

A 2-day-old boy in the newborn nursery was born at 39 weeks gestation to a 38-year-old woman by vaginal delivery. The mother received epidural fentanyl for severe painful contractions during labor, but the delivery was uncomplicated. Since birth, the boy has shown little interest in breast or bottle feeding. He spit up the last feed but has not vomited. He has not yet passed meconium. Physical examination shows a newborn with generalized hypotonia, a flat facial profile, low-set folded earlobes, and a single transverse crease on each palm. His abdomen is firm and distended. Digital rectal examination is notable for a tight anal canal and relieves an expulsion of gas and stool. Abdominal x-ray shows markedly distended loops of bowel with no gas in the rectum.



What is the most likely mechanism for his findings?

- ☐ A. Constipation from decreased levels of thyroxine
- ☐ B. Failure of neural crest cell migration during fetal intestinal development

generalized hypotonia, a narrow palpebral fissure, low-set lobed ears, and a single transverse crease on each palm. His abdomen is firm and distended. Digital rectal examination is notable for a tight anal canal and relieves an expulsion of gas and stool. Abdominal x-ray shows markedly distended loops of bowel with no gas in the rectum.



What is the most likely mechanism for his findings?

- ☐ A. Constipation from decreased levels of thyroxine
- ☐ B. Failure of neural crest cell migration during fetal intestinal development
- ☐ C. Intestinal ileus from exposure to maternal epidural analgesia
- ☐ D. Intestinal obstruction from inspissated meconium
- ☐ E. Invagination of a part of the intestine into itself

Submit

A 2-day-old boy in the newborn nursery was born at 39 weeks gestation to a 38-year-old woman by vaginal delivery. The mother received epidural fentanyl for severe painful contractions during labor, but the delivery was uncomplicated. Since birth, the boy has shown little interest in breast or bottle feeding. He spit up the last feed but has not vomited. He has not yet passed meconium. Physical examination shows a newborn with generalized hypotonia, a flat facial profile, low-set folded earlobes, and a single transverse crease on each palm. His abdomen is firm and distended. Digital rectal examination is notable for a tight anal canal and relieves an expulsion of gas and stool. Abdominal x-ray shows markedly distended loops of bowel with no gas in the rectum.



What is the most likely mechanism for his findings?

- ☐ A. Constipation from decreased levels of thyroxine [1%]
- ☒ B. Failure of neural crest cell migration during fetal intestinal development [87%]

What is the most likely mechanism for his findings?

- ☐ A. Constipation from decreased levels of thyroxine [1%]
- ☒ B. Failure of neural crest cell migration during fetal intestinal development [87%]
- ☐ C. Intestinal ileus from exposure to maternal epidural analgesia [4%]
- ☐ D. Intestinal obstruction from inspissated meconium [6%]
- ☐ E. Invagination of a part of the intestine into itself [2%]

Proceed to Next Item

Explanation:

User Id: [REDACTED]

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
"Squirt sign"	Positive	Negative

©UWorld

This infant has characteristic **dysmorphic features** of trisomy 21 (Down syndrome). Although congenital aganglionic megacolon (Hirschsprung disease) is usually an isolated birth defect, it is strongly associated with **Down syndrome**. Hirschsprung disease should be suspected in any neonate who has delayed passage of meconium as 99% of full-term infants stool within 48 hours of birth. The disorder results from failed development of the enteric nervous system of a variable portion of the distal gut and most commonly involves the **rectosigmoid**. The affected colonic segment cannot relax and

This infant has characteristic **dysmorphic features** of trisomy 21 (Down syndrome). Although congenital aganglionic megacolon (Hirschsprung disease) is usually an isolated birth defect, it is strongly associated with **Down syndrome**. Hirschsprung disease should be suspected in any neonate who has delayed passage of meconium as 99% of full-term infants stool within 48 hours of birth. The disorder results from failed development of the enteric nervous system of a variable portion of the distal gut and most commonly involves the **rectosigmoid**. The affected colonic segment cannot relax and therefore is chronically contracted. Newborns present with poor feeding, abdominal distention, failure to pass meconium, and even biliary emesis. Rectal examination can produce an explosive expulsion of gas and stool ("squirt sign") from temporary relief from the obstruction.

Classic abdominal x-ray findings of **complete bowel obstruction** are seen in this patient. If there is no evidence of perforation (eg, free air under the diaphragm), **contrast enema** can potentially delineate the level of obstruction. A **transition zone** may be seen between the narrowed aganglionic segment and the normally innervated, dilated colon (megacolon). The gold standard for diagnosis is **rectal suction biopsy**, which demonstrates the absence of ganglion cells. The treatment of choice is surgical resection of the aganglionic segment followed by anastomosis of the normal bowel to the anus.

The other primary diagnosis of consideration in any neonate with delayed meconium passage is meconium ileus (**Choice D**). Meconium ileus can be the earliest manifestation of cystic fibrosis due to abnormal chloride transport that results in tenacious secretions in multiple organs, including the intestines. The meconium is as thick as glue and difficult to propel, resulting in impaction in the ileum and a narrow, underused colon (**microcolon**). In contrast to Hirschsprung disease, rectal examination would not relieve any meconium due to its inspissated consistency. Meconium ileus is not associated with Down syndrome, making this diagnosis less likely.

(**Choice A**) Down syndrome is also associated with congenital hypothyroidism. However, the majority (>95%) of hypothyroid neonates are asymptomatic in the neonatal period as some maternal thyroxine crosses the placenta. Constipation can develop later. In addition, the rectal examination findings in this child are not typical of congenital hypothyroidism.

(**Choice C**) Some drugs (eg, magnesium sulfate, opiates, ganglionic blocking agents) administered to mother before delivery can result in delayed passage of meconium. However, the rectal examination findings and severity of this patient's presentation suggest an alternate cause.

(**Choice E**) Intussusception is the most common cause of intestinal obstruction in

resection of the aganglionic segment followed by anastomosis of the normal bowel to the anus.

The other primary diagnosis of consideration in any neonate with delayed meconium passage is meconium ileus (**Choice D**). Meconium ileus can be the earliest manifestation of cystic fibrosis due to abnormal chloride transport that results in tenacious secretions in multiple organs, including the intestines. The meconium is as thick as glue and difficult to propel, resulting in impaction in the ileum and a narrow, underused colon (**microcolon**). In contrast to Hirschsprung disease, rectal examination would not relieve any meconium due to its inspissated consistency. Meconium ileus is not associated with Down syndrome, making this diagnosis less likely.

(Choice A) Down syndrome is also associated with congenital hypothyroidism. However, the majority (>95%) of hypothyroid neonates are asymptomatic in the neonatal period as some maternal thyroxine crosses the placenta. Constipation can develop later. In addition, the rectal examination findings in this child are not typical of congenital hypothyroidism.

(Choice C) Some drugs (eg, magnesium sulfate, opiates, ganglionic blocking agents) administered to mother before delivery can result in delayed passage of meconium. However, the rectal examination findings and severity of this patient's presentation suggest an alternate cause.

(Choice E) Intussusception is the most common cause of intestinal obstruction in children age 6-36 months and typically presents with intermittent episodes of severe abdominal pain. It is not associated with delayed passage of meconium at birth, making this diagnosis unlikely.

Educational objective:

Hirschsprung disease should be suspected in any newborn with failure to pass meconium within 48 hours after birth, especially in the setting of Down syndrome. The presentation is consistent with complete bowel obstruction (eg, poor feeding, abdominal distension, absent air in the rectum).

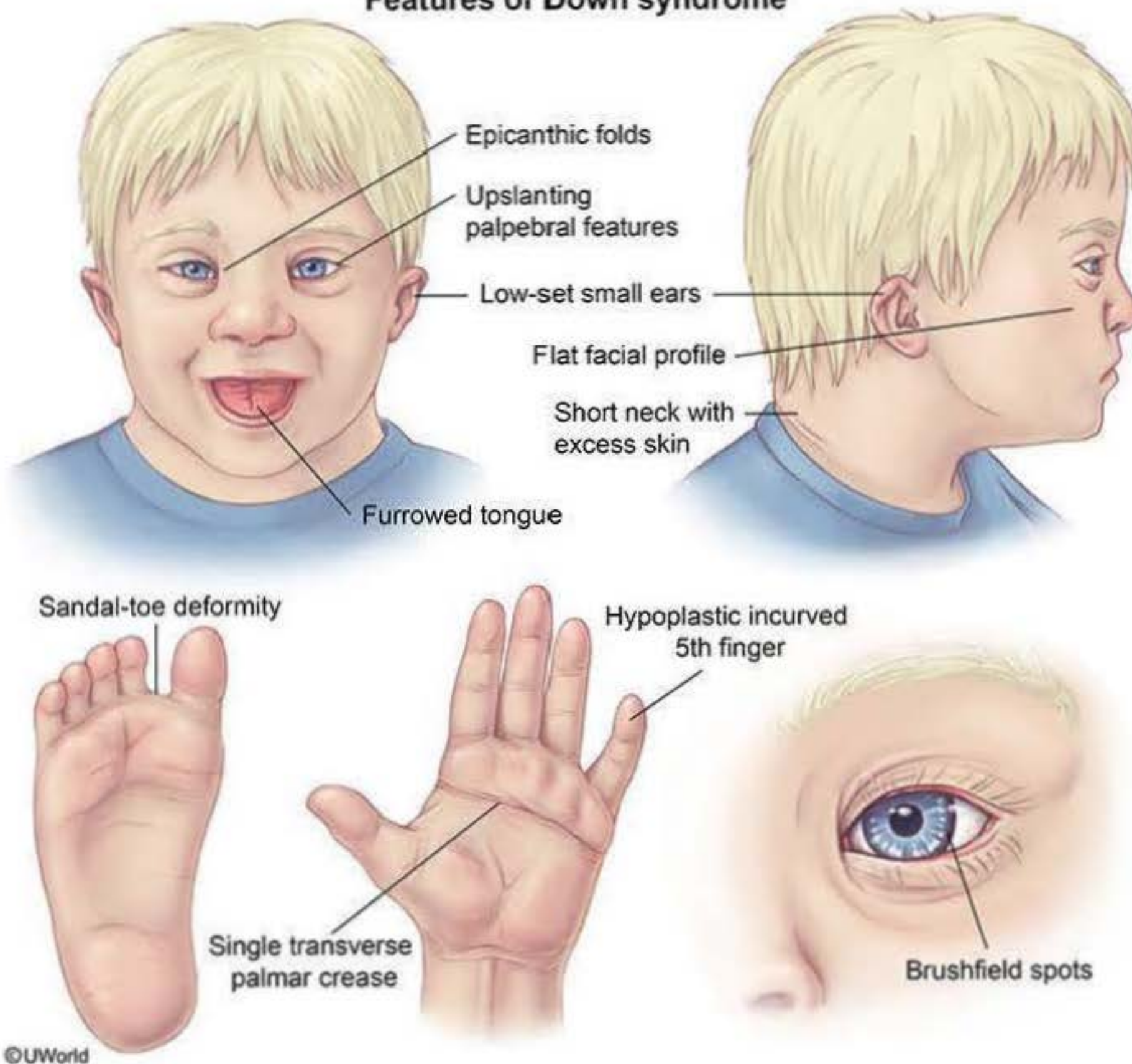
References:

1. [The constipated child: how likely is Hirschsprung's disease?](#)
2. [Hirschsprung disease.](#)
3. [Clinical features of Hirschsprung's disease associated with Down syndrome: a 30-year retrospective nationwide survey in Japan.](#)

Media Exhibit

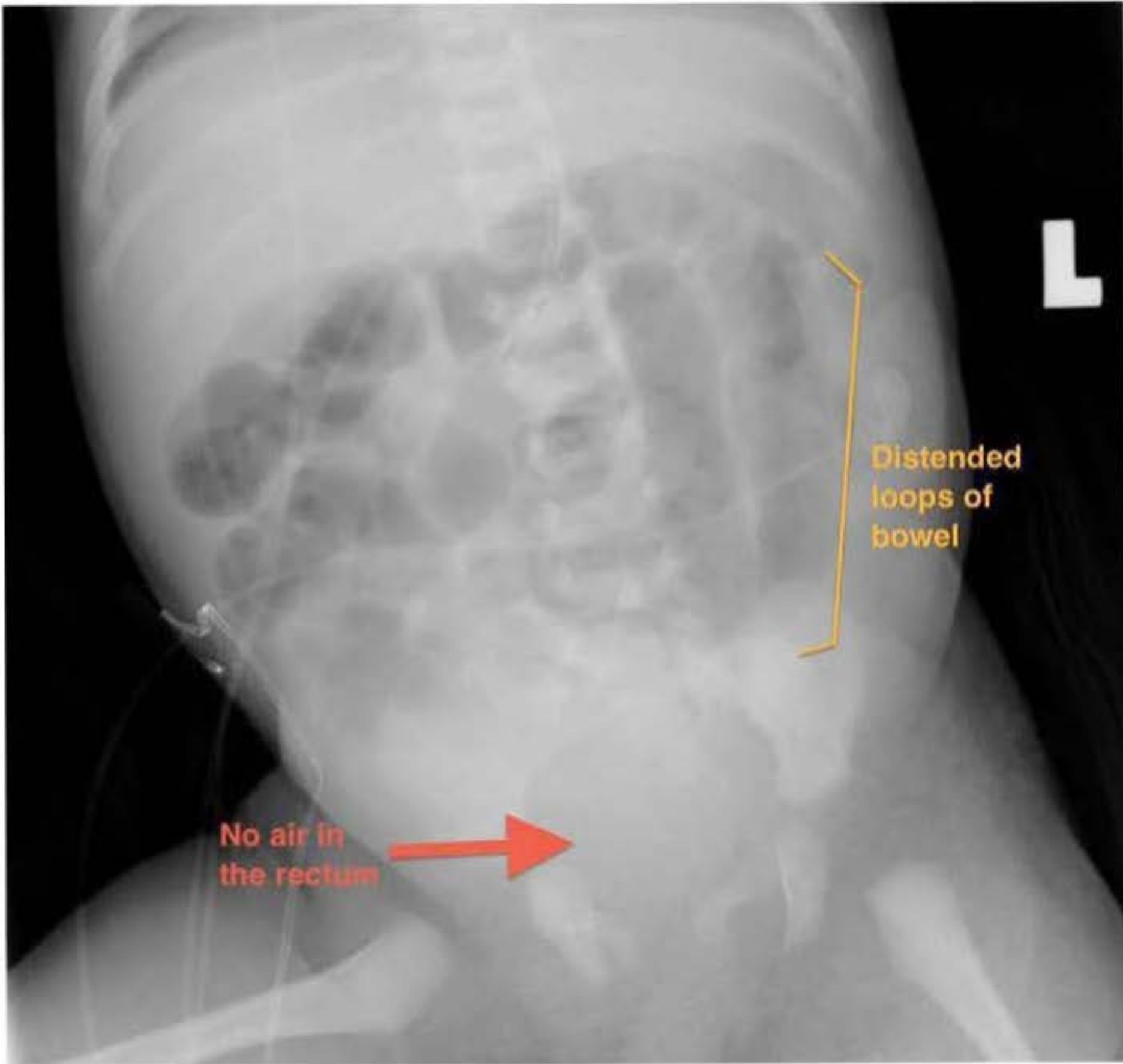
phic features of down syndrome

Features of Down syndrome



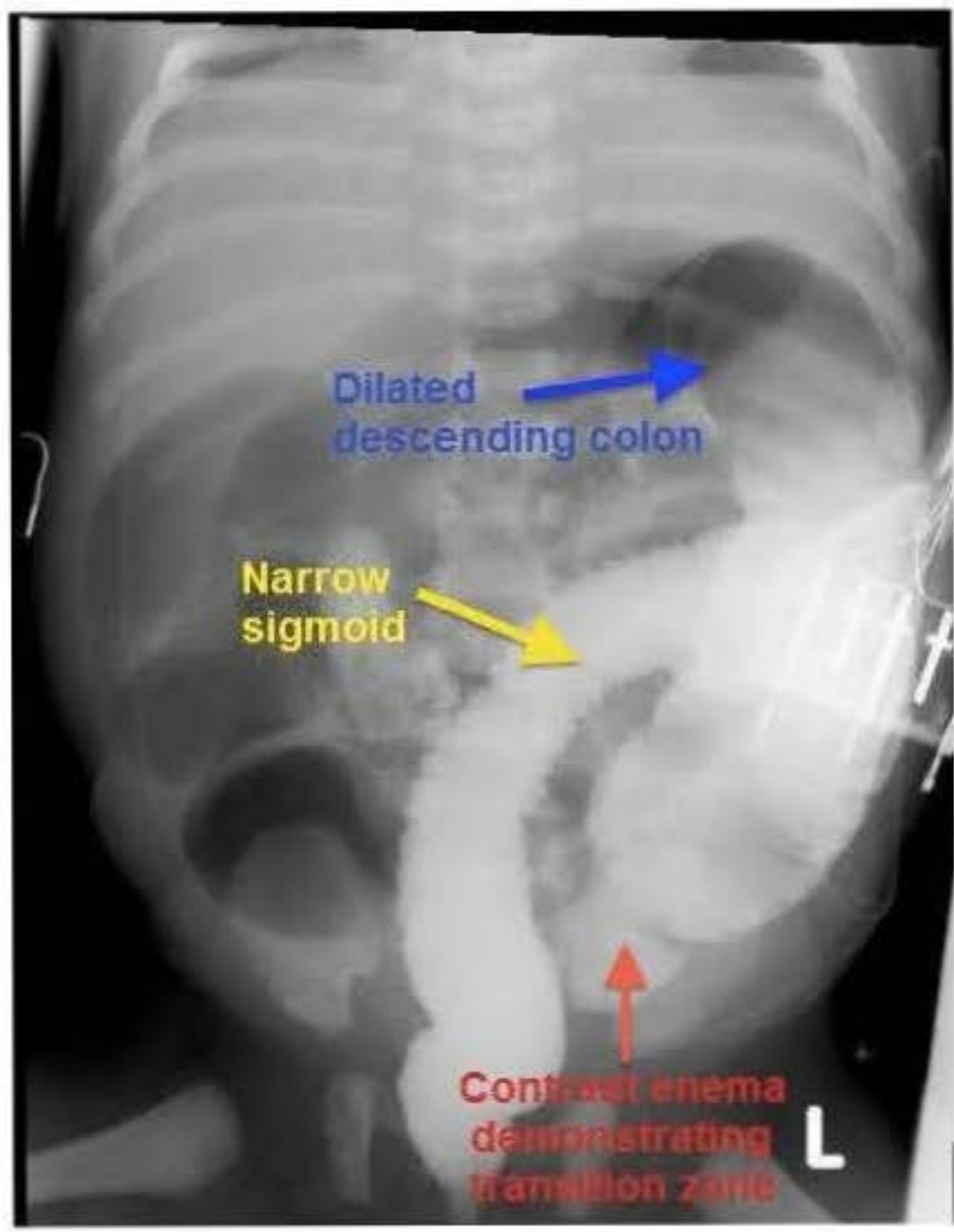
Media Exhibit

Small bowel obstruction



Media Exhibit

Hirschsprung disease



Media Exhibit

m ileus



Contrast enema
demonstrating
microcolon