1. An 8-year-old Caucasian boy is brought to his primary care pediatrician with a 6-month history of recurrent abdominal pain. The episodes occur 3 to 4 times per week and consist of “twisting” pain, in the mid- and lower abdominal quadrants. The episodes are occasionally associated with loose stools; otherwise, there are no additional gastrointestinal (GI) or systemic complaints. According to the mother, the boy’s appetite has diminished and there may have been some weight loss, but this is not documented. The mother also thinks that the child has not grown in 2 years. The past medical history is not contributory; he had been breast-fed for the first 12 months of life. There is no family history of any significant GI disease. The patient has a 13-year-old brother who had been diagnosed with type 1 diabetes mellitus 2 years earlier.

Which of the following tests is recommended?
A. 24-hr. pH-probe monitoring
B. Colonoscopy
C. Serum antitissue transglutaminase
D. Serum antigliadin antibodies
E. Serum H. pylori antibody titer

The recommended response is C.

This boy has celiac disease. This condition affects almost 1% of Caucasians and can present at any age following introduction of gluten. Presentation with recurrent abdominal pain, though not its most common presenting symptom, is well described. Diarrhea, reduced appetite, and weight loss are additional symptoms. In addition, there is a first-degree relative with type 1 diabetes, a condition well known to be associated with a high incidence of celiac disease. The best test to screen for celiac disease is the measurement of serum antitissue transglutaminase: the test is reported to have a sensitivity of almost 100%, with a somewhat lower specificity. Adding serum IgA antibodies to detect the occasional patient with IgA deficiency (about 3% of all celiac disease patients) can be useful. Antigliadin antibodies are much less specific and they should be omitted from the tests used in screening. There are no indications for colonoscopy in this child at this stage, as the procedure is not likely to show significant causes of recurrent abdominal pain in the absence of signs or symptoms of colonic involvement. H. pylori gastritis is poorly correlated with the symptoms described; furthermore, serum H. pylori antibody titer is a relatively poor test for this infection, with a low sensitivity. Finally, the clinical presentation does not suggest gastroesophageal reflux disease (GERD), making the indication for a pH probe not necessary.

2. A 16-year-old child known to have celiac disease since age 4 (diagnosed after a 2-year history of chronic diarrhea and weight loss) is seen as an outpatient 3 years after his last appointment (the patient had not kept the expected annual appointments). His mother believes that he is not adhering strictly to the diet, although he would not admit it. The patient displays a defiant attitude and denies any dietetic transgression. He does say that when on just 1 or 2 occasions in the past years he
may have eaten some gluten-containing food, this hasn’t bothered him at all, to the point he is not sure he has the disorder. Mom insists that he appears more tired, something he vehemently denies. Physical exam shows a normal teenage boy, not malnourished.

What is the best course of action?

A. Surmise that the patient may indeed have been ingesting substantial amounts of gluten and that he must have grown out of his gluten intolerance, as he has evidently not shown any of the symptoms he presented when first diagnosed, and his physical examination is within normal limits

B. Tell the patient that there is no such thing as transient celiac disease and he must stay on a diet as originally recommended

C. Plan a repeat esophagogastroduodenoscopy (EGD) with duodenal biopsy to verify if he is relapsed

D. Obtain serologic evidence of recurrence by checking anti-Endomysium antibodies (EMA) and, if results are positive, try to reinforce the need to stay on a diet

E. Obtain blood work for CBC, serum iron, and antigliadin antibodies

The recommended response is E.

Teenagers are well known to often resist compliance to pharmacologic and/or dietetic treatments. This boy had celiac disease and should have been seen at regular yearly intervals to monitor compliance and any medical/nutritional issues. Celiac disease is permanent, so there is no chance that he has outgrown it, although even long-term “apparent health” has been described in celiac patients who were not on a gluten-free diet; however, to simply state that he needs to stay on a diet without further action is unlikely to result in any change in compliance. A repeat EGD might show signs of relapse; however, this is not necessarily the case, as relapse of duodenal morphological changes can occur as late as 2 years in a minority of celiac patients eating gluten. Thus, to propose the biopsy directly would not be a good choice. Although EMA are highly specific, they have less sensitivity and are not the most reliable tests to judge minor dietary indiscretions. On the contrary, anti-gliadin antibodies are much less specific for celiac disease, but—being anti-food proteins antibodies unlike the EMA autoimmune antibodies—they are promptly responsive to the introduction of gluten. Thus, a well-followed gluten-free diet should result in almost undetectable serum values of anti-gliadin antibodies, both IgA and IgG. Furthermore, if the patient is indeed reacting to gluten ingestion and is easily tired as mother reports, checking him for possible iron-deficiency anemia is indicated, and may well serve to enforce the dietetic compliance.

3. A 5-year-old boy presents with a history of constipation. His mother reports that in the past 2 years, the child—who had been properly toilet-trained around age 2—had started to have less frequent defecations. The stools have been typically large, hard, and passed approximately every 3 to 4 days and with “a lot of straining.” In a couple of instances, streaks of fresh blood have been seen around stools. She informs you that several remedies such as fruit juices, prunes, and mineral oil have failed. The only notable finding in his past medical history is the need for many formula changes during his first year of life, as mom says he was “lactose intolerant.” This appears to have regressed after age 13 months and he now drinks
milk without problems. The physical examination is unremarkable, except for some stools palpated in the rectal vault, slightly heme-positive.

In addition to functional constipation, what would be a likely diagnosis for this patient’s symptoms?

A. Hirschsprung’s disease (HD)
B. Lactose intolerance
C. Cow’s milk protein allergy
D. Hypothyroidism
E. Ectopic, anteriorly displaced anus

The recommended response is C.

The history and physical both rule out HD; there is no malnutrition, history starts late (is neonatal in more than 90% of HD), stools are large and round (they are string-like in HD), and are present in the rectal vault. Likewise, an anteriorly-displaced anus is easily ruled out by the physical examination; this often-overlooked disorder typically causes constipation from an early age, as the angle between the rectum and the anal canal becomes too narrow for stools to be expelled normally. Although hypothyroidism can lead to constipation, for this problem to cause it other signs would have to be present; at this age in particular, short stature and slowed development. Lactose intolerance is rarely present at this young age and would lead to the opposite clinical manifestation of diarrhea. Cow’s milk protein allergy (CMPA), on the other hand, is a relatively recently recognized cause of constipation. Allergic symptoms may appear during infancy with GI manifestations such as vomiting and diarrhea (a condition that commonly leads to multiple formula changes and is often erroneously referred to as “lactose intolerance”), which can subside. However, a condition of proctitis may occur and continue in the following years with minimal overt symptomatology, essentially causing painful defecations and, hence, a voluntary retention of stools that is the ultimate reason for constipation. The response to withdrawal of milk and derivatives from the diet is typically dramatic.

4. A 6 week-old baby is presented for complaints of hematochezia. The baby had been delivered at full term without complications and the mother was able to breast-feed him from the beginning. His neonatal period was unremarkable and he has been gaining weight and developing normally. One week prior to the visit, mom noticed flecks of bright red blood in the diaper, mixed with somewhat looser stools. This presentation recurred twice in the following days and has caused great maternal anxiety. The mother explains that there are no other problems and that there is no family history of GI disorders. The mother is allergic to shellfish. The physical examination shows a happy, well-nourished, well-hydrated baby, without any positive physical findings.

What is the most common explanation for this baby’s symptoms?

A. Infectious colitis
B. Ulcerative colitis
C. Cow’s milk allergic colitis
Infectious colitides resulting in bloody stools are secondary to invasive bacterial pathogens. Such conditions are well known to occur throughout the pediatric age, including infancy. However, they are overall uncommon at this early age and their prevalence is even lower in developed countries. In addition, this baby has no other symptoms suggestive of an infectious cause (fever, inconsolable crying attributable to crampy abdominal pain, dysenteric diarrhea). Inflammatory bowel diseases, both ulcerative colitis (UC) and Crohn’s disease (CD), have been described below 1 year of age. However, they are exceedingly rare in this age group and practically unknown in a 6-week-old baby. In addition, they are cause of bright red blood passed without association with loose stools; in essence, this is a possible but unlikely diagnosis. Breast-fed babies are well known to suffer from CMA. This may result from reaction to milk proteins that are absorbed intact from mother’s GI tract and reach the baby’s gut lumen via breast milk. The resulting reaction leads to a distal colitis, endoscopically manifested by small, round pale yellow areas surrounded by a red halo. The typical symptomatology is the one described: the passage, by an otherwise perfectly healthy and growing infant, of streaks of fresh red blood from the rectum. This condition’s prevalence is not exactly known, but is thought to affect 1 out of 10 of all milk-allergic babies (25% to 30% of all infants), thus making it by far the most common cause of hematochezia in this age group.

5. The best course of action for the baby in the above vignette is:
A. Schedule a colonoscopy
B. Order a barium enema
C. Recommend discontinuation of breast feeding
D. Recommend dietetic changes to mother to eliminate all milk protein and reassess in 2 weeks
E. Start antibiotic coverage

The recommended response is D.

There is no need to perform procedures such as colonoscopy or barium enema in this case. The most likely diagnosis is obvious and direct therapeutic intervention is the best option. This would initially consist of recommending discontinuation of the intake of any milk protein-containing foods from the mother’s diet, while allowing breast-feeding to continue. This intervention is efficacious in about two-thirds of the cases and disappearance of hematochezia can be expected to occur within 7 to 10 days. Therefore, reassessing the child after this time is recommended. At that time the symptom persists (one-third of cases), then discontinuation of breast-feeding, to be replaced by an
extensively hydrolyzed formula, will be required. Clearly there is no rationale for an antibiotic coverage.

6. An Afro-American 13-year-old child is seen because of recurrent abdominal pain (RAP). He started to complain of vague mid-abdominal pain almost 2 years before; initially the episodes were mild, occurred about once every other week, and lasted a few minutes. Subsequently, however, they have appeared more frequently and their intensity may have increased. Currently, he has sharp pain at least twice a week, vaguely localized in the mid-abdominal area, rated 6 to 7 on a pain scale of 1 to 10, presenting in no clear association with any event, but sometimes following a meal. No nausea, vomiting, and/or constipation are reported. However, he does occasionally present loose stools and in 2 occasions they may have been blood-tinged. He reports no major change in appetite, but for the fear that eating may trigger the pain, his food intake has decreased, and his mother thinks he may have lost 2 to 3 kg in the past few months. No contributory elements are found in the past medical history. In review of systems, there has been in the past couple of years some concern about his growth in stature, and parents are planning a visit to an endocrinologist. No family history of GI disorder is reported. Both parents are tall. On physical examination, the boy appears thin; his weight is just below the 3rd percentile and his height is in the 10th percentile. The abdomen is full, with some tenderness at palpation of the mid and lower quadrants; there are no masses. His Tanner stage is II.

Which initial investigations are likely to provide the key for the diagnosis?
A. None, as this boy evidently is affected by functional RAP
B. An EGD
C. A plain X-ray film of the abdomen
D. Lab work: CBC with differential, ESR, CRP, ASCA, and p-ANCA titers, iron panel, albumin
E. Lab work: amylase, lipase, liver function tests (LFTs)

The recommended response is D.

Assessing a child for RAP is a daunting task. Functional causes are known to be very common, occurring in approximately 15% of school-age children and teenagers, and they may have protean manifestations. There are, however, elements in this patient’s history that seem to indicate an organic origin: his stunted growth, his weight loss, the presence of occasional bloody, loose stools, and the objective finding of abdominal tenderness, typically absent in functional RAP. Thus, performing no investigations would be a poor choice. An EGD is definitely an option, however, the patient does not present symptoms or signs clearly pointing to an upper GI involvement and, thus, the procedure is unlikely to provide support for a diagnosis at this initial stage. Even less informative would be a plain film of the abdomen, unlikely to disclose any abnormalities responsible for RAP. Pancreatitis would have presented with a more acute course, would not have caused any blood in the stools, and the location of the pain would have been different. Liver diseases do not cause RAP. Thus, to obtain the tests in option E is clearly not a valid option. Rather, this young teenager with somewhat inadequate skeletal growth and weight loss, RAP, and occasional bloody and loose stools may well be affected by
Crohn’s disease. It is in fact known that changes in linear growth and in weight gain can precede, often by years, any obvious GI signs/symptoms, to the point that it is not infrequent that children with CD are first evaluated by endocrinologists.

7. An 8-month-old girl is brought to the emergency department because of a 2-day history of vomiting (non-bloody, non-bilious), followed by diarrhea consisting of 6 to 7 bowel movements of watery, non-bloody stools. Her oral intake of a normal infant formula and stage I baby foods is reduced and she is irritable.

The most likely diagnosis is:
A. Cow’s milk protein allergy
B. Rotavirus enteritis
C. Lactose intolerance
D. Bacterial enterocolitis
E. Urinary tract infection (UTI)

The recommended response is B.

Cow’s milk protein allergy most commonly presents in the first 2 months of life, after formula is introduced, although (less commonly) it may also appear later; it is estimated to affect 2% to 3% of all artificially fed babies. Rotavirus is by far the most common agent of acute-onset vomiting and diarrhea in the age group 6 to 24 months. Diarrhea of bacterial origin is less commonly associated with emesis and induces a dysenteric picture, with mucus and blood in the stools. Lactose intolerance, though often transiently associated with Rotavirus diarrhea where it may be a secondary cause of diarrhea, is not a cause of vomiting, and it is never a primary cause of diarrhea in infancy. Finally, although UTI may be associated with vomiting and diarrhea, they are uncommon causes of this symptomatology. This infant has Rotavirus enteritis (B).

8. The emergency room doctor assesses the infant above. He finds her to be alert, afebrile, and mildly dehydrated. What is the best course of action?
A. Admit the patient, place her on nothing by mouth (NPO) status on IV maintenance fluids
B. Prescribe a lactose-free formula and send the patient home
C. Obtain a stool culture and perform IV rehydration in the emergency department
D. Start oral rehydration therapy (ORT) with a low-osmolarity solution and administer so to rehydrate within 3 to 4 hours; then send home on normal, age-appropriate diet (including normal, lactose-containing formula), recommending to continue ORT ad lib until diarrhea stops
E. Call the pediatric gastroenterologist on call for further management

The recommended response is D.

It is now well established that a typical, uncomplicated infant with acute diarrhea such as the one described should be treated with ORT, without any need for IV fluids or stool cultures. Rehydration should be performed quickly, within 3 to 4 hours, and ORT should
be continued throughout the duration of the diarrhea. In addition, it is well established that refeeding with a lactose-free formula is not necessary and, after rehydration, patients should be offered a completely normal diet. This situation should be managed by a primary care pediatrician without need for referral.

9. A 3-month-old Afro-American boy is admitted after his primary care pediatrician has found him to have failure to thrive (FTT) and unexplained chronic diarrhea. He was born at term with a weight of 3.2 Kg and was fed exclusively a normal infant formula since birth. At 3 weeks of age he presented with increased frequency of stools, which then became bulky. No other symptoms are reported and his oral intake is adequate. At 3 months, his weight is 4.1 kg and he continues to have 5 to 6 bowel movements per day of loose, occasionally mucousy, and often-abundant stools. From some tests done by his primary care doctor you learn that fecal fat test was positive for “split fat,” no increase in “neutral fat”; Hb was 9.3 g/dl and serum albumin was 2.9 g/dl. In family history, the patient has a 2-year-old brother and a 4-year-old sister, who are both healthy now. The brother, however, had “feeding intolerances” as a baby, and many formulas had to be changed, until he did well on a casein hydrolysate formula.

On physical examination, the patient appears malnourished but not dehydrated; eyelids are mildly edematous; abdomen is distended and full.

What is the most likely explanation of these findings?
A. Celiac disease
B. Poor feeding management
C. Giardiasis
D. Cystic fibrosis
E. Cow’s milk protein allergy

The recommended response is E.

Although celiac disease can have a very early onset, this may obviously only follow ingestion of gluten, so it is not a diagnostic possibility here. Poor feeding techniques are causes of FTT in early infancy, but we have no indications for such a problem here, and two previous siblings did not have growth problems; furthermore, the presence of diarrhea would not fit into this category. Giardiasis may have caused this child’s symptoms, as this parasitic infection may result in malabsorption, but at this early age this is a highly unlikely explanation, especially in developed countries. Cystic fibrosis is definitely in the differential diagnosis, even in the absence of respiratory symptoms; FTT with chronic diarrhea and protein malnutrition (low serum albumin) would support it. However, this condition is rare in Afro-Americans (about 1:15,000) and the fecal fat test would have shown increase in neutral, not split fat, as this disease is a maldigestion not a malabsorption condition. Cow’s milk protein allergy-induced enteropathy is the most likely explanation for the findings described. This entity is in addition a cause of protein-losing enteropathy, a likely underlying problem here.

10. After admission, what key investigation will provide support for the diagnosis?
A. RAST tests for milk proteins
B. Skin prick tests with milk proteins
C. Duodenal biopsy
D. Fecal leucocytes
E. WBC with differential

The recommended response is C.

Food allergic enteropathies are by definition not induced by an IgE-mediated mechanism, so the tests in A and B, that only detect, respectively, circulating and tissue-related IgE, will be worthless. The presence of fecal leucocytes might support a condition of milk-allergic colitis that can be associated, but is not necessarily coexistent with the enteropathy. Thus, the most direct evidence for the presence of milk-allergic enteropathy is provided by the morphological changes seen in the duodenal mucosa, typically patchy areas of villous atrophy with mucosal edema.

11. A 7-year-old child presents with a history of vomiting and RAP. His mother says he presented with vomiting the first time about a year ago; the episodes occurred in the afternoon, were repeated 4 to 5 times, and then subsided over night. Subsequently, he presented again with vomiting about 2 months later, this time the emesis, always non-bloody, non-bilious, was more intense. He had many episodes over the course of 3 days, accompanied by headaches, bouts of sharp abdominal pain, and complete lack of appetite. He was brought to an emergency room on the third day; some laboratory tests were obtained (the mother cannot recall which tests), he was rehydrated and sent home, where he recovered completely in a short time. Over the following months, the episodes have recurred 4 more times with similar characteristics. Of interest, the patient is reportedly totally well between these severe episodes. His past medical history is noncontributory—the patient has “always been healthy.” The family history is negative for GI disorders; his mother has migraine. In social history, it is noted that the parents have gone through a “tough” divorce about 3 years earlier. Mom is worried about a possible tumor but also states this may be “stress-related.”

The physical examination shows a well-developed, well-nourished, healthy-looking boy in no distress. No positive findings are seen.

What is the most likely diagnosis?
A. Functional recurrent abdominal pain
B. H. pylori gastritis
C. Intracranial tumor
D. Lactose intolerance
E. Cyclic vomiting syndrome

The recommended response is E.

Functional RAP is not typically associated with recurrent emesis: indeed, one of the key elements to suspect the functional origin of RAP is the absence of other concomitant signs/symptoms. While H. pylori chronic gastritis can be associated with recurrent vomiting, the symptoms are not usually so severe, nor do they recur with this periodicity; rather, they tend to be present more constantly. While the presence of an intracranial
lesion cannot be ruled out as possible cause of these symptoms associated with headaches; however, this would typically result in emesis at awakening, not be associated with RAP, and would not leave the patient totally symptom-free in between attacks. Lactose intolerance is defined as the clinical manifestation of inadequate lactose digestion by the small intestinal lactase, and the most common symptom is diarrhea, often associated to abdominal bloating; emesis is not caused by lactose intolerance. This patient has cyclic vomiting syndrome, a disorder thought to affect approximately 1:500 children. Of yet unclear origin, the disorder causes recurrent bouts of intense emesis, with typically 5 to 10 episodes per day over a period of 1 to 7 days, accompanied by headache, more or less severe abdominal pain, anorexia and, less commonly, also lethargy and photophobia. Characteristically, the episodes end abruptly with a sudden restoration of normal health, and these children are completely well between the crises.

12. A 12-month-old girl is referred by her primary care pediatrician for poor feeding, FTT, and irritability. The parents report that her appetite has been poor, particularly in the past 2 to 3 months, during which her weight has practically remained flat. In addition, she has a history of being fussy and irritable, especially during feedings, and of some infrequent regurgitations and rare emesis. There has been no diarrhea and no constipation. On 3 different occasions, CBC was normal, followed by normal serum iron, basic metabolic panel (BMP) and urinalysis were normal, so the patient is referred to you.

The patient was born at term with a weight of 2.95 kg. Her past medical history includes that she was breast-fed for about 1 month and then was switched to a normal infant formula. Regurgitations started at about 4 months and irritability began shortly thereafter. Occasionally, she would have projectile emesis, once apparently blood streaked. A change in formula first to Alimentum and then to Pregestimil (utilized in total for almost 1 month) did not result in any clinical improvement. Zantac in the amount of 6 mg/kg/day was given daily since age 4 months, but since no effect was seen, it was discontinued after 3 months.

Her pediatrician did not notice any appreciable change in the rate of weight gain until approximately the age of 6 months, when she began to fall off percentiles: the introduction of solid foods at that time did not help, and she actually started to refuse feedings more often.

In family history, her father reports allergies to peanuts and shrimp; a maternal uncle has Crohn’s disease, diagnosed at age 14.

On physical examination, the child is borderline malnourished, crying, and irritable. There are no other positive physical findings. Which of the following is the most likely GI condition affecting this girl?

A. Celiac disease
B. GERD
C. Cow’s milk protein allergy
D. Shwachman-Diamond syndrome
E. Eosinophilic esophagitis

The recommended response is E.
Poor weight gain can have a long list of causes, only some of which are linked to GI problems. In this case, the important lack of appetite with the resultant diminished oral intake may well be responsible for the observed FTT; the presence of emesis, even though only occasional, certainly adds to the possibility that the GI tract is involved. Celiac disease, though definitely a possibility, is not a likely diagnosis: there is no diarrhea, regurgitations and irritability started before introduction of gluten, and her serum iron (commonly low in this condition) is normal. Gastroesophageal reflux disease and a resultant peptic esophagitis, also is a possibility, but not the most likely: the symptoms began after the typical age of 2 months, did not respond to an adequate antacid regimen, and the introduction of solids made the symptoms worse, which is inconsistent with this diagnosis. Cow’s milk protein allergy may well be responsible for all of the signs and symptoms, and is further supported by the positive family history. However, the lack of response to a diet with extensive protein hydrolysates such as Alimentum and Pregestimil, used for an adequately long period, makes this diagnosis less likely. Shwachman-Diamond syndrome is characterized by cyclic neutropenia, metaphyseal dysostosis, and is the second most common cause of pancreatic insufficiency in childhood. Failure to thrive is the result of insufficient digestion and this results in chronic diarrhea; its absence, the presence of anorexia and vomiting, as well as the normality of the CBC that would have shown neutropenia, all make the diagnosis highly unlikely.

Eosinophilic esophagitis is the most likely GI diagnosis. This condition is characterized by an intense eosinophilic infiltrate in the esophagus at various levels but more commonly in its distal one-third, and is now increasingly recognized as a cause of clinically important esophagitis. It is often—but by no means always—associated with a condition of food allergy, commonly reported in family history as well. It is not associated with a peptic injury, does not respond to antacid therapy, and is not necessarily accompanied by peripheral eosinophilia. The esophagitis can be severe and often causes severe refusal of food intake (particularly solids) and blood-streaked emesis. It is evident at endoscopy, where it is seen as furrowing, whitish nodularities; it is diagnosed by pathology that shows more than 20 eosinophils/field.

13. You are asked to consult on a 6-day-old newborn girl who presents with diarrhea since birth. She has 8 to 10 bowel movements per day of watery stools that began on her first day of life, immediately after meconium was passed. No associated GI symptoms (no vomiting) except for a distended abdomen. However, the baby presented with tremors on the second day of life and a dextrostix showed blood glucose of 35 that was corrected. Since then, she has been on IV fluids at maintenance to keep her well hydrated.

The baby was breast-fed from birth and still is being breast-fed; the oral intake appears adequate and she sucks well. She was born at term, via a normal spontaneous vaginal delivery, from a healthy young mother, with the weight of 3.2 kg. There is no prenatal history of any significance; the mother was followed regularly during pregnancy with repeatedly normal ultrasounds. Family history is noncontributory and there are no instances of GI disorders. She is the first child of this couple. During the past 2 to 3 days, the following investigations were done:

- CBC with differential: normal
• Blood cultures, CSF cultures: negative
• BMPs: Na 135–140, Cl 108–112; K 3.7–4.5; CO₂ 19–21; pH 7.35–7.40; glucose 45–90
• X-ray of abdomen: increased amount of fluid distending the intestinal loops
• Stool cultures: repeatedly negative
• Stool virus search for Rotavirus, Adenovirus: negative
• UA: 1+ for reducing substances

You put the baby on NPO status and the diarrhea subsides promptly and completely. When feeding is resumed with a protein hydrolysate formula, the diarrhea recurs with the same characteristics. A stool output around 250 to 300 ml/day is recorded. The baby is put on total parenteral nutrition (TPN). After 2 more days, you switch feedings to the elemental formula Neocate, but the baby continues to have unchanged diarrhea over the ensuing 7 days. The most likely diagnosis is:

A. Cow’s milk protein allergy
B. Congenital Chloridorrhea
C. Congenital Na-losing diarrhea
D. Glucose-galactose malabsorption
E. Congenital microvillous atrophy

The recommended response is D.

It is highly unlikely that this baby has milk protein allergy; she presented immediately with diarrhea, without the typical free interval, was being breast-fed when diarrhea started, and did not respond to any of the formula changes that would have resolved the symptom. Options B and C refer to congenital disorders of electrolyte absorption that can result in neonatal diarrhea. In Chloridorrhea the Cl/HCO₃ antiport is defective, and this results in secretory diarrhea with low serum Cl and metabolic alkalosis. Thus, this disorder is ruled out by the normal levels of serum Cl and the lack of metabolic alkalosis. Congenital Na-losing diarrhea is a more rare disorder, in which the antiport Na/H is defective. This leads to a secretory diarrhea with large Na stool losses, hyponatremia, and severe metabolic acidosis, all elements not found in this patient. Both Chloridorrhea and Na-losing diarrhea, in addition, have a prenatal onset that causes polyhydramnios, something lacking in our patient.

Congenital microvillous atrophy is the leading cause of secretory diarrhea in the first weeks of life. It is due to a permanent damage of the enterocytes, the microvilli of which are not expressed at the apical border. The typical clinical presentation is a watery, profuse diarrhea starting in the first hours of life; the stools are watery and the stool output is very high (100 to 500 mL/kg/d), a volume comparable to or higher than that seen in cholera. The diarrhea is of a secretory type, thus, it persists at these high rates despite fasting. This is different from what seen in our patient: her stool output was never so large and the diarrhea clearly improved after NPO was instituted. This baby is affected by glucose-galactose malabsorption. This autosomal recessive disorder of the Na-coupled intestinal transporter for glucose and galactose implies the inability to absorb these
monosaccharides from birth, and results in osmotic type diarrhea when either one of these sugars (or their precursors, such as lactose) is ingested. Hypoglycemia is often present, as in our patient, when IV fluids with glucose are not given; a mild-to-moderate metabolic acidosis is also often seen. In most cases, the defect is thought to also involve the kidney and mild glycosuria can be found. The diarrhea typically disappears when the offending carbohydrates are removed. Thus, the patient exhibits all the classic signs of such a condition.

14. A 3-month-old infant, exclusively fed with formula, is brought to the attention of a doctor because of a 1-month history of regurgitations. These occur effortlessly after most meals and are of about 10 to 20 cc each; no blood has ever been noticed and there are no other associated symptoms. Weight gain is unaffected.

The most appropriate course of action is:
A. Reassure the parents and recommend only generic “reflux precautions”
B. Recommend a formula change, as the baby obviously cannot tolerate the current one
C. Recommend weaning to solid foods to prevent regurgitations
D. Start the baby on a proton pump inhibitor to prevent esophageal damage
E. Obtain an upper GI x-ray study

The recommended response is A.

This baby most likely presents with GER in its most common and benign presentation (“happy spitter”): functional GER, as opposed to pathogenic GER, or GERD. Functional GER occurs in almost all babies, while only in a small fraction (2% to 5%) is this entity severe enough to lead to complications such as excessive vomiting with resulting reduced caloric intake and FTT; esophagitis; occult GI blood loss; respiratory symptoms and even ALTE (acute, life-threatening episodes). When obviously none of such complications is present, simple reassurance is adequate, in consideration of the excellent prognosis: more than 70% of infants with GER will cease to have this problem by their first birthday. Cow’s milk protein allergy may present with similar complaints, but they are typically more severe, and tend to be associated with diarrhea and/or with insufficient weight gain. Although weaning to solid foods will reduce regurgitations in GER, this process is best started only when the baby is at least 4 to 5 months old. No investigations or drug treatments are required for such a presentation.

15. A 6-week-old baby boy is seen because of persistent jaundice, dark urine and acholic stools. Jaundice had been present since shortly after birth, but was not adequately assessed. He had a normal birth weight. No important elements are disclosed by family history. On physical examination, the baby is jaundiced, has both liver and spleen palpable 4 cm below the costal margins. The following laboratory studies are obtained:
- CBC: WBC 13.8 K/mmc; Hb 9.8 g/dl
- PT 13.8 sec; INR 1.2
- AST 280; ALT 350 U/L; GGT 400, Alkaline Phosphatase 550
• Bilirubin: total 9.3/conjugated 6.1 mg/dl
You perform a liver biopsy that shows: canalicular and cellular bile stasis and bile duct proliferation and portal fibrosis.” What diagnosis is consistent with this clinical picture?

A. Alagille’s syndrome
B. Biliary atresia
C. Idiopathic neonatal hepatitis
D. Neonatal hemochromatosis
E. Wilson’s disease

The recommended response is B.

Alagille’s syndrome occurs in approximately 1/100,000 live births, There is often a family history for related clinical features in at least 20% of cases. Classic pathology findings include bile duct paucity. Idiopathic neonatal hepatitis represents the most common diagnosis in infants with neonatal cholestasis. Often children have low birth weight and jaundice present within the first week. Acholic stools are uncommon. Often see giant cell transformation and extramedullary hematopoiesis on liver biopsy. Neonatal iron storage disease (or “hemochromatosis”) presents shortly after birth with fulminant hepatic failure and is, therefore, not in this differential diagnosis. Wilson’s disease, a rare autosomal recessive disease resulting in accumulation of copper in several organs, has not been reported in children younger than 4 years of age and is, thus, not consistent with this presentation. Biliary atresia accounts for 30% of cases of neonatal cholestasis. It is characterized by prolonged cholestatic jaundice in the newborn period, then hepatosplenomegaly. In those patients detected late in the process they present with abdominal distension, ascites and failure to thrive. The classic histopathology changes include variable canalicular and cellular bile stasis and bile duct proliferation.

16. You are asked to consult on the case of a 12-year-old child with a known diagnosis of Crohn’s disease. He has been on 20 mg of prednisone for 4 weeks now and is unable to wean below 15 mg for the last 2 months. Each time he weans his steroids he develops abdominal pain and diarrhea and stops eating. He is also noted to be < 10% for weight and < 3% for height. The family is asking for advice on how to get their child off of prednisone and are concerned about his growth failure and delayed puberty. The father has an adult height of 5’5” and the mother is 5’3’’ tall. The 9 year old sister is essentially the same height as her 12 year old brother with CD.

Which of the following management strategies would not be appropriate
A. Obtain a bone age
B. Consider 6-MP/azathioprine therapy
C. Consider caloric supplementation
D. Continue corticosteroids as the disease appears inactive on 20mg/day
E. Recommend vitamin D and calcium supplementation

The recommended response is D.
Growth failure can be attributed to many factors in this patient. Long term corticosteroid use has been shown to impair growth irrespective of disease activity. Moreover poor caloric intake as a result of persistent abdominal pain can limit intake resulting in weight loss and poor growth. In this case genetic factors may also contribute to the child’s short stature. A bone age would be helpful to see whether the child has delayed bone age as compared to his chronological age. In the case where the bone age is equivalent to the chronological age then the child most likely has genetic short stature. If the bone age is delayed as compared to the chronological age then malnutrition can certainly contribute to the short stature. In the case of CD, most patients have delayed bone age but the bone age is similar to the height age of the child (what age child would be if current height tracked to the 50%). In this scenario, additional calories would benefit the child and result in positive growth. Other causes of delayed bone age and delayed puberty include constitutional growth delay. A consultation with an endocrinologist would be helpful in certain scenarios and in the case of extreme growth delay; growth hormone may be a consideration. Thiopurines would be beneficial as a steroid sparing strategy and would be recommended in this case. Vitamin D and calcium supplementation is recommended in patients on long term corticosteroids. A QCT to assess bone density should be considered in children dependent on corticosteroids.

17. A 15-month-old-girl presents with diarrhea of 1-month’s duration, consisting of liquid, foul-smelling, greenish stools that are passed 5 to 6 times per day, with mucus but without visible blood. No vomiting was recorded but a diminished oral intake was noted, and she is thought to have lost 1 lb. Her past medical history includes short-gut syndrome; she was born prematurely at 33 weeks of gestational age and at day 6 of life presented with necrotizing enterocolitis (NEC). She underwent surgical resection of 60 cm of ileum, including the ileocecal valve. Subsequently, she was on TPN exclusively for 10 months and in the past 5 months also has been able to receive feedings first by nasogastric tube, then orally. Currently, she is receiving 700 ml of hyperalimentation by parenteral nutrition, providing 500 calories, and takes Pregestimil (a protein hydrolysates formula) by mouth in the amount of 16 oz, providing an additional 400 calories. Her weight 1 month ago was 8.5 kg. Physical examination reveals a somewhat weak-looking infant girl, mildly dehydrated, and borderline malnourished. Her weight is 8.1 kg and the abdomen is full and slightly distended, with hyperactive bowel sounds. Perianal skin is eroded and erythematous. What is the most likely explanation for this girl’s chronic diarrhea?

A. CMPA  
B. Celiac disease  
C. Giardiasis  
D. Small bowel bacterial overgrowth  
E. Chronic nonspecific diarrhea

The recommended response is D.

The diarrhea associated with CMPA typically develops during the first few months of life. Even though this patient has short gut, a condition thought to be more commonly...
associated with CMPA, the fact that she did well for over 4 months and that she is fed with a protein hydrolysate make this diagnosis unlikely. Celiac disease is not a consideration, given that there is no history of gluten in this girl’s diet. Giardiasis is a possible diagnosis, and should be ruled out by appropriate testing of stools for this protozoon 3 times. It is, however, an uncommon cause of chronic diarrhea in developed countries, and less likely than small bowel bacterial overgrowth (SBBO). Small bowel bacterial overgrowth is indeed highly suspected in this case, as the patient has had an extensive intestinal resection and the ileocecal valve (a known crucial factor in preventing high colonization of colonic bacteria) has been removed. Furthermore, the presence of skin erosions in the perianal area would suggest a very acidic stool pH, such as found in monosaccharide intolerance, a condition typical of SBBO.

18. For the patient in the vignette above, what is the best course of action?
A. Confirm diagnosis by a glucose breath test  
B. Perform a diagnostic EGD  
C. Obtain a stool culture to confirm the diagnosis of SBBO and identify the pathogen involved  
D. Start therapy with high doses of a probiotic  
E. Start therapy with oral antibiotics

The recommended response is A.

Options B and C are clearly wrong: the EGD won’t help in making the diagnosis, as if changes are seen in the duodenal mucosa they are unspecific and won’t be helpful in directing treatment. Although stool cultures are useful in ruling out an infectious enteritis. They are not needed to diagnose SBBO, as the bacteria responsible for this syndrome are not pathogenic ones but rather the ordinary colonic flora that becomes resident in the small intestine. Probiotics (option D) have been tried with inconsistent success in SBBO and no controlled trials have been published to support their usefulness. Empiric therapy with PO antibiotics (E) can be used only if a diagnostic procedure is not available. Indeed, a major drawback to the approach with empiric therapy is that treatment may require more than one antibiotic and sometimes cyclic treatment. Thus, because many antibiotics may be associated with adverse effects some of which may mimic symptoms of SBBO (such as diarrhea and abdominal pain), establishing a clear diagnosis is important. Obtaining the diagnosis with a glucose breath test is the best option. The combination of an elevated fasting breath H2 concentration and increased breath H2 excretion after a glucose load has been reported to have a specificity of 100% for SBBO.

19. Which of the following statements is not true when dealing with IBD in children?
A. Its incidence appears to be increasing worldwide  
B. Children with UC tend to have a higher likelihood of pancolitis at presentation  
C. Steroid sparing strategies is important for the treatment of pediatric IBD  
D. Isolated Small bowel disease is more common in children than adults  
E. Growth failure is more common in UC then CD
The recommended response is E.

The only false statement is reported in option E. Growth failure is more common in CD than UC. In patients with significant growth failure carrying a diagnosis of UC must be investigated for evidence of Crohn’s disease most noticeable in the more proximal small bowel. The video capsule endoscopy may be a very useful diagnostic tool in this case.

20. You examine a 1-month-old-boy who is brought to your attention for a distended abdomen. The parents tell you that he was born at full term by a normal delivery, he is their first-born, and was breast-fed from birth. He is reported to have had passage of meconium on day 2 of life. He did well and was sent home on day 3, where he continued to thrive while breast-fed, but the frequency of his bowel movements decreased, and currently he has 1 bowel movement of normal stools, passed with evident straining, every 2 to 3 days.

At physical examination, he is well nourished and alert. Abdomen is very distended and full: palpation does not elicit tenderness. No masses are evident. Perianal inspection is normal. Rectal exam: your finger enters with difficulty through a narrow anus, and you do not touch stools; at retraction, loose, yellowish stools are passed explosively. A plain x-ray of the abdomen confirms the distention and shows dilated loops of colon, but no stool accumulation.

What is the first priority in caring for this patient?
A. Rule out the diagnosis of Hirschsprung’s disease.
B. Provide him with a stool softener
C. Administer an enema
D. Change the diet to soy milk to eliminate cow’s milk protein introduced via breast milk
E. No action is needed, as this baby obviously is healthy

The recommended response is A.

Hirschsprung’s disease is a diagnostic possibility and needs to be ruled out as soon as possible. In fact, even though this case lacks the most obvious landmarks of the condition such as the delayed passage of meconium, this presentation is not unusual for patients who have a short segment. The physical examination findings are highly supportive of this diagnosis. Using stool softeners or enemas is not indicated, as this patient does not have hard stools (as seen both from history and at the physical examination), and they would be contraindicated in suspected HD. No signs consistent with CMPA are reported, and discontinuing breast-feeding would be a poor option. Finally, although this baby is well nourished, he is not healthy, as the abdomen is very distended and diagnostic steps need to be undertaken. Testing may involve first a barium enema and/or an anorectal manometry, followed—in cases still consistent with HD—by the definitive diagnostic test: the rectal biopsy.