble" sign on abdominal x-ray are strongly suggestive of duodenal obstruction. Duodenal atresia is strongly associated with Down syndrome.

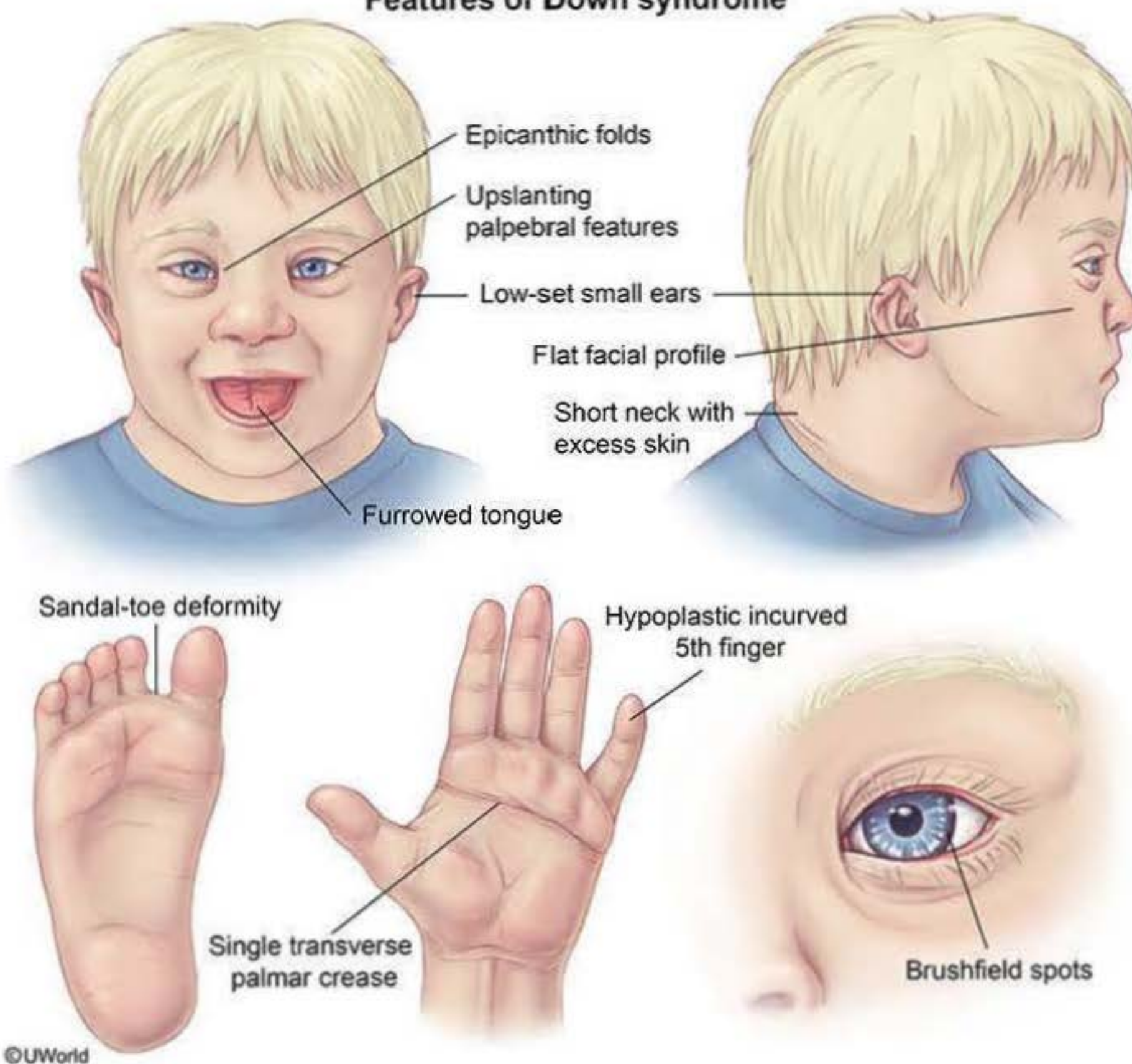
References:

1. **Duodenal atresia: associated anomalies, prenatal diagnosis and outcome.**
2. **Congenital duodenal obstruction in children: a decade's experience.**
3. **Prenatal ultrasonographic detection of gastrointestinal obstruction: results from 18 European congenital anomaly registries.**

Media Exhibit

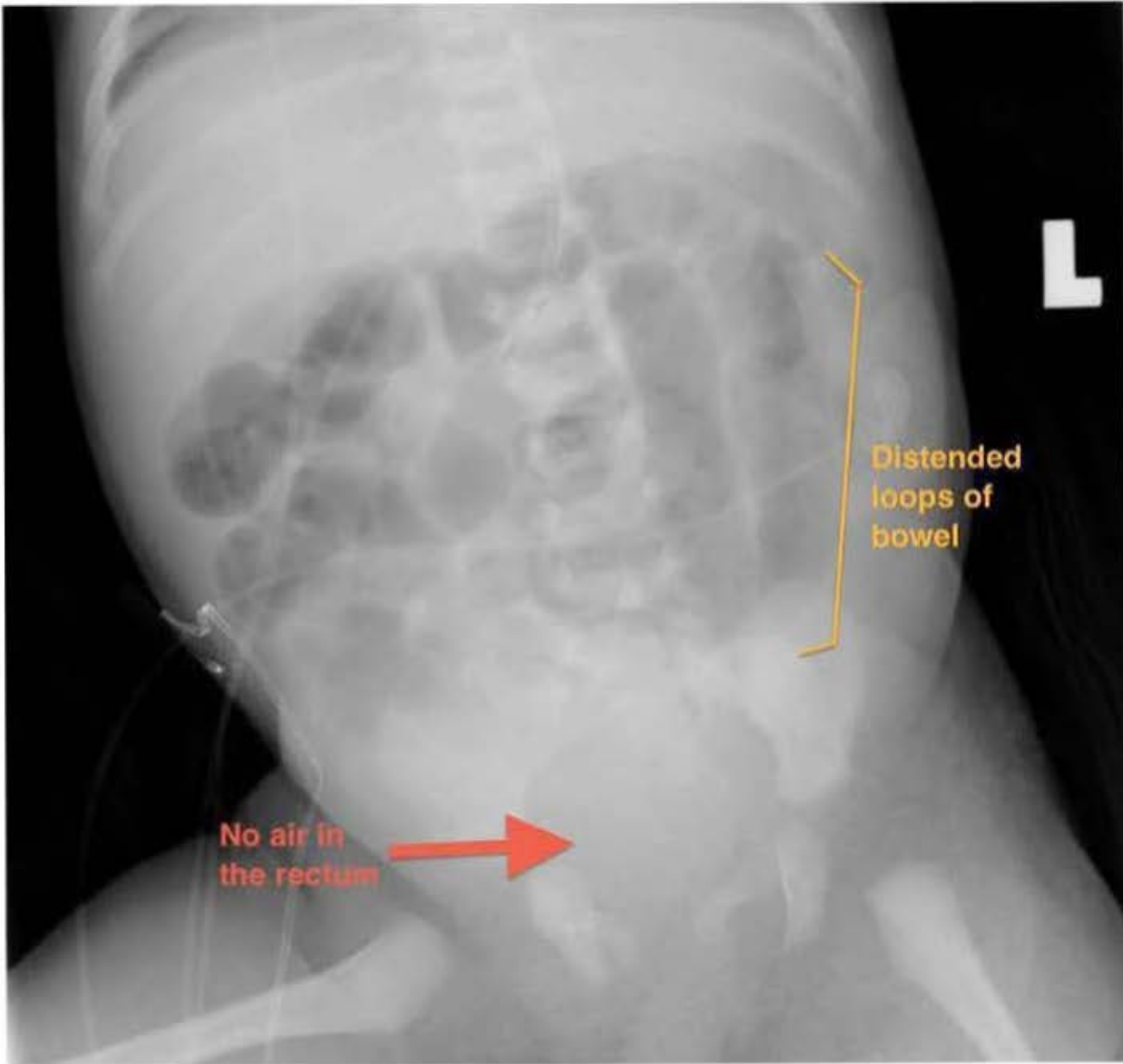
phic features of down syndrome

Features of Down syndrome



Media Exhibit

Small bowel obstruction



Media Exhibit

volvulus

