Symptoms and signs of lower gastrointestinal disease

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Abstract

This chapter provides a physiological basis for understanding a practical approach to patients presenting with the lower gastrointestinal symptoms. These symptoms can arise from disturbances of gut motility (diarrhoea, constipation), coordination and function of the anorectum (evacuation difficulty, faecal incontinence) and structural deficits of the hindgut (rectal bleeding). Abdominal pain is a common presentation, and can be related to physiological or anatomical changes in the gut. This chapter describes the clinical assessment of such patients to discern the likeliest diagnosis of their abdominal pain.

Keywords Abdominal pain; constipation; diarrhoea; faecal incontinence; MRCP

Key points

- Lower gastrointestinal symptoms are common and frequently have a benign cause, but eliciting alarm symptoms is key to excluding the possibility of organic disease
- With diarrhoea, determining the characteristics of the stool as secretory, osmotic, inflammatory or dysmotility-related helps define the likeliest causes
- With constipation, symptoms can help differentiate between slow transit, evacuation difficulty, a combination of these and irritable bowel syndrome
- Faecal incontinence, although common, is under-reported by patients; it is important to establish whether the symptoms result from impairment of the anal sphincter or the rectum
- Although most cases of rectal bleeding have benign local causes, it is essential to consider the possibility of colonic diseases amenable to treatment

Diarrhoea

Stool consistency is determined by the interaction between water secretion and absorption within the bowel. When secretion exceeds absorption, watery stools are produced. Diarrhoea is objectively defined as a stool weight >200 g within a 24-hour period. However, because this is not a practical diagnostic feature in clinical practice, three or more loose or liquid stools (Bristol Stool Form Scale (BSFS) type 5 or above; Figure 1) per day is used.[1] The frequency and consistency of normal bowel movements vary widely, and it is important to elicit whether there has been a change from the norm.

Acute diarrhoea (typically defined as lasting <4 weeks)[1] is likely to be the result of infection. Questioning regarding recent foreign travel and food consumption may elicit the source, although it is important also to consider acute diarrhoea as the initial presentation of a chronic cause. In the UK, if certain infections are suspected, this can require prompt notification to Public Health England to prevent possible epidemics. The most common of these is food poisoning.

Chronic diarrhoea (symptoms over 4 weeks) is a common presentation, affecting up to 5% of populations in the developed world.[1] Although an infective cause (e.g. *Giardia*) is possible, more probable differential diagnoses exist. It is helpful to think of these in terms of the underlying pathophysiology, which can be categorized into three main groups. History-taking should include stool characteristics as well as associated features (Table 1). It is important to remember that medications can also cause diarrhoea.

Secretory diarrhoea

Clinical features: watery, large-volume frequent bowel movements occur. Often, there is no associated pain. Diarrhoea does not settle with periods of fasting.

Pathophysiology: there is increased Cl^- secretion and impaired Na^+ transport caused by activation of transport processes within the enterocyte (often caused by a peptide or toxin). This leads to increased fluid secretion and decreased absorption, causing watery stools.

Examples:

- Toxins, for example from *Escherichia coli* and *Vibrio cholerae*.
- Hormones, for example carcinoid tumours and vasoactive intestinal peptide.
- Bile acid diarrhoea (increased levels of bile salts in the colon stimulate electrolyte secretion).

Osmotic diarrhoea

Clinical features: bulky or 'pasty' stools are seen, which settle with fasting or avoidance of the causative substance. Steatorrhoea-type stool can occur: pale stools that are difficult to flush.

Pathophysiology: the cause is ingestion of poorly absorbed substances that are osmotically active (often magnesium salts

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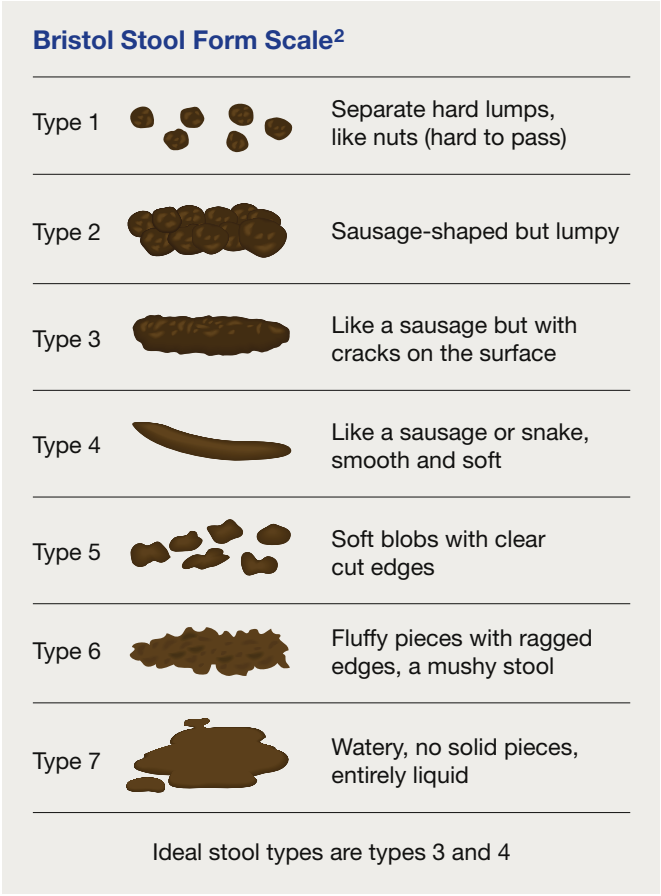

Figure 1

and sugars such as mannitol). This leads to fluid retention within the bowel lumen.

Examples:

- Malabsorption disorders — coeliac disease, bowel resection and bacterial overgrowth.
- Maldigestion disorders — lactase deficiency and pancreatic insufficiency (as seen in chronic pancreatitis and cystic fibrosis).
- Overuse of osmotic laxatives.

Associated features

- Steatorrhoea (pale stools that are difficult to flush) indicates small bowel or pancreatic pathology
- Abdominal pain relieved by bowel movement suggests irritable bowel syndrome
- Weight loss and nocturnal diarrhoea indicate an organic cause — this must be investigated
- Light-headedness, tachycardia and low urine output can occur secondary to dehydration
- Blood in the stool indicates inflammatory bowel disease or infectious colitis
- Oral ulceration and perianal disease are seen in Crohn's disease

Table 1

Motility-related diarrhoea

Clinical features: this is associated with cramping abdominal pain. There is a wide variation in stool consistency, and episodic constipation can occur.

Pathophysiology: a variable rate of transit through the digestive tract is related to gut, nerve or muscle dysfunction. Decreased contact time between the intestinal contents and the mucosa prevents sufficient reabsorption.

Examples:

- Irritable bowel syndrome (IBS).
- Stimulant laxative overuse.
- Intestinal resection.

In patients with inflammatory bowel disease (IBD), the underlying pathophysiology is likely to be a combination of mechanisms and it can therefore be difficult to treat. Patients can complain of blood in the stool and have additional symptoms such as fever, cramping abdominal pain and tenesmus.

All patients should be asked about symptoms of dehydration, a common complication of diarrhoea. Inability to retain oral fluids and signs of severe dehydration require hospital admission.

Constipation

Constipation is a frequent complaint, experienced by 1 in 7 adults and 1 in 3 children (depending on the criteria used).[3] In the developed world, women are more commonly affected than men, and prevalence increases with age. Over one-third of women experience constipation during pregnancy, especially in the first two trimesters. The costs of constipation are high — data from the UK for 2015 suggest that the cost of unplanned hospital admission for constipation was £145 million, laxative costs were £101 million and constipation caused a reduction of 2.4 days per month of work or school productivity.[3]

The symptoms of constipation are multiple, but the standard for classification in clinical trials is set by the Rome IV criteria (Table 2). These criteria are, however, of little value in clinical practice given their complexity of application. A more practical classification depends on the individual's perception of normal bowel habit and refers to either infrequency of or difficulty with defecation. Patients with self-reported constipation are most bothered by symptoms of straining, abdominal discomfort and a sensation of incomplete evacuation. Describing stool consistence can be difficult for some patients, and the BSFS (comparing with types 1 and 2) can be helpful.

It is important to classify constipation into primary and secondary causes because of the differences in management. Secondary constipation can be a consequence of systemic conditions, medications or structural abnormalities within the gastrointestinal tract. Specific causes of secondary constipation are shown in Table 3. Primary constipation is related to colonic or anal dysfunction and can be characterized pathophysiologically into four main categories:

- slow transit
- defecation disorder
- a combination of slow transit and defecation disorder
- normal transit.

Rome IV criteria for functional constipation

1. Must include two or more of the following:
   a. Straining during more than one-fourth (25%) of defecations
   b. Lumpy or hard stools (Bristol Stool Form Scale 1–2) more than one-fourth (25%) of defecations
   c. Sensation of incomplete evacuation more than one-fourth (25%) of defecations
   d. Sensation of anorectal obstruction/blockage more than one-fourth (25%) of defecations
   e. Manual manoeuvres to facilitate more than one-fourth (25%) of defecations (e.g. digital evacuation, support of the pelvic floor)
   f. Fewer than three spontaneous bowel movements per week
2. Loose stools are rarely present without the use of laxatives
3. Insufficient criteria for irritable bowel syndrome

Table 2

Slow transit constipation

This is typically a disorder of young women who often give a history of constipation since childhood. The cause can relate to colonic neuromuscular dysfunction. The reduced motility (in both colon and small bowel) leads to a prolonged transit time and hence an infrequent urge to defecate and a harder, drier and small volume stool. Abdominal pain and bloating are also features.

Defecation disorders

This can arise from a structural or a physiological disorder. Mechanical causes include a rectocele (anterior bulge of the rectum into the posterior wall of the vagina), rectal prolapse or intussusception (of the rectoanal unit). The latter may be related to the physiological component, which can in turn be related to a failure of relaxation of the pelvic floor muscles or paradoxical contraction of the internal anal sphincter ('anismus') during attempted defecation. These pelvic floor factors are often contributed to by ineffective propulsion from the abdominal musculature. Patients present with an inability to void their rectum despite urge, so they can strain despite having soft stools, and can have a sense of incomplete emptying. They may need to adopt uncomfortable positions on the toilet to void, and use digital manoeuvres to aid defecation.

Secondary causes of constipation

- **Medications** – opiates, ferrous supplements, tricyclic antidepressants, diuretics, antipsychotics
- **Intrinsic colonic pathology** – colorectal cancer, diverticular disease
- **Metabolic/endocrine** – hypercalcaemia, coeliac disease, hypothyroidism, hypocalcaemia
- **Neurological** – spinal cord injury, multiple sclerosis, Parkinson's disease
- **Psychological** – depression, anorexia nervosa, bulimia, affective disorders abuse

Table 3

Normal transit constipation

This is a term given to the presence of constipation symptoms without a demonstrable delay in transit or pelvic floor dysfunction. The symptoms can be just as intrusive for the patient as those of the other pathophysiological subtypes.

A small proportion of patients presenting with constipation have a megacolon. This can be either inherited or acquired, results in dilatation of the rectum and can involve the more proximal colon. The dilatation associated with megarectum results in a loss of tone and slowed transit, so patients present with features of both slow transit and difficult evacuation.

Assessment

The clinical history is key to identifying possible alarm features (Table 4) and secondary causes of constipation. With regard to primary causes, although there is a great deal of symptom overlap, history-taking can point more towards one form of pathophysiology (as outlined above).

Abdominal examination is essential to identify potential secondary causes (especially endocrine or neurological). An abdominal faecal mass can suggest a megacolon. A digital rectal examination is important to exclude faecal impaction (remembering that a small amount of soft stool is present in the normal rectum), and to identify possible colonic lesions causing the problem. Equally important, careful examination with the finger can help to identify paradoxical pelvic floor or sphincter contraction during attempted voiding. Identifying haemorrhoids that have complicated chronic constipation, or the presence of anal fissures or rectal prolapse, is a key aspect of the perineal examination.

Alarm features of constipation[2]

- Age >50 years
- Short history of symptoms
- Male sex
- Unintentional weight loss
- Nocturnal symptoms
- Anaemia
- Rectal bleeding
- Abdominal mass
- Recent antibiotic use
- Family history of bowel or ovarian cancer

Table 4

Faecal incontinence

Faecal incontinence is the involuntary passage of stool (and flatus). It affects up to 10% of adults, increasing in prevalence with age.[4] This is, however, likely to be an underestimate because of the perceived stigma attached to the symptoms. Patients experience a significant decrease in quality of life, leading to low mood and social isolation. It is important to be aware that faecal incontinence is a symptom, not a diagnosis, and often has a complex aetiology with many contributory factors (Table 5).

Factors important for continence (Table 6)

- Function of the internal (involuntary) and external (voluntary) anal sphincters.

- Neurological motor control over the anal canal and pelvic floor muscles.
- Neurological sensory function to sense contact at an appropriate volume and timing.
- Anal cushions (as a mechanical barrier).
- Stool consistency.
- Rectal compliance (to accommodate stool delivered by the colon).

Defects in any one or more of these can cause faecal incontinence. Common causes of incontinence are cited in Table 5. Faecal incontinence is clinically classified into two symptom types, as described below.

Urge incontinence:

- There is a sudden need to defecate with insufficient warning to reach a toilet.
- Causes are diarrhoea, weakness or defects of the external anal sphincter, and loss of normal anorectal sensation.
- The symptom of urgency itself (even without incontinence) is distressing and limits quality of life.

Passive incontinence:

- Loss of faeces or flatus occurs without sensory awareness, resulting in soiling of underwear or insensate loss of wind.
- Causes are diarrhoea, weakness or defects of the internal anal sphincter, damage to the anal cushions and loss of rectal compliance.

Symptom assessment should include classifying the kind of incontinence (as above), as well as the frequency and consistency of stool passed. It is important to enquire about the obstetric, neurological and surgical history to determine the potential aetiology. The history needs to be sensitively taken to address the patient's potential embarrassment and hesitance. In addition, the impact on the patient's lifestyle needs to be addressed — include dietary patterns, lifestyle adjustments and medications used. Assessment of symptom burden and quality of life impact can be supplemented by using severity scales such as the Cleveland Clinic Incontinence or St Mark's Incontinence scores.

General examination can reveal evidence of abdominal scars or neurological disease. Perianal inspection may reveal evidence of obstetric trauma, significant haemorrhoids or rectal prolapse (which can present at any age). Digital rectal examination is essential to assess anal tone at rest (internal anal sphincter function) and on contraction (external anal sphincter function). It can also aid the diagnosis of chronic constipation (which may be causing overflow diarrhoea).

Common causes of faecal incontinence

- **Obstetric**
- **Iatrogenic** — sphincterotomy, hamorrhoidectomy, fistula surgery, anal stretch
- **Direct trauma** — impalement injury, non-consensual anal intercourse
- **Radiation damage**
- **Colorectal** — rectal prolapse, prolapsing haemorrhoids, internal anal sphincter atrophy
- **Congenital** — imperforate anus, anal agenesis
- **Neurological** — spina bifida, spinal cord injury, multiple sclerosis
- **Urogynaecological** — pelvic organ prolapse
- **Medical** — irritable bowel syndrome, inflammatory bowel disease, obesity, diabetes, coeliac disease, drugs

Table 5

Physiology of continence

Arrival of faeces or flatus into the rectum is detected by stretch receptors and is, at low volumes, accommodated. At critical fill volume, new content causes relaxation of the internal anal sphincter If defecation is inconvenient, voluntary contraction of the external anal sphincter occurs. This allows anal pressure to remain higher than rectal pressure and delays evacuation

Table 6

Rectal bleeding

The passage of blood per rectum (haematochezia) is common, affecting approximately 10% of UK adults at any one time. Although it is most often caused by benign conditions of the colon or anus, serious conditions such as malignancy and IBD must be excluded (Table 7). Haemodynamic instability associated with large volume rectal bleeding should provoke consideration of an upper gastrointestinal source.

It is important to be precise about the exact symptom a patient is describing. Details of whether blood is seen mixed in the stool, in the toilet bowl or only on wiping can help to localize the source: blood seen only on wiping after defecation is likely to be have an anal cause (fissure, haemorrhoids), whereas blood mixed in the stool can have originated from higher up in the gastrointestinal tract.

Key points in the history

The following should be noted:

- **Duration and frequency of bleeding** — is it acute, intermittent or chronic?
- **Volume of blood** — are there streaks or frank blood? The passage of clots suggest heavier bleeding.
- **Colour of blood** — bright red blood is usually from the left colon or distal to that; dark blood is typically caused by a more proximal lesion.
- **Change in bowel habit.**
- **Associated symptoms** — haematemesis suggests an upper gastrointestinal cause; passage of mucus can be associated

Common causes of rectal bleeding

- Haemorrhoids
- Anal fissure
- Diverticular disease
- Colorectal or anal malignancy
- Inflammatory bowel disease
- Colonic polyps
- Ischaemic colitis
- Infection

Table 7

with IBD or rectal adenomas; dizziness and syncope can indicate large volume blood loss; associated pain and fevers raises the possibility of diverticular disease.

- **Other alarm features** – these include weight loss, an abdominal or rectal mass, a personal or family history of IBD or bowel cancer, and anaemia.
- **Past medical history** – remember to include IBD, coagulopathies and conditions requiring anticoagulation.
- **Drug history** – anticoagulants and conditions requiring anticoagulation are especially noteworthy.
- **Travel history.**

Age is important in determining the likeliest differential diagnoses of rectal bleeding: new rectal bleeding in patients >50 years of age should raise the possibility of malignancy or diverticular disease, with consideration of urgent referral for investigation. Alongside abdominal examination, all patients require a digital rectal examination to exclude a palpable rectal mass. It is important to remember that the discovery of haemorrhoids or an anal fissure on examination does not exclude other causes of rectal bleeding.

Abdominal pain

Abdominal pain is a common presentation to both primary and secondary care. Annually, abdominal pain is responsible for 300,000 emergency admissions and a further 90,000 non-urgent hospital admissions in England alone.[5] The differential diagnoses of abdominal pain are vast and arise from a variety of different body systems (Table 8); because of this, history-taking is of the utmost importance to ascertain the most likely diagnosis. Despite investigation, many patients with chronic abdominal pain (lasting >6 months) have no evidence of organic pathology.

The character of the pain aids identification of the underlying pathology. Visceral pain is a consequence of distension, inflammation or ischaemia of the viscera or the capsules in which they are encased. This pain is characteristically described as a diffuse in location and dull in character. The general pain location correlates to the embryological origin of the affected organ. Because of its association with the autonomic nervous system, pain can be associated with autonomic features such as sweating, pallor and changes in vital signs. Conversely, somatic pain arises from irritation to the parietal peritoneum that lines the abdominal and pelvic cavities. This produces well-localized, sharp pain that the patient can accurately point to using one finger.

Common differential diagnoses of abdominal pain

Site	Differential diagnosis
Small bowel	• Duodenal ulcer and/or perforation • Small bowel obstruction • Irritable bowel syndrome • Crohn's disease
Colon	• Diverticulitis • Large bowel obstruction • Inflammatory bowel disease • Ischaemic colitis
Liver and biliary system	• Hepatitis • Acute cholecystitis • Ascending cholangitis • Liver abscess • Liver failure
Kidney and urological tract	• Pyelonephritis • Cystitis • Renal stones • Acute urinary retention • Malignancy
Gynaecological tract	• Pelvic inflammatory disease • Ectopic pregnancy • Endometriosis • Dysmenorrhoea • Mittelschmerz • Ovarian torsion • Ruptured ovarian cyst
Other	• Acute appendicitis • Acute pancreatitis • Diabetic ketoacidosis • Lower lobe pneumonia • Abdominal aortic aneurysm • Irritable bowel syndrome • Splenic rupture • Familial Mediterranean fever • Porphyria

Table 8

History

- **Location** – is it well-localized or diffuse? Epigastric pain is related to foregut structures; mid-abdominal pain relates to midgut structures; suprapubic pain derives from hindgut pathology.
- **Onset** – sudden-onset pain can be secondary to infarction, rupture or perforation; gradual pain can arise from chronic inflammation or malignancy.
- **Character** – is there a dull ache (visceral pain) or a sharp, stabbing pain (somatic pain)? Wave-like colicky pain suggests intestinal obstruction or renal or biliary colic.
- **Radiation** – shoulder tip pain results from diaphragmatic irritation; renal colic radiates to the groin; back pain can be referred from an abdominal aortic aneurysm or pancreatic conditions.
- **Exacerbating and alleviating factors** – correlation with meals can indicate peptic ulcer disease, cholecystitis or small bowel pathology; pain alleviated by defecation suggests rectal pathology or IBS; pain worsened by movement may be peritonitis.
- **Associated symptoms** – the presence of vomiting suggests gastroenteritis, appendicitis, pancreatitis or small bowel obstruction; respiratory symptoms can suggest lower lobe pneumonia; rectal bleeding may indicate IBD.
- **Last menstrual period (in female patients of childbearing age)** – mid-cycle pain suggests Mittelschmerz; missed periods raise the suspicion of ectopic pregnancy.
- **Past medical and surgical history** – adhesions from previous abdominal surgery can lead to small bowel obstruction.

- **Social history** – alcohol excess is a known risk factor for pancreatitis; there is an association between smoking and the development and severity of Crohn's disease.

Examination should include general inspection, looking for signs of cachexia. Peripheral stigmata of liver disease such as spider naevi, bruising, palmar erythema and jaundice increase speculation of liver cirrhosis. Abdominal examination should include palpation of all areas (the abdomen classically being divided into a 3 × 3 grid), beginning away from the affected area. Tenderness, guarding and rebound tenderness should be noted. Palpable masses require urgent investigation. It is important to note that even large volume ascites can be painless. Hyperactive bowel sounds suggest intestinal obstruction, whereas reduced or absent bowel sounds can be present in peritonitis or paralytic ileus. Inspection of the inguinal area and external genitalia is vital to exclude hernias and testicular torsion, respectively.

In light of the differential diagnoses (Table 8), a digital rectal examination to exclude gynaecological and anorectal causes is an important component of the examination in the patient with abdominal pain. Once consent is granted, a pregnancy test should be performed in all women of childbearing age who present with abdominal pain, to exclude gynaecological emergencies such as ruptured ectopic pregnancy. ◆

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TEST YOURSELF

To test your knowledge based on the article you have just read, please complete the questions below. The answers can be found at the end of the issue or online here.

Question 1

A 36-year-old man presented with a 3-month history of opening his bowels five or six times a day. The stools were watery and occasionally pale in colour. There was occasional nocturnal diarrhoea. He had type 2 diabetes mellitus and was taking a sulfonylurea. He had had a cholecystectomy 2 years previously. There had been no weight loss, and the results of blood tests, including tissue transglutaminase, were normal.

Which of the following are the likeliest causes of his diarrhoea?

A. Bile acid diarrhoea
B. Diverticular disease
C. Drug-induced diarrhoea
D. *Helicobacter pylori* infection
E. Small intestinal bacterial overgrowth

Question 2

A 27-year-old woman presented with a 5-year history of abdominal bloating, weight gain and constipation, as well as recent anal discomfort with no bleeding. The bloating was eased by bowel opening, which occurred every 5–7 days. Stool consistency varied from pellet-like to semi-formed. She strained at stool on every occasion.

What is the mostly likely finding on rectal examination?

A. A tear in the anal skin is normal with chronic straining, and unlikely to be a cause of her pain
B. Contraction of the internal anal sphincter is normal on bearing down
C. Low resting anal pressure excludes an anal fissure
D. Rectal prolapse is possible despite her young age
E. The presence of stool in the rectum confirms the diagnosis of faecal impaction

Question 3

A 53-year-old woman had presented with four stereotyped episodes in the previous 8 months. She had developed acute abdominal pain with vomiting; on each occasion, this had settled within 24 hours, after which she had felt well. She did not pass stools for a day or two, and tended to avoid meals for that period. On the latest occasion, the pain had persisted for 36 hours. Her weight was steady, and her only previous hospitalization was for peritonitis complicating acute appendicitis at age 13 years.
On clinical examination, she was uncomfortable and afebrile. Her abdomen was tender, with active bowel sounds.

What is the most likely diagnosis?

A. Diverticular disease
B. Endometriosis
C. Inflammatory bowel disease
D. Irritable bowel syndrome
E. Subacute intestinal obstruction