

- Pulmonary valve stenosis
- Webbed neck
- Failure to thrive, usually mild
- Abnormalities of cardiac conduction and rhythm
- ± Intellectual disability

Treatment

- Refer to genetic service
- Evaluation of cardiac status
- Consider vision, hearing, clotting status, possible epilepsy

§ Angelman syndrome

Genetic profile

- Abnormal chromosome 15

Clinical features (a wide spectrum)

- Hand flapping
- ‘Puppet’-like ataxia
- Frequent laughter/smiling
- Microcephaly by age 2 years
- Developmental delay
- Speech impairment
- Seizures
- Cannot live independently

Diagnosis based on clinical features and genetic studies.

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Treatment with minocycline is promising.

§ Progeria

Sporadic mutation of LMA gene that codes for a protein leads to early cell death.

Accelerated ageing—manifests in early childhood causing premature death (median age 12 years) from vascular disease. No known treatment.

Sex chromosome abnormalities

Klinefelter syndrome¹⁰

This is due to an extra X chromosome, resulting in a male phenotype and occurring in 1 in 800 live births. Approximately 2 out of 3 are never recognised.



DxT lanky men + small testes + infertility → Klinefelter syndrome

Genetic profile

- 47, XXY genotype
- The extra X chromosome is usually of maternal origin
- About 30 or more variants of the disorder

Clinical features

Marked variation but usually:

- tall men with long limbs
- small firm testes ≤2 cm (10 mL)
- infertility (azoospermia)

There may be:

- sparse facial hair
- reduced libido
- learning difficulties, especially reading
- intellectual ability may range from normal to disability
- gynaecomastia
- increased risk of DVT, breast cancer and diabetes (screening indicated)

Diagnosis

- Increased gonadotrophin, low to normal testosterone

Treatment

- Transdermal testosterone

⌚ Turner syndrome (gonadal dysgenesis)

This is due to only one X chromosome, occurring in 1 in 4000 live female newborns; 99% of conceptions are miscarried.¹⁶



DxT short stature + webbed neck + facies → Turner syndrome

Genetic profile

- 45 chromosomes of XO karyotype (typical Turner karyotype in 50% of cases)
- Many are mosaics (e.g. 45X/46XX chromosomes)
- Phenotypes vary

Clinical features of typical XO karyotype

- Short stature—average adult height 143 cm
- Primary amenorrhoea in XO patient; infertility
- Webbing of neck
- Typical facies: micrognathia, low hairline
- Lymphoedema of extremities
- Cardiac defects (e.g. coarctation of aorta)

Mental deficiency is rare.

Treatment

- Hormone-based (e.g. growth hormone, hormone replacement therapy)

Intersex states

These are uncommon chromosomal disorders of sexual development (DSD) in which the appearance of the external genitalia is either ambiguous or at variance with the individual's chromosomal sex. Intersex is an umbrella term to describe a wide range of natural body variations. The inappropriate term 'hermaphrodite' should be avoided.

The conditions include:

- mixed gonadal dysgenesis
- ovotesticular disorder DSD
- 46, XX DSD (androgenised females)
- 46, XY DSD (underandrogenised males)

The latter may be caused by inadequate production of androgen or inadequate response to androgen, which includes the 'androgen insensitivity syndrome'. This syndrome, made prominent through athletes with a male genotype but female phenotype, results from a mutation of the gene encoding the androgen receptor.

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⌚ Congenital adrenal hyperplasia

See [CHAPTER 14](#).

Developmental delay and intellectual disability

- Chromosomal microdeletion syndromes (preceding chromosomal conditions; some such as FKS and tuberous sclerosis show features of ASD)
- Autism spectrum disorder (ASD): if diagnosed, refer to paediatrician for chromosomal microarray investigation.
- Fetal alcohol spectrum disorder (FASD)

⌚ Fetal alcohol spectrum disorder (FASD)¹⁹

FAS is the most severe form of the fetal alcohol spectrum disorders. These are caused by the teratogenic effects of alcohol (not a chromosomal abnormality) and is estimated to involve 2 in 1000 live births. The phenotype varies with the dosage and gestational timing of the alcohol exposure.¹⁹ Caution is needed not to overdiagnose.



DxT abnormal facies + growth retardation + microcephaly + history of alcohol intake during pregnancy → fetal alcohol spectrum disorder

Clinical features of FASD

See [FIGURE 23.2](#) .

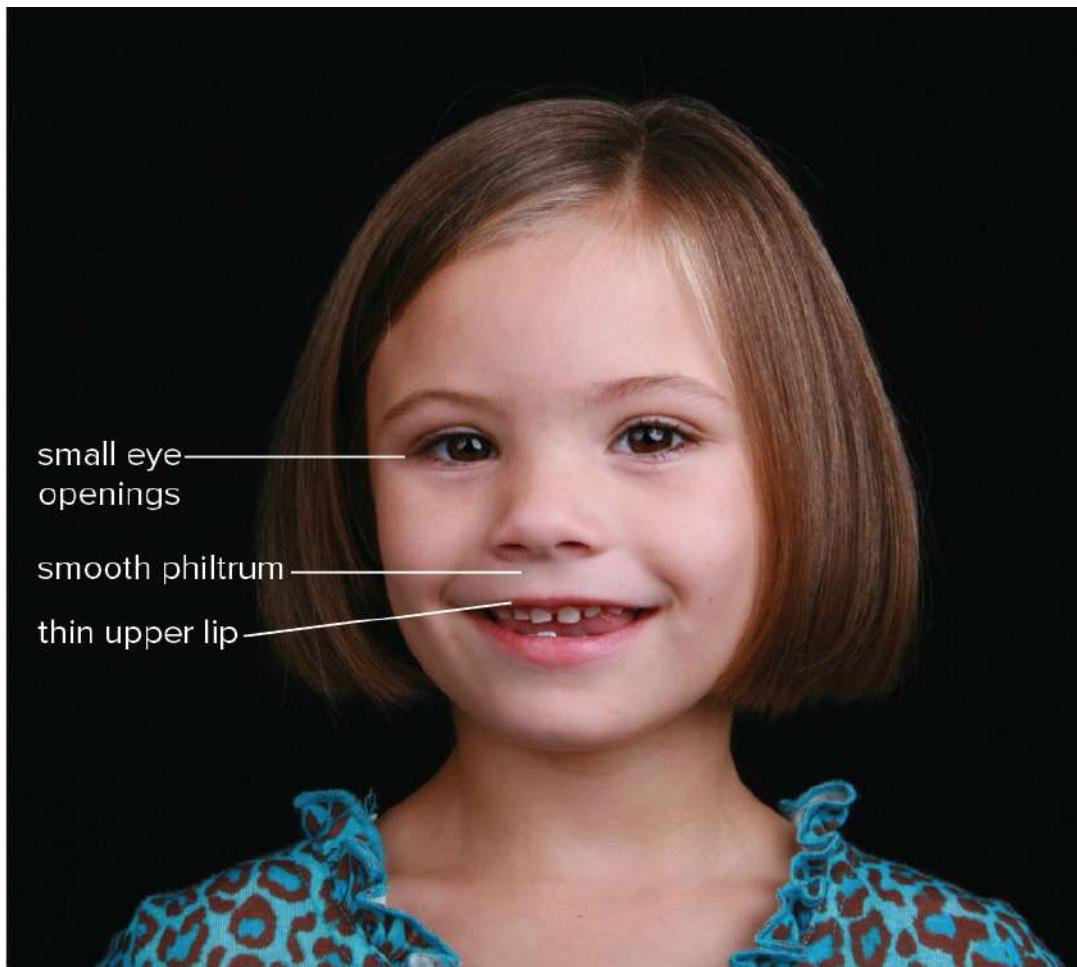


FIGURE 23.2 Fetal alcohol syndrome: facial features

Source: Rick's Photography/Shutterstock

- Markedly underweight until puberty
- Learning difficulties
- Microcephaly
- Characteristic facies (needs 2 of *)

shortened palpebral fissures*

long, smooth featureless philtrum*

thin upper lip*

upturned nose

- Hyperactivity
- Congenital heart disease often seen
- Skeletal abnormalities

Diagnosis based on alcohol history in pregnancy.

Management

- Early diagnosis and intervention
- By preventive strategies with community education about the harmful effects of drinking, especially in early pregnancy
- Counselling; addressing environmental and behavioural factors

Other types of hereditary disorders

The many examples include familial hypercholesterolaemia (AD) and the AR disorders for which genetic testing is available, namely Gaucher disease, glycogen storage disease, phenylketonuria, skin disorders, polycystic kidney disease, galactosaemia, homocystinuria, the porphyrias and the mitochondrial disorders.

Single gene cardiac disorders

Includes:

- cardiomyopathies
- arrhythmia syndromes, e.g. long QT syndrome
- sudden cardiac death families

Congenital long QT syndrome

This is an autosomal dominant condition with predisposition to ventricular arrhythmias, syncopal/fainting spells and sudden death, particularly during exercise. Confirm or exclude by ECG when suspected—interval 0.5–0.7 seconds. Management includes sports restrictions, beta blockers and pacemaker or AICD.

Familial hypertrophic cardiomyopathy

This is an AD disorder with several genetic mutations. It is the most common cause of sudden cardiac death among athletes.

Clinical features

- Fatigue
- Exertional dyspnoea and chest pain
- Palpitations
- Dizziness/syncope

Diagnosis by ECG (LV hypertrophy) and doppler echocardiography.

Insertion of AICD may prevent sudden death.

Familial hyperlipoproteinaemia²⁰

There are several types of genetic disorder of lipid metabolism including the better-known familial hypercholesterolaemia and familial combined hyperlipidaemia. The former is identified by elevated cholesterol, corneal arcus juvenalis, tendon xanthomas in the patient or their first- and second-degree relatives and also by a DNA mutation. Homozygous patients present with atherosclerosis disease in childhood and early death from myocardial infarction. Heterozygotes may develop the disorder in their 30s or 40s.

This is common, being 1 in 500 Caucasians, and 1 in 70 in Lebanese and Afrikaners. Eighty per cent are undiagnosed and missing out on preventive treatments. GPs have an important role in screening people with premature ischaemic heart disease to look for phenotype characteristics of the condition.

Familial cancer²¹

The majority of cancer is not inherited; rather it is acquired because of genetic mutations in several genes of a cell in a specific tissue during an individual's lifetime. Twenty to twenty-five individuals in a population of 1000 have a family history of bowel or breast cancer.⁸

However, some people carry inherited genetic mutations from conception that predispose them to developing certain cancers, particularly colorectal, breast and ovarian cancer, at a relatively young age. Up to 5% of some cancers are considered familial and the genetic basis of some of these is now understood. Most are AD with 50% of offspring being affected.

The three most significant familial cancer inherited susceptibility syndromes are:

- hereditary breast–ovarian cancer syndrome (*BRCA*₁ and *BRCA*₂ genes)
- hereditary non-polyposis colorectal cancer (HNPCC)
- familial adenomatous polyposis (FAP)

Features of breast–ovarian cancer syndrome^{9,10}

- Mutations in either of the two genes—*BRCA*₁ and *BRCA*₂—result in a strong predisposition for both breast and ovarian cancer
- Mutations present in about 1 in 800 of the general population (male and female), who are carriers
- Dominant inheritance
- The risk of developing breast cancer is 10-fold and 40–80% of cases occur before the age of 70 years²²
- The prognosis in these women is the same as for sporadic cases
- Early age of onset of breast cancer
- Male breast cancer (6% in males with *BRCA*₂ gene mutation)
- Coexistence of ovarian and breast cancer in the same family
- Carriers of mutations *may* be at an increased risk of prostate cancer, pancreatic cancer and colorectal cancer, although this is controversial for the latter two

Risk indicators for familial breast–ovarian cancer

- Two first-degree or second-degree relatives on one side of the family with cancer
- Individuals with age of onset of cancer <50 years
- Individuals with bilateral or multifocal breast cancer
- Individuals with ovarian cancer
- Breast cancer in a male relative
- Jewish ancestry

Colorectal cancer²¹

Both sexes have a risk of approximately 5% of developing bowel cancer in their lifetime. In

some this risk is increased due to an inherited predisposition.

The two key disorders are HNPCC and FAP.

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Lynch syndrome (hereditary non-polyposis colorectal cancer)

- Caused by a defect in one of the genes responsible for DNA mismatch repair
- Affects 1 in 1000 individuals
- Autosomal dominant
- Early age of onset
- Increased risk of certain extracolonic cancers, including endometrial, stomach, ovary and kidney tract cancers
- Screening should occur every 1–2 years from 25 years of age or 5 years earlier than affected family member developed it

Familial adenomatous polyposis

- Less common than HNPCC; affects about 1 in 10 000
- Caused by a mutation in the *APC* gene
- Usually hundreds or thousands of polyps
- Eventually almost 100% of cases develop colon cancer without prophylactic colectomy
- Median age of diagnosis 40 years
- Small increased risk of other cancers (e.g. thyroid, cerebral)
- Screening should occur annually from between 12–15 and 30–35 years of age, and then every 3 years

Individuals at risk

For Lynch syndrome (HNPCC):

- Three or more close relatives with bowel cancer
- Two or more close relatives with bowel cancer and:
 - more than one bowel cancer in same relative

- onset of bowel cancer before 50 years
- a relative with endometrial cancer or ovarian cancer

For FAP:

- A relative with bowel cancer with polyposis
- Individuals with multiple polyposis

Other cancers where family history is significant

- Melanoma: an inherited mutation in certain genes (e.g. *BRAF* gene) is considered to be involved in up to 5% of melanomas. Having a first-degree relative affected almost doubles a person's risk.
- Prostate: family history is a risk factor; some genes (e.g. *BRAC₁* and *BRAC₂*) are susceptible. Refer if a significant family history.
- Several other cancers can develop as a mutation, e.g. stomach, pancreas, kidney, thyroid, uterus.

The role of the GP in familial cancer²¹

The GP has an important role in identifying potential high-risk patients and families and in addressing their concerns.

- Take a family history and involve at least three generations.
- Map a family tree: include any diagnosed breast, ovarian or colorectal cancers in any relative and *any* type of cancer in first- or second-degree relatives on either side of the family.
- Record the age of onset and site of cancer in first-degree relatives.
- Confirm reports of cancer from medical records.
- Assess risk (high, low or intermediate) using guidelines from your country's national cancer guidelines, e.g. the NHMRC guidelines in Australia.
- Reassure low-risk patients but provide general preventive and screening guidelines.
- Refer all patients at potentially high risk to a familial cancer clinic.

Services at these clinics involve:

- risk assessment
- genetic testing

- counselling, including pre- and post-testing
- surveillance advice

Management is based on early detection and potential prophylactic methods, for example:

- for breast cancer—regular imaging and clinical examination
- for ovarian cancer—transvaginal ultrasound and serum CA-125 detection
- for FAP and HNPCC—annual colonoscopy, faecal occult blood test

Patients at high risk will ask about prophylactic colectomy, mastectomy and oophorectomy, for which there are reasonable indications, but it is necessary to refer to a cancer geneticist for expert evaluation before such decisions are made.

Other inherited conditions

Gaucher disease

Gaucher disease, which is due to a deficiency of the lysosomal enzyme glucocerebrosidase, leads to anaemia and thrombocytopenia as a result primarily of hypersplenism. There is chronic bone pain and ‘crises’ of bone pain. Consider it in children with fatigue, bone pain, delayed growth, epistaxis, easy bruising and hepatosplenomegaly. Replacement enzyme therapy is available.

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Glycogen storage disease (liver glycogenoses)

This is a group of inherited disorders caused by a deficiency of one or more enzymes involved in glycogen breakdown, leading to the deposition of abnormal amounts of glycogen in tissues, especially the liver. The best-known type is 1A (von Gierke disorder), an autosomal recessive disorder due to deficiency of glucose-6-phosphatase (G-6-P). It is seen in several ethnic groups. It typically causes growth retardation, hepatomegaly, renomegaly, hypoglycaemia (can be severe), lactic acidosis and hyperlipidaemia. Children have characteristic morphological features—short, doll-like facies with fat cheeks, thin extremities and large abdomen (hepatomegaly).

Diagnosis is by abnormal plasma lactate and lipid levels, liver biopsy and recently by gene analysis for the G-6-P gene.

Treatment is aimed to prevent hypoglycaemia and lactic acidosis via frequent carbohydrate feedings, such as uncooked cornstarch and overnight nasogastric glucose infusion. The prognosis is poor.

The porphyrias

The three most common porphyrias are acute intermittent porphyria, porphyria cutanea tarda (the

commonest) and erythropoietic protoporphyrina, which are caused by deficiencies of the third, fifth and eighth enzymes, respectively, of the haem biosynthesis pathway. Their clinical features are quite different.

Acute intermittent porphyria

This autosomal dominant disorder is the most serious of the porphyrias although it remains clinically silent in the majority of patients who carry the trait. It is due to a deficiency of porphobilinogen (PBG) deaminase.

Clinical features

- Recurrent unexplained abdominal pain crises
- Usually young women (teens or 20s)
- Recurrent psychiatric illnesses, abnormal behaviour
- Acute peripheral or nervous system dysfunction (e.g. peripheral neuropathy, hypotonia)
- PBG in urine during attack
- Hyponatraemia
- Attacks precipitated by various drugs (e.g. anti-epileptics, alcohol, sulfonamides, barbiturates)



DxT severe abdominal pain + abnormal illness behaviour + 'red' urine → acute intermittent porphyria

Diagnosis

- Urine PBGs (high) and serum sodium (very low) during 'attack'
- Erythrocyte PBG deaminase testing to screen relatives

Treatment

- Eliminate triggers and avoid 'unsafe' drugs
- High-carbohydrate diet, glucose oral or IV for attacks
- IV haematin (haemarginate)

⌚ Tuberous sclerosis (epiloia)

This is an autosomal dominant disorder due to mutations in one of two genes located on

chromosomes 9 and 16. A feature is tube-like growths that affect multiple systems including the brain.



DxT facial rash + intellectual disability + seizures → tuberous sclerosis

The above triad of features is classic but not applicable to all cases.

Predictive genetic testing

The Human Genetics Society of Australia strongly advises against predictive or presymptomatic genetic testing of children for disease where there is no pre-emptive treatment in childhood. It should only be carried out in children if an effective treatment or preventive strategy is available. It raises issues of confidentiality, informed consent and harmful effects on self-esteem.¹⁰

Routine genetic testing is only advisable for high-risk individuals, such as those with a family history.

The ethical issues for adults are also considerable and for the individual the decision is difficult and requires considerable counselling via a clinical genetics service. This applies particularly to those at risk of Huntington disease and other adult-onset neurodegenerative conditions for which no preventive treatment exists.

Reproductive genetic screening²³

The triple test is often used for subfertility screening. It tests for cystic fibrosis, fragile X syndrome and spinal muscular atrophy. A survey by the Murdoch Children's Research Institute identified one in 20 carriers in a study of prospective parents.

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Newborn screening

With parental consent, blood from the infant's heel is usually screened for 25–45 conditions including cystic fibrosis, phenylketonuria (compare the Guthrie test), congenital hypothyroidism, galactosaemia and other rare disorders of metabolism. This test has the potential for global screening of disorders.

Prenatal screening and diagnosis of genetic disorders²⁴

Approximately 2% of births are associated with congenital abnormalities, of which 1 in 7 are chromosomal, the most common of which is Down syndrome (trisomy 21). Antenatal screening tests that can now be performed for several conditions are mainly:

- screening tests for Down syndrome and other trisomies
- screening tests for thalassaemias/haemoglobinopathies
- second-trimester ultrasound scans for fetal abnormalities, such as neural tube defects (NTD) and abdominal wall defects (AWD)

Screening for Down syndrome

This has a live birth incidence of 1.4 per 1000 in Australia.²⁰ The risk of conceiving a child with Down syndrome increases proportionally with age. For a woman aged 21, it is 1 in 1000, while for a woman aged 35, it is 1 in 275 and for a woman aged 45, it is 1 in 20.

The tests available to test for Down syndrome include the following':^{5,25,26}

1. combined first-trimester screening tests (maternal serum screening/MSST; cell-free fetal DNA at 10–12 weeks gestation; nuchal translucency ultrasound at 11–14 weeks)
2. second-trimester MSST (4 analytes): alpha fetoprotein, oestriol, free beta hCG, inhibin A. This test is basically for women presenting later in pregnancy. A final risk is calculated by a computer program which combines other factors such as EDD age and age of gestation
3. non-invasive prenatal test (NIPT) from 10–21 weeks maternal serum. This cell-free DNA screening should be offered as a choice to women. This aneuploidy test usually covers three trisomies: 21 Down syndrome, 18 Edward syndrome, 13 Patau syndrome. A follow-up ultrasound is recommended for Down syndrome. It has the potential to screen multiple disorders as it examines the genetic material of the fetus in maternal serum
4. diagnostic tests (chorionic villus sampling, amniocentesis). The most reliable method is obtaining fetal tissue by these last means but there is a significant risk of miscarriage (1 in 100 for chorionic villus sampling and 1 in 200 for amniocentesis)

Consanguinity

Consanguinity is the situation where a couple shares one or more common ancestors. Consanguineous relationships occur in most societies and in some cultures are associated with particular advantages and religious traditions.⁹

First cousins share a pair of grandparents and are statistically at an increased risk for autosomal recessive conditions. The baseline risk that any couple will have a baby with a birth defect is 3–4%.⁹ In addition, empirical data show that for first cousin marriage there is an additional 4% risk of having a child with a birth defect—this includes malformations such as intellectual impairment and many rare autosomal recessive disorders. So the combined risk is 8% irrespective of a positive family history, which is an important consideration in counselling these people.⁹

For siblings the risk of increase in birth defects is 30% and 19% for second cousins.

Rare but helpful diagnostic tips

- Dark black urine on exposure = alkaptonuria



DxT arthritis + pigmentation of ear cartilage + grey–black urine with alkalisation → alkaptonuria

- Red urine on exposure = porphyria
- Blue nappies/diapers = tryptophan malabsorption syndrome
- Maple syrup odour (urine and perspiration) = maple syrup urine disease (AR) (an amino acid metabolic disorder)



DxT odour + hypertonicity + seizures (infancy) → maple syrup urine disease

-
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- Mousy body odour = phenylketonuria
 - ‘Fish-like’ mouth with micrognathia = Turner syndrome
 - ‘Chipmunk’ facies = thalassaemia major
 - ‘Doll-like’ facies = glycogen storage disease (G-6-P deficiency)
 - Elfin face = Williams syndrome
 - Butterfly-like facial rash in children = tuberous sclerosis
 - Weak suction + delayed sitting and crawling + hypotonia = Prader–Willi syndrome
 - ‘Happy puppet’ features = Angelman syndrome
 - Characteristic eyes: wide-spaced, down-slanting openings, ptosis = Noonan syndrome
 - Long fingers and limbs = Marfan syndrome
 - High-pitched meowing cry, low-set ears, mental retardation = ‘cat-cry’ syndrome (maladie du cri du chat)
 - Blue sclera = osteogenesis imperfecta

Patient education resources

Hand-out sheets from *Murtagh's Patient Education* 8th edition:

- Autism spectrum disorder
- Cystic fibrosis
- Down syndrome
- Fragile X syndrome
- Spinal muscular atrophy
- Tourette syndrome

Resources

Centre for Genetic Education, NSW Health: www.genetics.edu.au

Cancer Council Australia, family cancer clinics helpline: 13 11 20

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Part 3 Presenting symptoms and problem solving in general practice

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24 Abdominal pain

A great fit of the stone in my left kidney: all day I could do but three or four drops of water, but I drunk a draught of white wine and salet oyle, and after that, crabs' eyes in powder with the bone in the carp's head and then drunk two great draughts of ale with buttered cake; and I voyded with an hour much water and a stone as big as an Alexander seed. God be thanked!

JOHN DEE 1594

Abdominal pain represents one of the top 15 presenting symptoms in primary care¹ and varies from a self-limiting problem to a life-threatening illness requiring immediate surgical intervention. Abdominal pain can be considered to be acute, subacute, chronic or recurrent. It can embrace all specialties, including surgery, medicine, gynaecology, paediatrics, geriatrics and psychiatry. For acute abdominal conditions it is important to make a rapid diagnosis in order to reduce morbidity and mortality. Most cases of ‘acute abdomen’ require surgical referral. Lower abdominal pain in women adds another dimension to the problem and will be presented in a separate chapter ([CHAPTER 95](#)).

Key facts and checkpoints

- The commonest causes of the acute abdomen in a general practice series were: acute appendicitis (21%), the colics (16%) and mesenteric adenitis (16%).²
- An international study involving referral to 26 surgical departments in 17 countries revealed the most common conditions to be: non-specific abdominal pain (34%), acute appendicitis (28%) and cholecystitis (10%).¹
- As a general rule, upper abdominal pain is caused by lesions of the upper GIT and lower abdominal pain by lesions of the lower GIT.

- Colicky midline umbilical abdominal pain (severe) → vomiting → distension = small bowel obstruction (SBO).
- Midline lower abdominal pain → distension → vomiting = large bowel obstruction (LBO).
- If cases of acute abdomen have a surgical cause, the pain nearly always precedes the vomiting (contrast with gastroenteritis)
- Mesenteric artery occlusion must be considered in an older person with arteriosclerotic disease or in those with atrial fibrillation presenting with severe abdominal pain or following myocardial infarction.
- Up to one-third of presentations of abdominal pain have no specific cause found.

A diagnostic approach

A summary of the separate diagnostic models for acute abdominal pain and chronic abdominal pain are presented in TABLES 24.1 and 24.2 .

Table 24.1 Acute abdominal pain (adults): diagnostic strategy model (excluding trauma)

Probability diagnosis

- Acute gastroenteritis
- Acute appendicitis
- Mittelschmerz/dysmenorrhoea
- Irritable bowel syndrome
- Biliary colic/renal colic

Serious disorders not to be missed

Cardiovascular:

- myocardial infarction (usually inferior)
- ruptured AAA
- dissecting aneurysm of aorta
- mesenteric artery occlusion/ischaemia

Neoplasia:

- large or small bowel obstruction

Severe infections:

- acute salpingitis

- peritonitis/spontaneous bacterial peritonitis
- ascending cholangitis
- intra-abdominal abscess

Pancreatitis

Ectopic pregnancy

Small bowel obstruction/strangulated hernia

Sigmoid volvulus

Perforated viscus

Pitfalls (often missed)

Acute appendicitis

Muscle/myofascial tear

Pulmonary causes:

- pneumonia
- pulmonary embolism

Faecal impaction (elderly)

Herpes zoster

Rarities:

Porphyria

Epiploic appendagitis

Lead poisoning

Henoch–Schönlein purpura

Haemochromatosis

Haemoglobinuria

Addison disease

Seven masquerades checklist

Depression

Diabetes (ketoacidosis)

Drugs (esp. narcotics)

Anaemia (sickle cell)

Endocrine disorder (thyroid storm, Addison)

Spinal dysfunction → referred pain

UTI (including urosepsis)

Is the patient trying to tell me something?

May be very significant.

Consider Munchausen syndrome, sexual dysfunction and abnormal stress/anxiety.

Table 24.2 Chronic or recurrent abdominal pain (adult): diagnostic strategy model**Probability diagnosis**

Irritable bowel syndrome
 Diverticular disease
 Mittelschmerz/dysmenorrhoea
 Peptic ulcer/gastritis

Serious disorders not to be missed

Cardiovascular:

- mesenteric artery ischaemia
- AAA

Neoplasia:

- bowel/stomach cancer
- pancreatic cancer
- ovarian tumours

Severe infections:

- hepatitis
- recurrent PID

Pitfalls (often missed)

Adhesions
 Appendicitis
 Food allergies
 Lactase deficiency
 Constipation/faecal impaction
 Chronic pancreatitis
 Crohn disease
 Endometriosis
 Diverticulitis

Rarities:

Tropical infections (e.g. hydatids, melioidosis, malaria, strongyloides)
 Uraemia
 Lead poisoning
 Porphyria
 Sickle-cell anaemia
 Hypercalcaemia

Addison disease

Seven masquerades checklist

Depression

Drugs

Endocrine disorder (Addison disease)

Spinal dysfunction

UTI

Is the patient trying to tell me something?

A strong possibility: consider hypochondriasis, anxiety, sexual dysfunction, Munchausen syndrome.

Probability diagnosis

Common reasons are common: gastroenteritis/food poisoning accounts for so many GP presentations that occasionally a case will have examination findings of an acute abdomen. The other most common causes of acute abdomen are acute appendicitis, irritable bowel syndrome, the various ‘colics’ and ovulation pain (mittelschmerz). Mesenteric adenitis is common in children. The various causes of chronic or recurrent abdominal pain are presented in

[TABLE 24.2](#) . A study on chronic abdominal pain³ showed that the commonest reasons (approximate percentages) were no discoverable causes (50%), minor causes including muscle strains (16%), irritable bowel syndrome (12%), gynaecological causes (8%), peptic ulcers and hiatus hernia (8%).

Serious disorders not to be missed

Most of the causes of the acute abdomen are serious and early diagnosis is mandatory to reduce mortality and morbidity.

It is vital not to misdiagnose a ruptured ectopic pregnancy, which causes lower abdominal or suprapubic pain of sudden onset, or the life-threatening vascular causes, such as a ruptured or dissecting aortic aneurysm, mesenteric artery occlusion and myocardial infarction (which can present as epigastric pain).

Perforated ulcers (now uncommon) and strangulated bowel, such as volvulus of the sigmoid and entrapment of the small bowel in a hernial orifice or around adhesions, also demand an early diagnosis.

There are some important ‘red flag’ symptoms and signs¹ of abdominal emergencies demanding urgent attention (see box).

Red flag pointers for acute abdominal pain

History	Signs
Collapse at toilet (intra-abdominal bleeding)	Pallor and sweating
Lightheadedness	Hypotension
Ischaemic heart disease	Atrial fibrillation or tachycardia
Progressive vomiting, pain, distension	Fever
Menstrual abnormalities	Prostration
Malignancy	Rebound tenderness and guarding
Lack of flatus	Decreased urine output

Dangers of misdiagnosis

- Ectopic pregnancy → rapid hypovolaemic shock
- Ruptured AAA → rapid hypovolaemic shock
- Gangrenous appendix → peritonitis/pelvic abscess
- Perforated ulcer → peritonitis
- Obstructed bowel → gangrene

Pitfalls

A very common pitfall is missing acute appendicitis, especially in the elderly, in children, in pregnancy and in those taking steroids, where the presentation may be atypical. Early appendicitis presents typically with central abdominal pain that shifts to the right iliac fossa (RIF) some 4–6 hours later. It can be difficult to diagnose early on. It can cause diarrhoea with abdominal pain, especially if a pelvic appendix, and can be misdiagnosed as acute gastroenteritis.

Disaccharidase deficiencies, such as lactase deficiency, are associated with cramping abdominal pain, which may be severe. The pain follows some time, maybe hours, after the ingestion of milk and is accompanied by the passage of watery stool. The association with milk may go unrecognised.

Herpes zoster, especially in the older person with unilateral abdominal pain in the dermatomal distribution, is a trap. Referred pain from conditions above the diaphragm, such as myocardial

infarction, pulmonary embolism and pneumonia, can be misleading. The rare general medical causes—such as diabetes ketoacidosis, acute porphyria, Addison disease, lead poisoning, tabes dorsalis, sickle-cell anaemia, haemochromatosis and uraemia—often create a diagnostic dilemma.

Specific pitfalls

- Misdiagnosing a ruptured ectopic pregnancy in a woman using contraception or with a history of normal menstruation or where the brownish vaginal discharge is mistaken for a normal period.
- Failing to examine hernial orifices in a patient with intestinal obstruction.
- Misleading temporary improvement (easing of pain) in perforation of gangrenous appendix or perforated peptic ulcer.
- Overlooking acute mesenteric artery obstruction in an older person with colicky central abdominal pain.
- Attributing abdominal pain, frequency and dysuria to a urinary infection when the cause could be diverticulitis, pelvic appendicitis, salpingitis or a ruptured ectopic pregnancy.
- Failing to examine testes.

Seven masquerades checklist

Depression, diabetes, drugs, spinal dysfunction and UTI can all cause abdominal pain: acute, subacute or chronic. Abdominal pain and even tenderness can accompany diabetic ketoacidosis. Drugs that can cause abdominal pain are listed in TABLE 24.3 .

Table 24.3 Drugs to consider as a cause of abdominal pain

Many illicit drugs
Alcohol
Antibiotics (e.g. erythromycin)
Aspirin
Corticosteroids
Cytotoxic agents
Tricyclic antidepressants (e.g. imipramine)
Iron preparations
Nicotine
NSAIDs/COX-2 inhibitors

Sodium valproate

Phenytoin

Spinal dysfunction of the lower thoracic spine and thoracolumbar junction can cause referred pain to the abdomen. The pain is invariably unilateral, radicular in distribution and related to activity. It can be confused with intra-abdominal problems such as biliary disease (right-sided), appendicitis and Crohn disease (right side), diverticular disorder (left-sided) and pyelonephritis.

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Psychogenic considerations

Psychogenic factors can be most relevant, especially in recurrent or chronic abdominal pain where no specific cause can be identified. Certainly consider psychosocial issues in adolescents and young adults presenting with recurrent pain.

Munchausen syndrome is hospital admission by deception, often with severe abdominal pain without convincing clinical signs or abnormal investigation. Diagnosis requires a high level of suspicion.

The clinical approach

History

The urgency of the history will depend on the manner of presentation, whether acute or chronic. Pain has to be analysed according to its quality, quantity, site and radiation, onset, duration and offset, aggravating and relieving factors and associated symptoms and signs.

Special attention has to be paid to:

- anorexia, nausea or vomiting
- micturition
- bowel function
- menstruation/contraception
- drug intake

Key questions

Point to where the pain is and where it travels to.

Questions to ask:

- What type of pain is it: is it constant or does it come and go?
- How severe would you rate it from 1 to 10?
- Have you ever had previous attacks of similar pain?
- What else do you notice when you have the pain?
- Do you know of anything that will bring on the pain? Or relieve it?
- What effect does milk, food or antacids have on the pain?
- Have you noticed any sweats or chills or burning of urine?
- Are your bowels behaving normally? Have you been constipated or had diarrhoea or blood in your motions?
- Have you noticed anything different about your urine?
- What medications do you take?
- How much aspirin do you take?
- Are you smoking or drinking heavily or taking illicit drugs?
- Have you travelled recently?
- What is happening with your periods? Is it mid-cycle or are your periods overdue?
- Does anyone in your family have bouts of abdominal pain?
- Do you have a hernia?
- What operations have you had for your abdomen? Appendix, gall bladder?

Examination

A useful checklist for conducting the examination is:

- general appearance
- oral cavity
- vital parameters: temperature, pulse, BP, respiratory rate
- chest: check heart and lungs for upper abdominal pain (especially if absent abdominal signs)
- abdomen: inspection, auscultation, palpation and percussion (in that order)

Abdominal examination should be performed with the patient lying flat with one pillow under the head and the abdomen uncovered from xiphisternum to groin. Consider the following:

- inguinal region (including hernial orifices) and femoral arteries
- rectal examination
- vaginal examination (females): for suspected problems of the fallopian tubes, uterus or ovaries
- thoracolumbar spine (if referred spinal pain suspected)
- urine analysis: white cells, red cells, glucose and ketones, porphyrins
- special clinical tests: Murphy sign (a sign of peritoneal tenderness with acute cholecystitis); psoas and obturator signs
- look for hernias: Spigelian hernias occur through defects in transversus abdominal muscle lateral to the rectus sheath—usually below the level of the umbilicus

Guidelines

- *Palpation*: start away from the painful side, palpate with gentleness—note any guarding or rebound tenderness: guarding indicates peritonitis; rebound tenderness indicates peritoneal irritation (bacterial peritonitis, blood). Feel for maximum site that corresponds to focus of the problem
- *Patient pain indicator*: the finger pointing sign indicates focal peritoneal irritation; the spread palm sign indicates visceral pain
- *Atrial fibrillation*: consider mesenteric artery obstruction
- *Tachycardia*: sepsis and volume depletion
- *Tachypnoea*: sepsis, pneumonia, acidosis
- *Pallor and ‘shock’*: acute blood loss
- *Auscultation*: note bowel activity or a succussion splash (best before palpation and percussion)

Causes of a ‘silent abdomen’: diffuse sepsis, ileus, mechanical obstruction (advanced).

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If distension, consider the six Fs: fat, fluid, flatus, faeces, fetus, frightening growths.

Hypertympany indicates mechanical obstruction.

Physical signs may be reduced in the elderly, grossly obese, severely ill and those using corticosteroids.

Investigations

The following investigations may be selected if indicated:

- haemoglobin—anaemia with chronic blood loss (e.g. peptic ulcer, cancer, oesophagitis)
- blood film—abnormal red cells with sickle-cell disease
- WCC—leucocytosis with appendicitis (75%),⁴ acute pancreatitis, mesenteric adenitis (first day only), cholecystitis (especially with empyema), pyelonephritis
- ESR—raised with cancer, Crohn disease, abscess (but non-specific)
- C-reactive protein (CRP)—use in diagnosing and monitoring infection, inflammation (e.g. pancreatic). Preferable to ESR
- liver function tests—hepatobiliary disorder
- serum amylase and/or lipase (preferable)—if raised to greater than three times normal upper level acute pancreatitis is most likely; also raised partially with most intra-abdominal disasters (e.g. ruptured ectopic pregnancy, perforated peptic ulcers, ruptured empyema of gall bladder, ruptured aortic aneurysm)
- faecal elastase—chronic pancreatitis
- pregnancy tests—urine or serum β-HCG: for suspected ectopic
- *Helicobacter pylori* testing
- urine:
 - blood: ureteric colic (stone or blood clot), urinary infection
 - white cells: urinary infection, appendicitis (bladder irritation)
 - bile pigments: gall bladder disease
 - porphobilinogen: porphyria (add Ehrlich aldehyde reagent)
 - ketones: diabetic ketoacidosis
 - air (pneumaturia): fistula (e.g. diverticulitis, other pelvic abscess, pelvic cancer)
- faecal blood—mesenteric artery occlusion, intussusception ('redcurrant jelly'), colorectal cancer, diverticulitis, Crohn disease and ulcerative colitis

Radiology

The two main screening tests are ultrasound and CT scan.⁵ Plain abdominal X-ray is an alternative, if more readily available. The following tests can be considered according to the clinical presentation:

- ultrasound: good for hepatobiliary system, kidneys and female pelvis. Look for:

- gallstones
- ectopic pregnancy
- pancreatic pseudocyst
- aneurysm aorta/dissecting aneurysm
- hepatic metastases and abdominal tumours
- thickened appendix
- paracolic collection

Note: can be affected by gas shadows

- CT scan: gives excellent survey of abdominal organs including masses and fluid collection:

- pancreatitis (acute and chronic)
- undiagnosed peritoneal inflammation (best)
- trauma
- diverticulitis
- leaking aortic aneurysm
- retroperitoneal pathology
- appendicitis (especially with oral contrast)

- plain X-ray abdomen (erect and supine): look for (see FIG. 24.1):

- kidney/ureteric stones—70% opaque⁴
- biliary stones—only 10–30% opaque
- air in biliary tree
- calcified aortic aneurysm
- marked distension sigmoid → sigmoid volvulus

distended bowel with fluid level → bowel obstruction

enlarged caecum with large bowel obstruction

blurred right psoas shadow → appendicitis

‘coffee bean’ sign → volvulus

a sentinel loop of gas in left upper quadrant (LUQ) → acute pancreatitis

- chest X-ray: air under diaphragm → perforated ulcer
- IVP
- contrast-enhanced CT or X-ray (e.g. Gastrograffin meal): diagnosis of bowel leakage
- barium enema
- HIDA nuclear scan—diagnosis of acute cholecystitis (good when US unhelpful)
- ERCP: shows bile duct obstruction and pancreatic disease
- MRI scan (especially useful with contrast)

Other tests:

- ECG
- endoscopy upper GIT
- sigmoidoscopy and colonoscopy

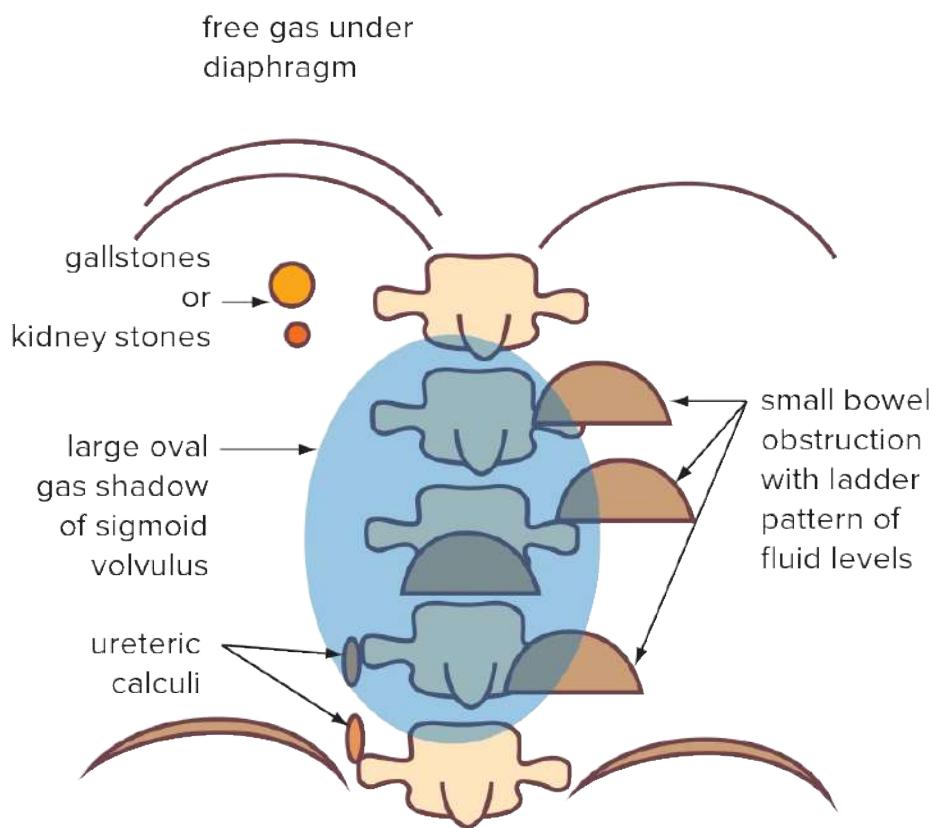


FIGURE 24.1 The acute abdomen: signs to watch for on plain abdominal X-ray

Diagnostic guidelines

General rules

- Upper abdominal pain is caused by lesions of the upper GIT.
- Lower abdominal pain is caused by lesions of the lower GIT or pelvic organs.
- Early severe vomiting indicates a high obstruction of the GIT.
- Acute appendicitis features a characteristic ‘march’ of symptoms: pain → anorexia, nausea → vomiting.

Pain patterns

The pain patterns are presented in [FIGURE 24.2](#). Colicky pain is a rhythmic pain with regular spasms of recurring pain building to a climax and fading. It is virtually pathognomonic of intestinal obstruction. Ureteric colic is a true colicky abdominal pain, but so-called biliary colic

and kidney colic are not true colics at all.

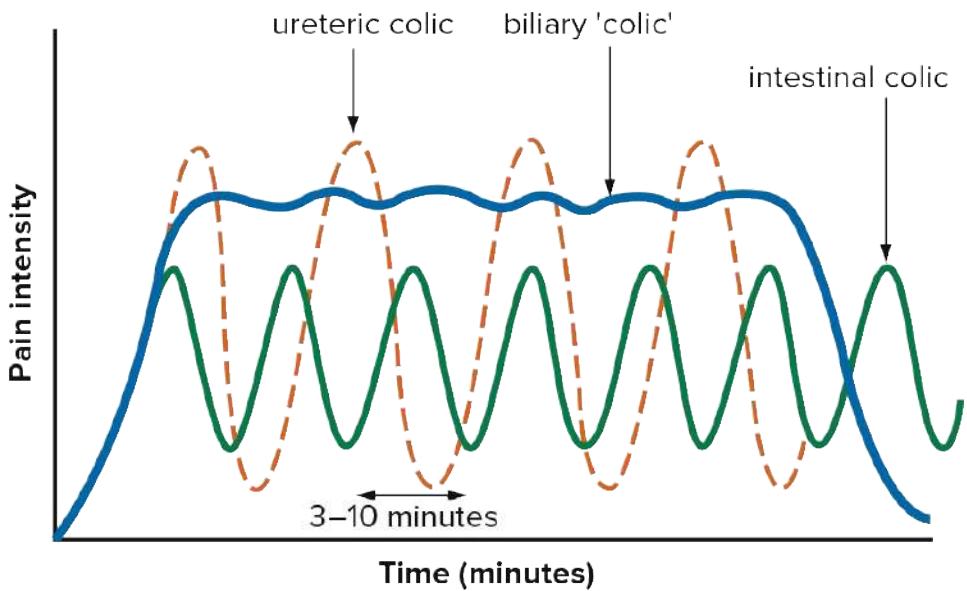


FIGURE 24.2 Characteristic pain patterns for various causes of ‘colicky’ acute abdominal pain

Site of pain

Typical pain sites of abdominal pain (general guidelines only) are presented in [FIGURE 24.3](#). Epigastric pain usually arises from disorders of the embryologic foregut, such as the oesophagus, stomach and duodenum, hepatobiliary structures, pancreas and spleen. However, as some disorders progress the pain tends to shift from the midline to the right (gall bladder and liver) or left (spleen). Perumbilical pain usually arises from disorders of structures of the embryologic midgut, while structures from the hindgut tend to refer pain to the lower abdomen or suprapubic region.

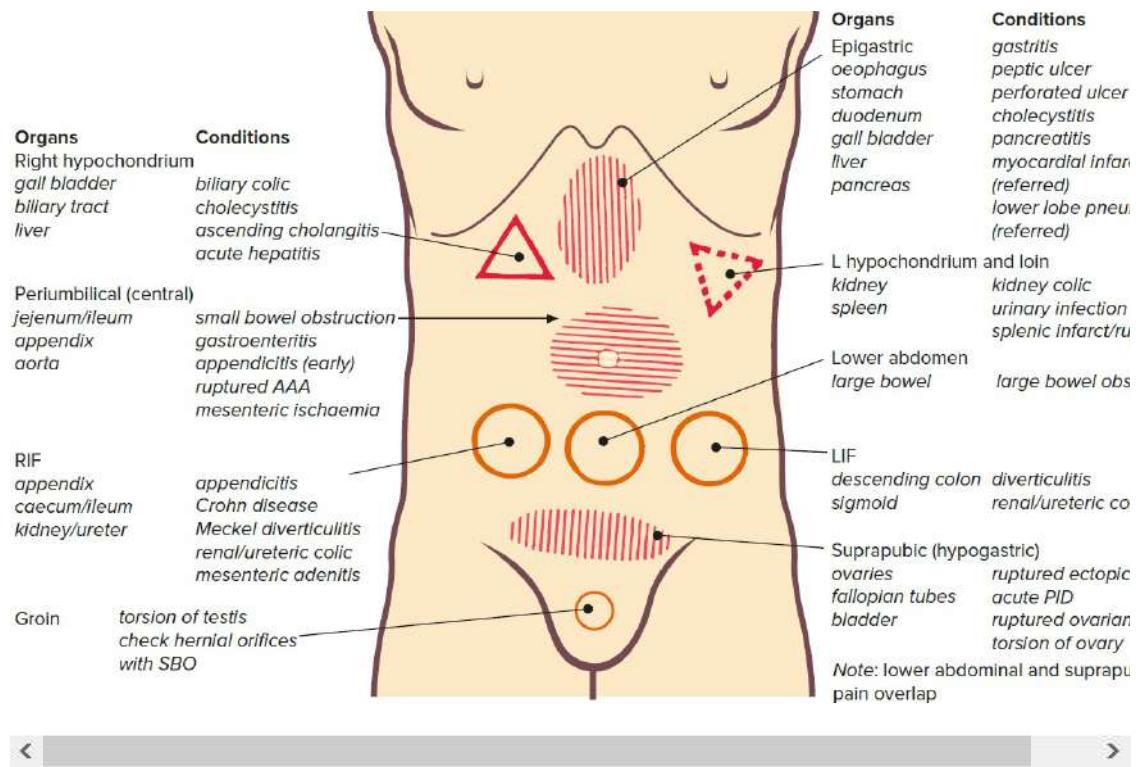


FIGURE 24.3 Typical sites of various causes of acute abdominal pain

The intra-abdominal sensory receptors can be considered as innervating visceral or parietal peritoneum. Visceral mechanoreceptors are triggered by intestinal distension or tension on mesentery or blood vessels while nociceptors are triggered by mechanical, thermal and chemical stimuli. The pain from viscera is felt as diffuse and poorly localised, while stimulation of parietal peritoneal nociceptors gives a pain that is experienced directly at the site of insult.

Abdominal pain in children

Abdominal pain is a common complaint in children, especially recurrent abdominal pain, which is one of the most common complaints in childhood. The problem causes considerable anxiety in parents and it is important to differentiate the severe problems demanding surgery from non-surgical problems. About one in 15 will have a surgical cause for pain.⁶ A good rule is to rule out a urinary infection with urinalysis.

Table 24.4 Acute abdominal pain in children

The causes of abdominal pain can be considered in the diagnostic model category.

- 1 Common causes/probability diagnosis:
 - infant 'colic'

- gastroenteritis (all ages)
 - mesenteric adenitis
-

2 Serious causes, not to be missed:

- intussusception (peaks at 6–9 months)
 - acute appendicitis (mainly 5–15 years)
 - bowel obstruction/strangulated hernia
-

3 Pitfalls:

- child abuse
 - constipation/faecal impaction
 - torsion of testes
 - lactose intolerance
 - peptic ulcer
 - infections: mumps, tonsillitis, pneumonia (esp. right lower lobe), EBM, UTI
 - adnexal disorders in females (e.g. ovarian)
 - acute pancreatitis
-

4 Rarities:

- Meckel diverticulitis
 - Henoch–Schönlein purpura
 - sickle crisis
 - lead poisoning
-

5 Seven masquerades checklist:

- type 1 diabetes
 - drugs
 - UTI
-

6 Psychogenic consideration:

- important cause
-

⌚ Infant ‘colic’ (period of infant distress)

This is the occurrence in a well baby of regular, unexplained periods of inconsolable crying and fretfulness, usually in the late afternoon and evening, especially between 2 weeks and 16 weeks of age. No apparent cause can be found, and the word ‘colic’ refers to the historical assumption that the crying is caused by abdominal pain. It is very common, occurring in about one-third of infants and lasting for a period of at least three weeks.

Clinical features

- Baby between 2 and 16 weeks old
- Prolonged crying—at least 3 hours
- Occurrence at least 3 days a week
- Crying worst at around 10 weeks of age
- Crying during late afternoon and early evening
- Child flexing legs and clenching fists as if severe ‘stomach ache’
- Normal physical examination

Management

Reassure and explain. Advise the parents:

- Use gentleness (such as subdued lighting where the baby is handled, soft music, speaking softly, quiet feeding times). Page 261
- Avoid quick movements that may startle the baby.
- Make sure the baby is not hungry—avoid underfeeding.
- Provide demand feeding (in time and amount).
- Make sure the baby is burped, and give posture feeding.
- Provide comfort from a dummy or pacifier.
- Provide plenty of gentle physical contact.
- Cuddle and carry the baby around (e.g. take a walk around the block).
- A carrying device such as ‘snuggly’ or ‘Mei Tai Sling’ allows the baby to be carried around at the time of crying.
- Make sure the mother gets plenty of rest during this difficult period.
- Do not worry about leaving a crying child for 10 minutes or so after 15 minutes of trying consolation.

Medication

Drugs are not generally recommended, but some preparations have tradition, if not much science, behind them (e.g. simethicone [Infacol wind drops]).

⌚ Intussusception

Intussusception is the diagnosis that should be foremost in one's mind with a child aged between 3 months and 2 years presenting with sudden onset of severe colicky abdominal pain, coming at intervals of about 15 minutes and lasting for 2–3 minutes. Early diagnosis, within 24 hours of onset, is essential, for after this time there is a significant rise in morbidity and mortality. A segment of bowel telescopes into the adjoining distal segment (e.g. ileocaecal segment), resulting in intestinal obstruction. It is usually idiopathic but can have a pathological lead point (4–12 years) (e.g. polyp, Meckel diverticulum).

Typical clinical features⁷

See FIGURE 24.4 .

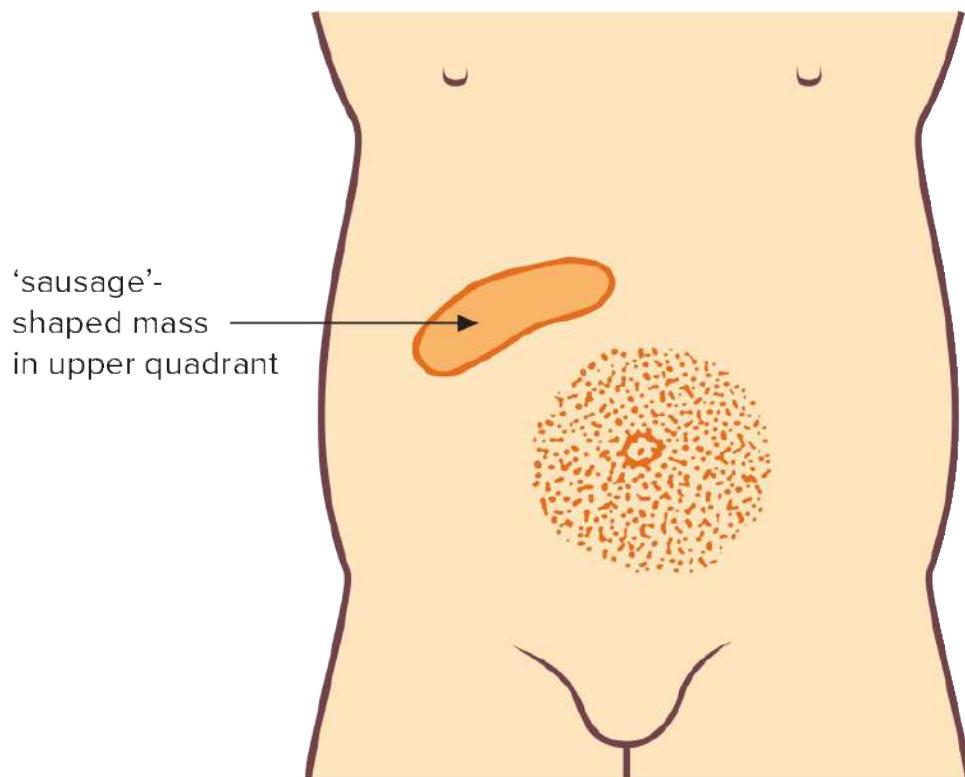


FIGURE 24.4 Typical features with pain distribution of acute intussusception

- Male babies > female
- Range: birth to school age, usually 5–24 months
- Sudden-onset acute pain with shrill cry
- Vomiting

- Lethargy
- Pallor with attacks
- Intestinal bleeding: redcurrant jelly (60%)⁷



DxT pale child + severe 'colic' + vomiting → acute intussusception

Signs

- Pale, anxious and unwell
- Sausage-shaped mass in right upper quadrant (RUQ) anywhere between the line of colon and umbilicus, especially during attacks (difficult to feel)
- Signe de dance (i.e. emptiness in RIF to palpation)
- Alternating high-pitched active bowel sounds with absent sounds
- Rectal examination: ± blood ± hard lump

Diagnosis

- Ultrasound
- Enema using oxygen or barium (with caution) used for diagnosis and treatment

Treatment⁷

- Hydrostatic reduction by air or oxygen from the 'wall' supply (preferred) or barium enema
- Surgical intervention may be necessary

Differential diagnosis

- Acute gastroenteritis: can be difficult in those cases where there is some loose stool with intussusception and with blood and mucus without much watery stool in gastroenteritis. However, usually attacks of pain are of shorter duration, and there is loose watery stool, fever and no abdominal mass. If doubtful, refer as possible intussusception.
- Impacted faeces can lead to spasms of colicky abdominal pain—usually an older child with a history of constipation.
- Other causes of intestinal obstruction (e.g. irreducible inguinal hernia, volvulus, intra-abdominal band).

Drugs

In any child complaining of acute abdominal pain, enquiry should be made into drug ingestion. A common cause of colicky abdominal pain in children is cigarette smoking (nicotine); consider other drugs such as marijuana, cocaine and heroin.

§ Acute appendicitis in children

This may occur at any age, being more common in children of school age (10–12 years) and in adolescence, and uncommon in children under 3 years of age. Special problems of early diagnosis occur with the very young (younger than 3 years) and in intellectually disabled children, many of whom present with peritonitis.

Vomiting occurs in at least 80% of children with appendicitis and diarrhoea in about 20%. The temperature is usually only slightly elevated but in about 5% of cases it exceeds 39°C.⁴

In children the physical examination, especially eliciting abdominal (including rebound) tenderness, and the rectal examination demand considerable tact, patience and gentleness. Jumping or hopping induces pain.

A serious point of confusion can occur between pelvic appendicitis, causing diarrhoea and vomiting, and acute gastroenteritis. A high CRP level >50 mg/L is a feature of appendicitis.⁶ A particularly severe case of apparent gastroenteritis, especially if persistent, should be regarded as pelvic appendicitis until proved otherwise. Ultrasound is the preferred imaging.

§ Mesenteric adenitis

This presents a difficult problem in differential diagnosis with acute appendicitis because the history can be very similar. At times the distinction may be almost impossible. In general, with mesenteric adenitis localisation of pain and tenderness is not as definite, rigidity is less of a feature, the temperature is higher, and anorexia, nausea and vomiting are also lesser features. The illness lasts about five days followed by a rapid recovery. Comparisons between the two are presented in TABLE 24.5, but if in any doubt it is advisable to consider the problem as acute appendicitis, admit for observation and be prepared for laparoscopy/laparotomy.

Table 24.5 Comparison of the features of acute appendicitis and mesenteric adenitis in children (guidelines only)

	Acute appendicitis	Mesenteric adenitis
Typical child	Older	Younger
Site of onset of pain	Midline Shifting to	RIF Can be midline

	right	
Preceding respiratory illness	Uncommon	Invariable: URTI or tonsillitis
Anorexia, nausea, vomiting	++	±
Colour	Usually pale	Flushed: malar flush
Temperature	N or ↑	↑ ↑ → ↑ ↑ ↑
Abdominal palpation	Tender in RIF Guarding ± Rigidity	Tender in RIF Minimal guarding Usually no rigidity
Rectal examination	Invariably tender	Often tender but lesser degree
Psoas and obturator tests	Usually positive	Usually negative
Full blood examination	Leucocytosis	Lymphocytosis

RIF = right iliac fossa

Mesenteric adenitis can sometimes present an anaesthetic risk and patients are usually quite ill in the immediate postoperative period. Treatment is symptomatic and includes ample fluids and paracetamol.

Recurrent abdominal pain

Recurrent abdominal pain (RAP)—three distinct episodes of abdominal pain over 3 or more months—occurs in 10% of school-aged children. In only 5–10% of children will an organic cause be found so that in most cases the cause remains obscure.⁸

Causes (organic)

An organic cause, however, must be considered and excluded. Organic disease is more likely if:

- the pain is other than perumbilical
- the pain radiates rather than remains localised
- the pain wakes the child from sleep
- the pain is accompanied by nausea and vomiting
- the child is not completely well between attacks
- there is associated weight loss or failure to thrive

Possible causes

- Constipation
- Childhood migraine equivalent (pain with extreme pallor)
- Lactose intolerance (symptoms related to milk ingestion)
- Intestinal parasites (may disturb child about 60 minutes after falling asleep)

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Investigations

- Urine analysis and MSU
- FBE and ESR
- Plain X-ray (assesses faecal retention)

Non-organic RAP

Clinical features

Typical clinical features include:

- acute and frequent colicky abdominal pain
- pain localised to or just above umbilicus
- no radiation of pain
- pain lasts less than 60 minutes
- nausea and frequent vomiting are usually absent
- diurnal (never wakes the child at night)
- minimal umbilical tenderness
- anxious child
- obsessive or perfectionist personality
- one or both parents intense about child's health and progress

Psychogenic factors

Although psychogenic factors are very relevant in individual cases there is scant hard evidence to

support the widely held hypothesis⁸ that such factors account for the vast majority of RAP. Some children will have obvious psychological problems or even be school avoidant, a common factor being family disruption.

Management⁸

- Give explanation, reassurance and support (ensure that the patient is involved in the discussion).
- Reassurance can only be given following a careful examination and thoughtfully chosen investigations.
- Avoid investigations, especially radiological if possible (FBE and MCU are okay).
- Acknowledge that the child has pain.
- Emphasise that the disorder is common, and usually traverses childhood without ill effects.
- Recommend simple measures (e.g. local warmth, brief rest for painful episodes).
- Advise review if episodes change in nature, pain persists for hours or there are new symptoms.
- Identify any life stresses and provide insight therapy.
- Enquire about family structures and function, and school performance.
- Discourage identification with the sick role.
- Refer for psychological assessment and counselling if necessary.

Abdominal pain in older people

Older people can suffer from a wide spectrum of disorders. Ischaemic events, emboli, cancer (in particular) and diverticulae of the colon are more common in old age; duodenal ulcer is less so. Those causes of abdominal pain that occur with more frequency include:

- vascular catastrophies: ruptured AAA, mesenteric artery occlusion
- perforated peptic ulcer
- biliary disorders: biliary pain and acute cholecystitis
- diverticulitis
- sigmoid volvulus
- strangulated hernia

- intestinal obstruction
- cancer, especially of the large bowel
- herpes zoster, causing unilateral root pain
- constipation and faecal impaction

Problems arise with management because the pain threshold is raised (colic in particular is less severe) and there is an attenuated response to infection so that fever and leucocytosis can be absent. Non-specific signs, such as confusion, anorexia and tachycardia, might be the only systemic evidence of infection.

Abdominal aortic aneurysm

An AAA may be asymptomatic until it ruptures or may present with abdominal discomfort and a pulsatile mass noted by the patient. There tends to be a family history and thus screening is appropriate in such families. Ultrasound screening is advisable in first-degree relatives over 50 years.

The risk of rupture is related to the diameter of the AAA and the rate of increase in diameter. The normal diameter of the abdominal aorta, which is palpated just above the umbilicus, is 10–30 mm, being 20 mm on average in the adult; an aneurysm is greater than 30 mm in diameter.⁹ Refer if ≥ 40 mm. Greater than 50 mm is significantly enlarged and is chosen as the arbitrary reference point to operate because of the exponential rise in risk of rupture with an increasing diameter. Refer all cases. The patency of a Dacron graft after 5 years is approximately 95% (see FIG. 24.5). Newer techniques include endovascular aneurysm repair.

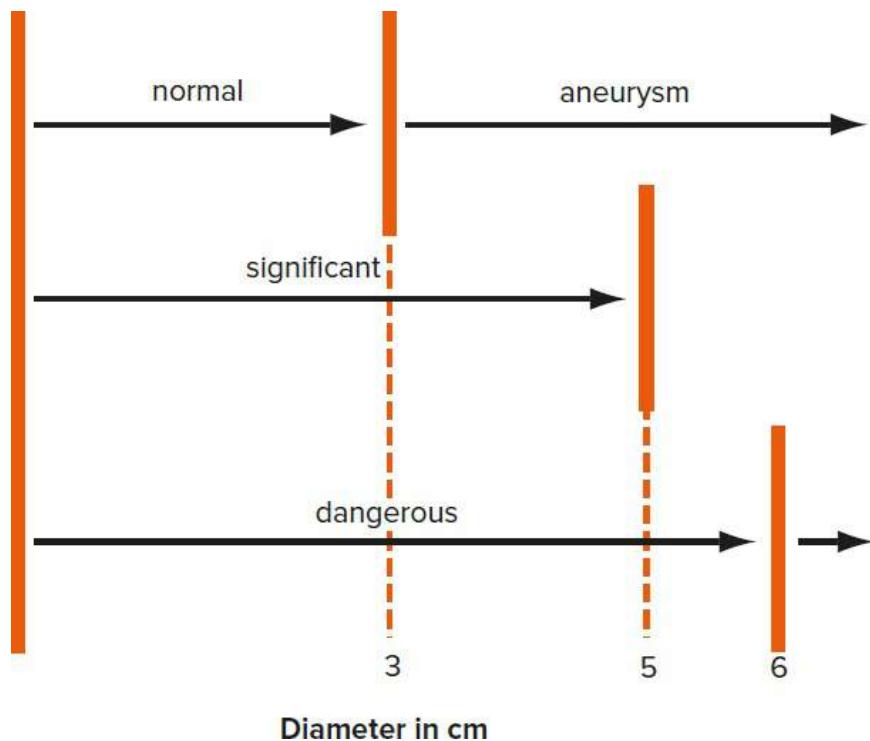


FIGURE 24.5 Guidelines for normal and abnormal widths of the abdominal aorta in adults (to exact scale)

Investigations

- Ultrasound (good for screening) in relatives >50 years (obesity a problem)
- CT scan (clearer imaging). Helical/spiral scan is investigation of choice
- MRI scan (best definition)

⌚ Rupture of aneurysm

This is a real surgical emergency in an elderly person who presents with acute abdominal and perhaps back pain with associated circulatory collapse (see FIG. 24.6). The patient often collapses at toilet because they feel the need to defecate and the resultant Valsalva manoeuvre causes circulatory embarrassment.

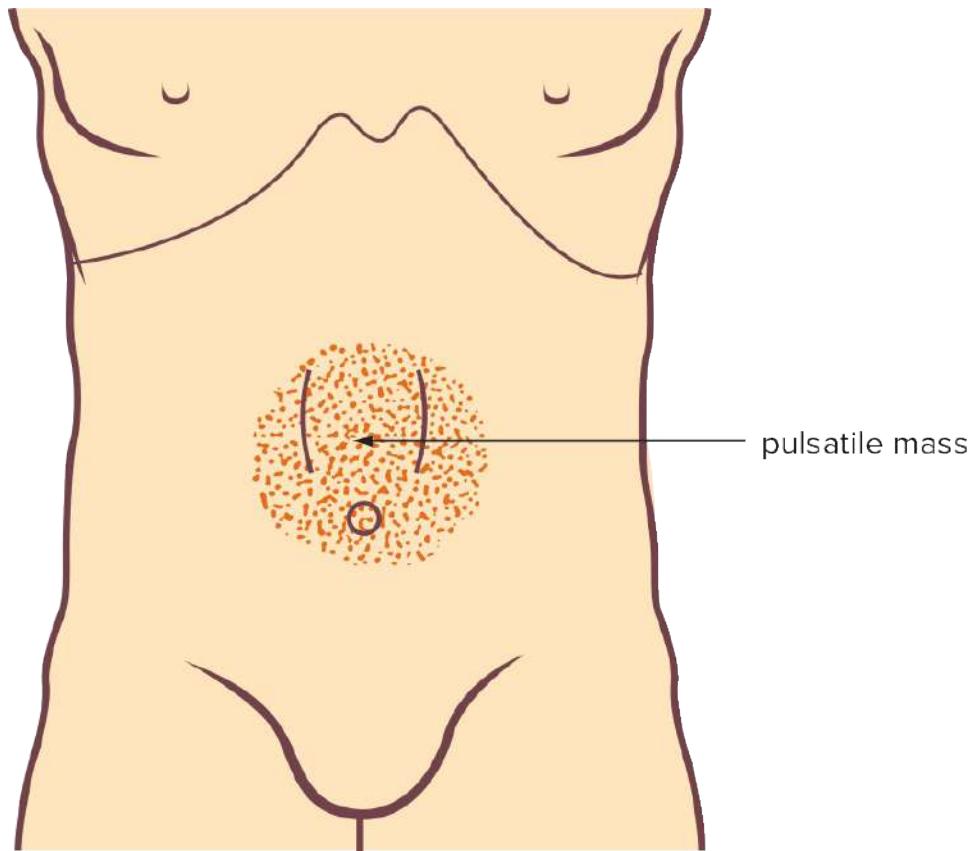


FIGURE 24.6 Typical pain distribution of a ruptured abdominal aortic aneurysm

The patient should be transferred immediately to a vascular surgical unit, which should be notified in advance. Two important emergency measures for the ‘shocked’ patient are intravenous access for plasma expanding fluid (a central venous line is best if possible) and swift action.



DxT intense abdominal pain + pale and ‘shocked’ ± back pain → ruptured AAA

Mesenteric artery occlusion

Acute intestinal ischaemia arises from superior mesenteric artery occlusion from either an embolus or a thrombosis in an atherosclerotic artery. Another cause is an embolus from atrial fibrillation. Necrosis of the intestine soon follows if intervention is delayed.

Clinical features

- Central perumbilical abdominal pain—gradually becomes intense. Patients develop a ‘fear of

eating'

- Profuse vomiting
- Watery diarrhoea—blood in one-third of cases (eventually) (refer to CHAPTER 34)
- Patient becomes confused



DxT anxiety and prostration + intense central pain + profuse vomiting ± bloody diarrhoea → mesenteric arterial occlusion

Signs

- Localised tenderness, rigidity and rebound over infarcted bowel (later finding)
- Absent bowel sounds (later)
- Shock develops later
- Tachycardia (may be atrial fibrillation and other signs of atheroma)

Investigations

- CRP may be elevated intestinal alkaline phosphatase.
- X-ray (plain) shows ‘thumb printing’ due to mucosal oedema on gas-filled bowel. CT scanning gives the best definition while mesenteric arteriography is performed if embolus is suspected. However, it is commonly only diagnosed at laparotomy.

Management

Early surgery may prevent gut necrosis but massive resection of necrosed gut may be required as a life-saving procedure. Early diagnosis (within a few hours) is essential.

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Note:

- Mesenteric venous thrombosis can occur but usually in people with circulatory failure.
- Inferior mesenteric artery occlusion is less severe and survival more likely.

⌚ Acute retention of urine

Acute retention of urine with a volume of 600+ mL usually causes severe lower abdominal pain, which may not be apparent in a senile or dementing person. Find and treat the cause. Apart from the common cause of an enlarged prostate or prostatitis, it can also result from bladder neck obstruction by faecal loading or other pelvic masses or anticholinergic drugs. It is often

precipitated by extreme cold or an excess of alcohol. Neurogenic causes include multiple sclerosis, spinal injury and diabetes.

Management

- Perform a rectal examination and empty rectum of any impacted faecal material.
- Catheterise with size 14 Foley catheter to relieve obstruction and drain (give antibiotic cover).
- Have the catheter in situ and seek a urological opinion. Send specimen for MCU.
- If there is any chance of recovery (e.g. if the problem is drug-induced), withdraw drug, leave catheter in for 48 hours, remove and give trial of prazosin 0.5 mg bd or terazosin.
- In some instances, it may be worth giving analgesics, ambulating the patient and attempting voiding by standing up to the sound of running water. A hot bath may also provide a simple solution.
- Check for prostate cancer and renal impairment.
- Perform neurological examination of lower limbs and perianal area.

Chronic retention of urine

May be painless, of insidious onset and present with overflow of urine. Bladder capacity may be >1.5 L.

Faecal impaction

Faecal impaction is encountered typically in the aged, bedridden, debilitated person. Its clinical presentation may closely resemble malignant obstruction.¹⁰ Spurious diarrhoea can occur, which is known as ‘faecal incontinence’ (see [CHAPTER 26](#)).

Acute appendicitis

Acute appendicitis is mainly a condition of young adults but it affects all ages (although uncommon under 3 years). Despite its declining incidence, it is the commonest surgical emergency and special care has to be taken with the very young and the very old. The symptoms can vary because of the different positions of the appendix. It is basically a clinical diagnosis.

Clinical features

See [FIGURE 24.7](#) . Typical clinical features are:

- usually under 30 years of age
- initial pain is central abdominal (sometimes colicky)

- increasing severity and then continuous
- shifts and localises to RIF within 6 hours
- may be aggravated by walking (causing a limp) or coughing
- sudden anorexia
- nausea and vomiting a few hours after the pain starts
- ± diarrhoea and constipation

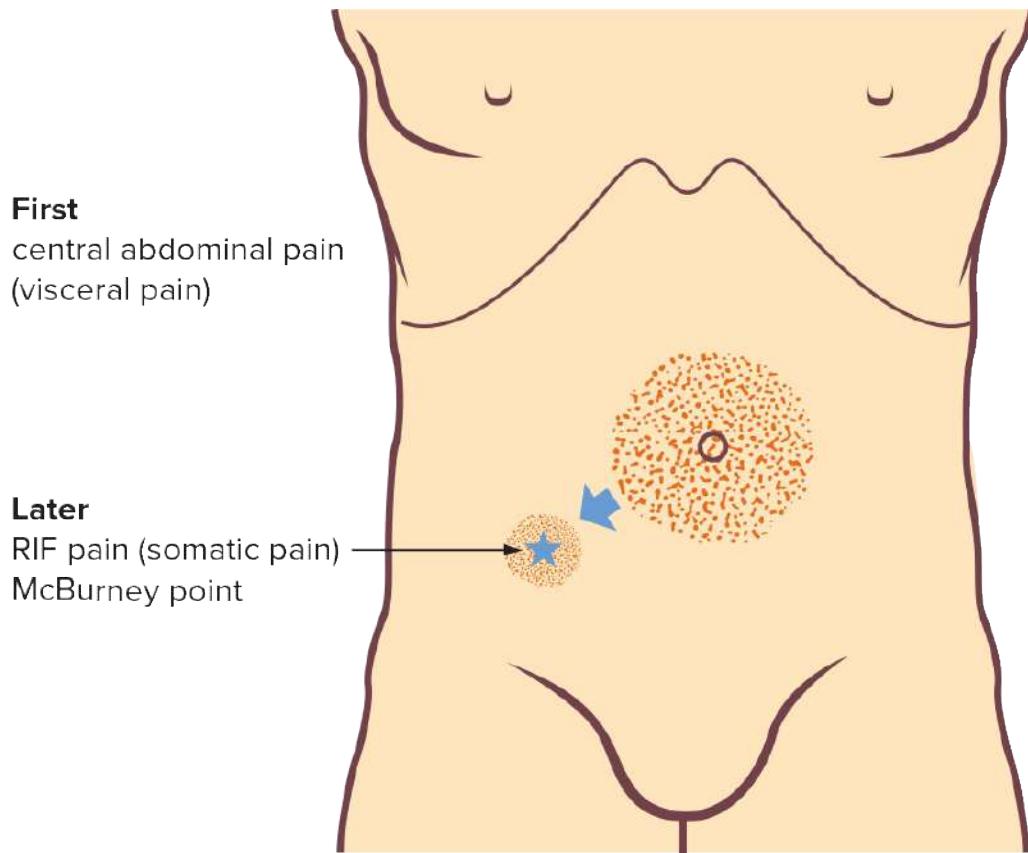


FIGURE 24.7 Typical pain distribution of acute appendicitis



DxT localised RIF pain + a/n/v + guarding → acute appendicitis

Signs

- Patient looks unwell

- Flushed at first, then pale
- Furred tongue and halitosis
- May be febrile—low-grade fever
- Tenderness in RIF, usually at McBurney point
- Local rigidity and rebound tenderness
- Guarding
- ± Superficial hyperaesthesia
- ± Psoas sign: pain on resisted flexion of right leg, on hip extension or on elevating right leg (due to irritation of psoas especially with retrocaecal appendix) Page 266
- ± Obturator sign: pain on the examiner flexing patient's right thigh at the hip with the knee bent and then internally rotating the hip (due to irritation of internal obturator muscle)
- Rovsing sign: rebound tenderness in RIF while palpating in LIF
- PR: anterior tenderness to right, especially if pelvic appendix or pelvic peritonitis

Variations and cautions

- Abscess formation → localised mass and tenderness
- Retrocaecal appendix: pain and rigidity less and may be no rebound tenderness; loin tenderness; positive psoas test
- Pelvic appendix: no abdominal rigidity; urinary frequency; diarrhoea and tenesmus; very tender PR; obturator tests usually positive
- Elderly patients: pain often minimal and eventually manifests as peritonitis; can simulate intestinal obstruction
- Pregnancy (occurs mainly during second trimester): pain is higher and more lateral; harder to diagnose; peritonitis more common
- Perforation more likely in those who are very young, elderly or have diabetes

Investigations

Investigations, including imaging, are of limited value:

- blood cell count shows a leucocytosis (75%) with a left shift
- urea and electrolytes—to assess hydration prior to surgery

- CRP—elevated
- ultrasound shows a thickened appendix (86% sensitivity, 81% specificity);¹¹ affected by gas shadow
- plain X-ray may show local distension, blurred psoas shadow and fluid level in caecum
- CT scan (94% sensitivity, 98% specificity) also allows other causes, especially in the female pelvis, to be evaluated¹²
- laparoscopy
- β-HCG

Management

Immediate referral for surgical removal—the gold standard. If perforated, cover with cefotaxime and metronidazole. If abscess, radiologic drainage, antibiotics ± interval appendicectomy. It is reasonable to offer a conservative approach for uncomplicated, low-grade cases, with careful monitoring and antibiotics in selected patients. One detailed study showed that surgery was the safest option.¹³

Small bowel obstruction

The symptoms depend on the level of the obstruction (see TABLE 24.6). The more proximal the obstruction, the more severe the pain.

Table 24.6 Small bowel obstruction: difference between a high and a low obstruction

	High	Low
Frequency of spasms	3–5 minutes	6–10 minutes
Intensity of pain	+++	+
Vomiting	Early, frequent Violent	Later Less severe
Content:	Gastric juices, then green	Faeculent (later)
Dehydration and degree of illness	Marked	Less prominent
Distension	Minimal	Marked

Main causes

- Outside obstruction (e.g. adhesions—commonest cause, previous laparotomy), strangulation in hernia or pockets of abdominal cavity (see FIG. 24.8)—this may lead to a ‘closed loop’ obstruction.¹⁴
- Lumen obstructions (e.g. foreign body, trichobezoar, gallstones, intussusception, malignancy).

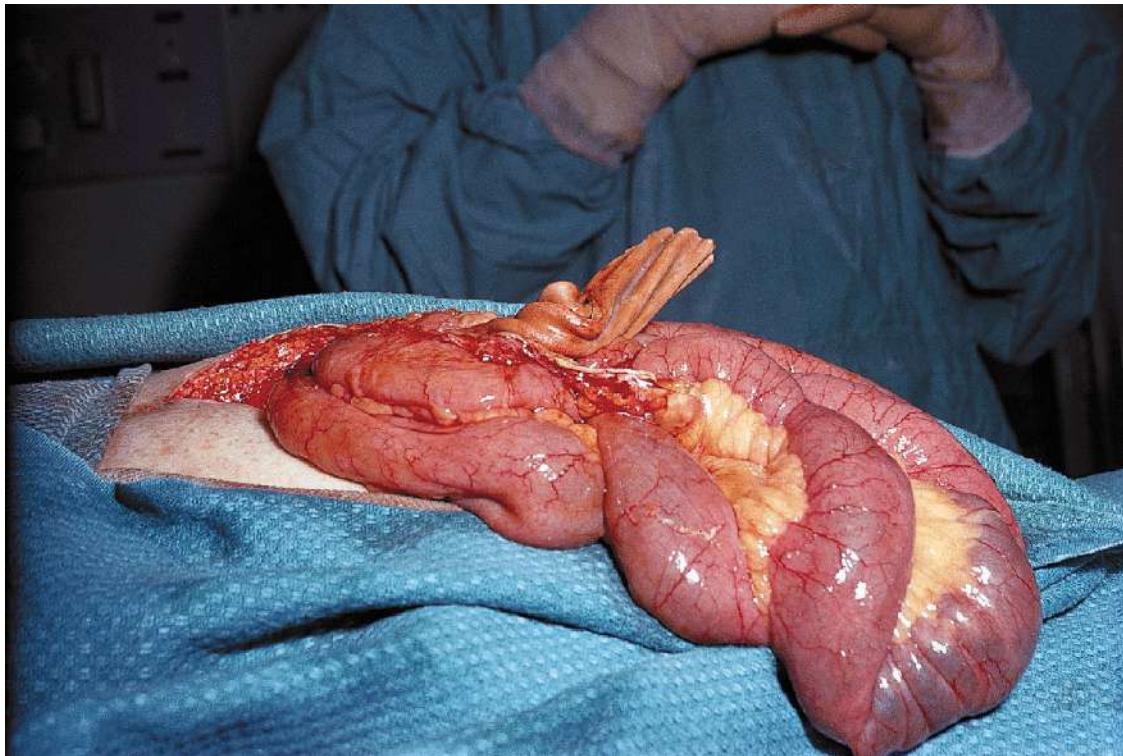


FIGURE 24.8 Operative findings (corrugated drainage material) in a 65-year-old man with subacute bowel obstruction after a 21-year history of nagging abdominal pain following a cholecystectomy

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Clinical features

- Severe colicky epigastric and perumbilical (mainly) pain (see FIG. 24.9)
- Spasms every 3–10 minutes (according to level), lasting about 1 minute
- Vomiting
- Absolute constipation (nil after bowel emptied)

- No flatus
- Abdominal distension (esp. if lower SBO)

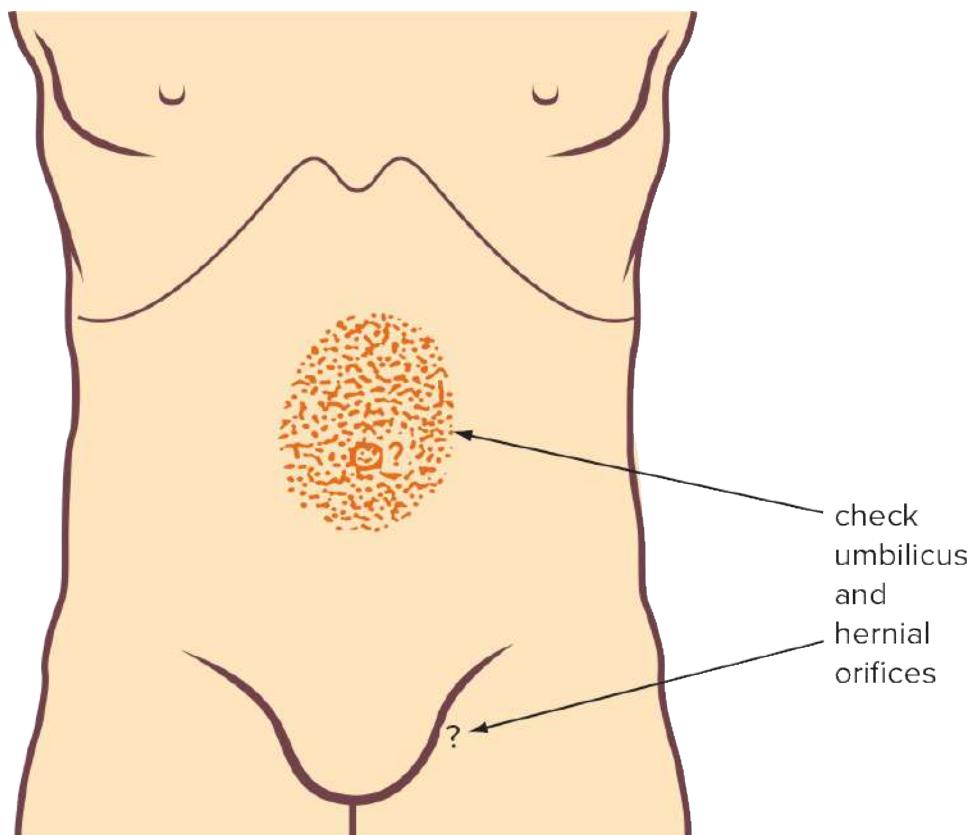


FIGURE 24.9 Typical pain distribution of small bowel obstruction



DxT colicky central pain + vomiting + distension → SBO

Signs and tests

- Patient weak and sitting forward in distress
- Visible peristalsis, loud borborygmi
- Abdomen soft (except with strangulation)
- Tender when distended
- Increased sharp, tinkling bowel sounds
- Dehydration rapidly follows, especially in children and elderly

- PR: empty rectum, may be tender

Note: check all hernial orifices, including umbilicus

- X-ray: plain erect film confirms diagnosis—‘stepladder’ fluid levels (4–5 for diagnosis) in 3–4 hours

Gastrografin follow-through for precise diagnosis with caution. It can cause severe diarrhoea and may be therapeutic in adhesive obstruction.

- ± CT scan (especially if extrinsic causation)

Management

- IV fluids and bowel decompression with nasogastric tube
- Laparotomy or hernia repair

¶ Paralytic ileus

Temporary arrest of peristalsis is common after abdominal surgery. Other causes include drugs, e.g. opioids, TCAs.

Symptoms

- Nausea
- Vomiting
- Vague abdominal discomfort
- Distension
- Constipation/obstipation

Signs

- Silent abdomen
- ↓ Bowel sounds

Tests

- X-ray: air accumulation in small bowel and colon

Management

- Drip and suction

⌚ Large bowel obstruction

The cause is commonly colorectal cancer (75% of cases), especially on the left side, but it can occur in diverticulitis or in volvulus of the sigmoid colon (10% of cases) and caecum.¹⁰ Sigmoid volvulus is more common in older men and has a sudden and severe onset. The pain is less severe than in SBO. Be wary of the non-surgical causes, simple constipation or acute pseudo-obstruction of the colon (Ogilvie syndrome). Consider ileus.

Clinical features

- Sudden-onset colicky pain (even with cancer)
- Each spasm lasts less than 1 minute
- Usually hypogastric midline pain (see FIG. 24.10)
- Vomiting may be absent (or late)
- Absolute constipation (obstipation), no flatus

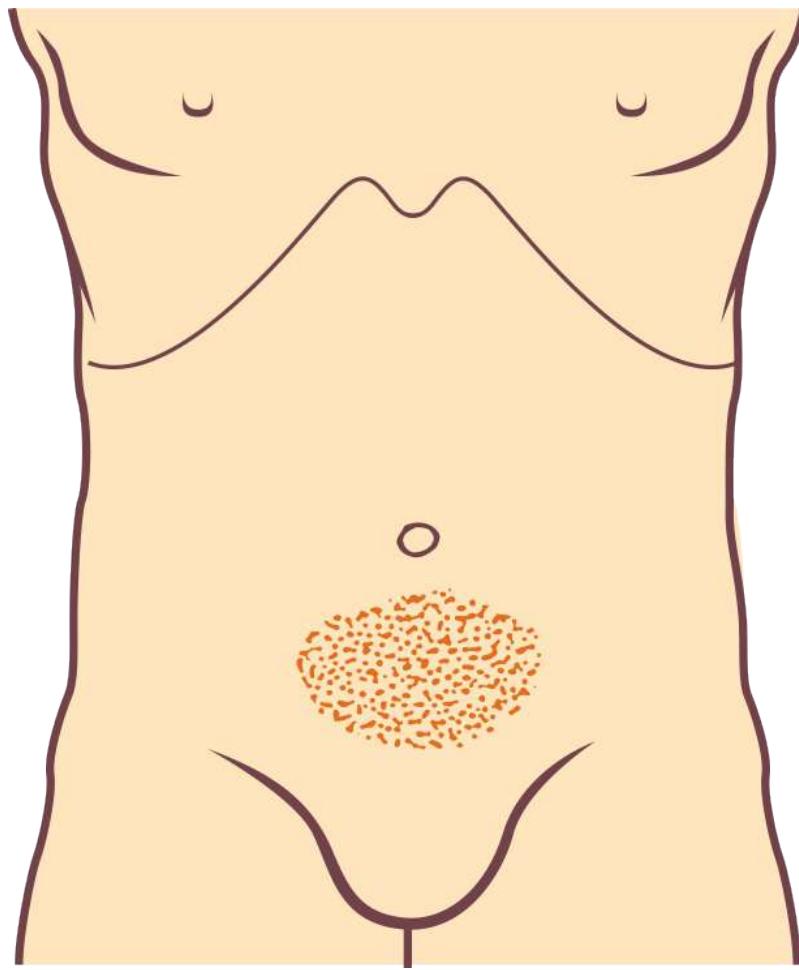


FIGURE 24.10 Typical pain distribution of large bowel obstruction



DxT colicky pain + distension ± vomiting → LBO

Signs and tests

- Increased bowel sounds, especially during pain
- Distension early and marked
- Local tenderness and rigidity
- PR: empty rectum; may be rectosigmoid cancer or blood. Check for faecal impaction
- X-ray: distension of large bowel with separation of haustral markings, especially caecal distension

sigmoid volvulus shows a distended loop and ‘coffee bean’ sign

Gastrografin enema confirms diagnosis

Management

- Drip and suction
- Surgical referral

Perforated peptic ulcer

Perforation of a peptic ulcer can cause acute abdominal pain both with and without a prior history of peptic ulcer. It is an acute surgical emergency requiring immediate diagnosis. Consider a history of drugs, especially NSAIDs. Perforated ulcers may follow a heavy meal. There is usually no back pain. May be painless with steroids.

The maximal incidence is 45–55 years and more common in males, and a perforated duodenal ulcer is more common than a gastric ulcer.

Consider the clinical syndrome in three stages:

1. prostration
2. reaction (after 4 hours)—symptoms may improve
3. peritonitis (after 4 hours)—severe pain

Clinical features

See [FIGURE 24.11](#) . Typical clinical features are:

- sudden-onset severe epigastric pain
- continuous pain but lessens for a few hours
- epigastric pain at first, and then generalised to whole abdomen
- pain may radiate to one or both shoulders (uncommon) or right lower quadrant
- nausea and vomiting (delayed)
- hiccough is a common late symptom

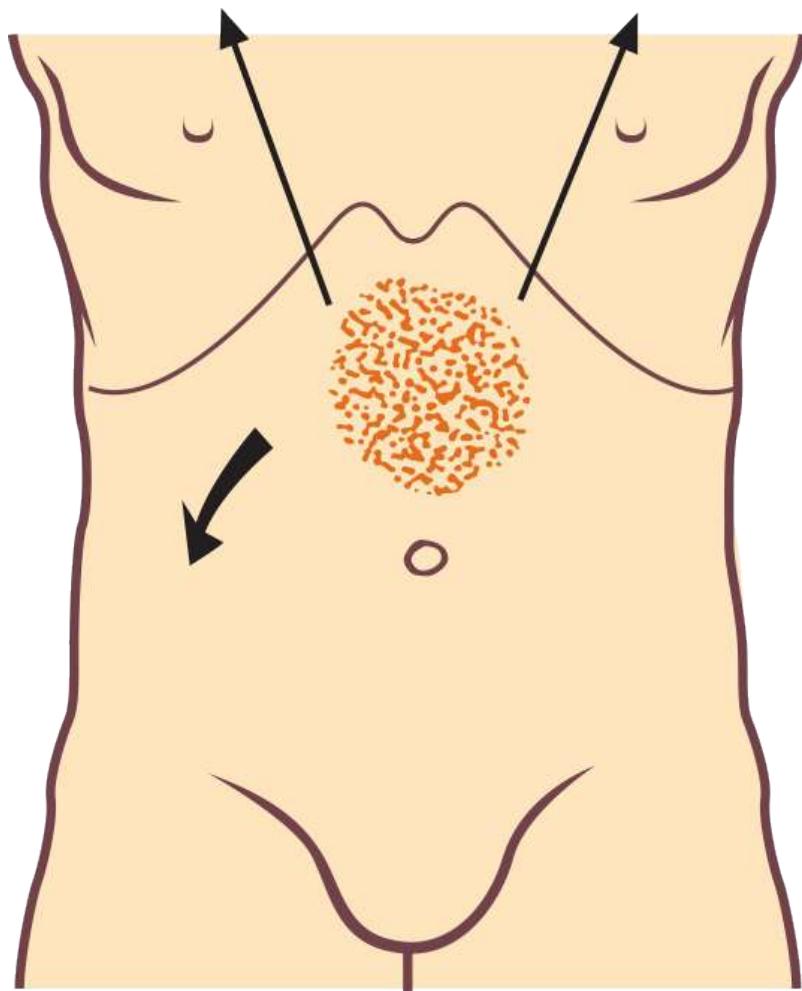


FIGURE 24.11 Typical features of perforated peptic ulcer with typical pain radiation



DxT sudden severe pain + anxious, still, 'grey', sweaty + deceptive improvement → perforated peptic ulcer

Signs and tests (typical of peritonitis)

- Patient lies quietly (pain aggravated by movement and coughing)
- Pale, sweating or ashen at first
- Guarding, board-like rigidity
- Maximum signs at point of perforation
- No abdominal distension

- Contraction of abdomen (forms a ‘shelf’ over lower chest)
- Bowel sounds reduced (silent abdomen)
- Shifting dullness may be present
- Pulse, temperature and BP usually normal at first
- Tachycardia (later) and shock later (3–4 hours)
- Breathing is shallow and inhibited by pain
- PR: pelvic tenderness
- X-ray: chest X-ray may show free air under diaphragm (in 75%)—need to sit upright for prior 15 minutes

limited Gastrograffin meal can confirm diagnosis

CT scan preferable, if available and safe

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Management

- Pain relief
- Drip and suction (immediate nasogastric tube)
- Broad-spectrum antibiotics
- Immediate laparotomy after resuscitation
- Conservative treatment may be possible (e.g. later presentation and Gastrograffin swallow indicates sealing of perforation)

Ureteric colic

Kidney (renal) colic is not a true colic but a constant pain due to blood clots or a stone lodged at the pelvic–ureteric junction; ureteric colic, however, presents as severe true colicky pain due to stone movement, dilatation and ureteric spasm. Fortunately, the majority of urinary calculi are small and will pass spontaneously.

Guidelines:

- loin pain—stone in kidney
- kidney/ureteric colic—ureteric stone

- strangury—stone in bladder

Clinical features

- Maximum incidence 30–50 years (M > F)
- Intense colicky pain: in waves, each lasting 30 seconds with 1–2 minutes respite
- Begins in loin and radiates around the flank to the groin, thigh, testicle or labia (see FIG. 24.12)
- Usually lasts <8 hours
- ± Vomiting

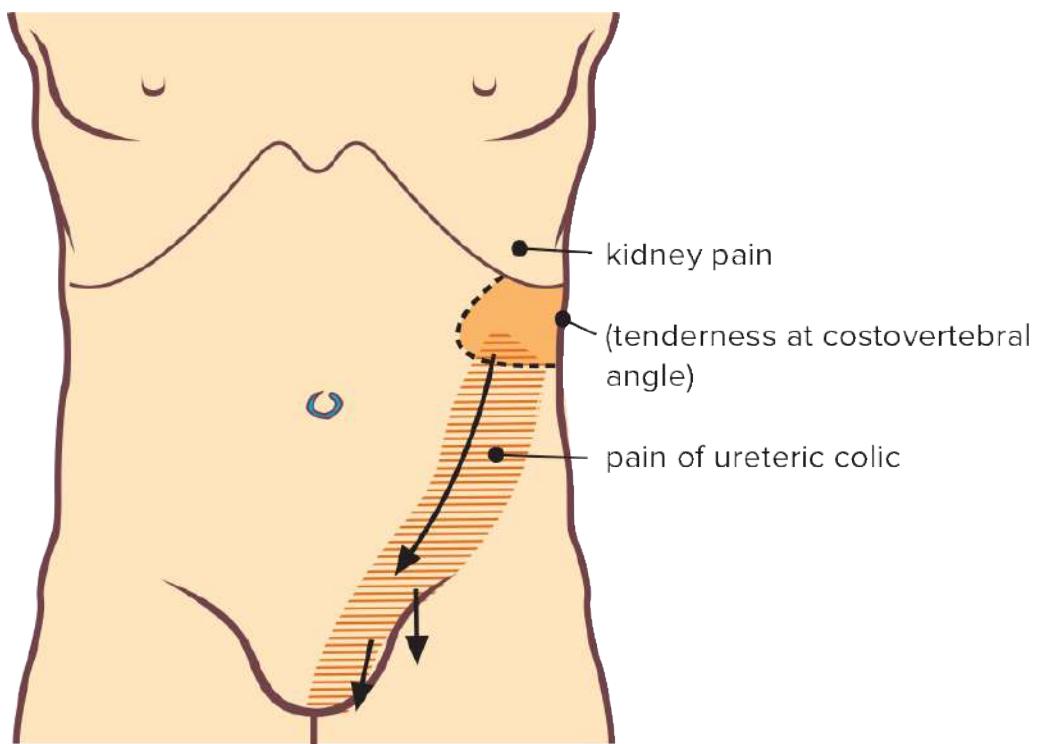


FIGURE 24.12 Ureteric colic: typical radiation of pain in left ureteric colic



DxT intense pain (loin) → groin + microscopic haematuria → ureteric colic

Signs

- Restlessness: may be writhing in pain

- Pale, cold and clammy
- Tenderness at costovertebral angle
- ± Abdominal and back muscle spasm
- Smoky urine due to haematuria

Diagnosis

- Urine: microscopy; blood testing strip (negative does not exclude calculus)
- Plain X-ray: most stones—kidney, ureter, bladder (75%)—are radio-opaque (calcium oxalate and phosphate)
- IVP: confirms opacity, level of obstruction, kidney function and any anatomical abnormalities
- Ultrasound: may locate calculus but will exclude obstruction
- Non-contrast spiral KUB-CT is the ‘gold standard’ (sensitivity 97%, specificity 96%) (will show easily missed radiolucent¹¹ uric acid stones)

Management

If the diagnosis is in doubt (especially if narcotic addiction is suspected) get the patient to pass urine in the presence of an examiner and test for haematuria. While awaiting passage of urine, an indomethacin suppository may be tried for pain relief.

Routine treatment (average size adult)

- Morphine 2.5 to 5 mg IV¹⁵ statim then titrate to effect
or
fentanyl 50–100 mcg IV then titrate to effect.
- Avoid high fluid intake, especially IV fluids—provokes distension of ureter and aggravates pain.
- Most cases settle and the patient can go home when pain relief is obtained and an IVP arranged for the next day.
- Further pain can be alleviated by indomethacin suppositories but should be limited to two a day.
- An effective alternative treatment is diclofenac 75 mg IM injection, then 50 mg (o) tds for 1 week. Several clinical trials have shown that NSAIDs by IM injection, including ketorolac (10–30 mg IM [or IV] 4–6 hourly), are effective and at least as efficacious as opioids.^{15,16,17}

Consider an anti-emetic.

Outcome and follow-up

- The calculus is likely to pass spontaneously if <5 mm (90% <4 mm pass spontaneously).¹⁶
- If >7 mm, intervention will usually be required by extracorporeal shock wave lithotripsy or surgery.
- If the person passes the calculus, he or she should retrieve it and present it for analysis.
- A repeat IVP may be necessary if there is evidence of obstruction for more than 3 weeks.
- Persistent obstruction causes sepsis.
- The cause of the ‘stone’ should be considered. Search for causes such as hyperparathyroidism, hypercalcaemia, hyperoxaluria and UTI.
- Fever with ureteric colic indicates an obstructed infected kidney.

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When to refer¹⁶

Any of the following:

- stone >7 mm in diameter
- high-grade or bilateral obstruction
- gross hydronephrosis
- fever/UTI
- unremitting pain
- stone fails to progress
- type 2 diabetes
- staghorn calculus
- presence of solitary kidney

Facts about urinary tract calculi

- The prevalence is 1 to 3 per 1000 population per year¹⁶
- The lifelong incidence is 10%

- The recurrence is up to 75% (most within 2 years)
- The typical age range is 20–50 years (peaks at 28 years)
- Pregnancy is a risk factor
- Male to female ratio = 3:1
- The incidence is inversely proportional to fibre intake and proportional to ingestion of animal protein and persistently low urinary volume
- Formed from urinary supersaturation with calcium (calcium oxalate, 75–80%), uric acid (7%) and cysteine (rare): also infected calculi (struvite)— Mg^+ , NH_4^+ , PO_4^- (5%)

‘Phony’ colic

Some patients who present with typical colic may be feigning their pain mainly because they are opioid dependent and seeking drugs by deception. As ureteric colic can affect young people (peak age 28 years) this can be a very difficult management issue, even for the experienced.

The use of CT scanning would help in locating calculi in such patients. If in doubt, an appropriate agent would be ketorolac 10–30 mg IM.

It is advisable to obtain a specimen of urine passed in the presence of an examiner and then tested for microscopic blood. While awaiting passage of urine an indomethacin suppository may be tried for pain relief.

Recurrent urinary calculi

Investigations

- Serum electrolytes, urea, creatinine
- Serum calcium, phosphate, uric acid, magnesium
- Serum alkaline phosphatase
- Urine sample—microbiology and culture
- At least two consecutive 24-hour urine samples
- Stone analysis
- IVP

Dietary advice is given in [CHAPTER 5](#) .

Biliary pain

Abdominal pain can be produced by contraction of the biliary tree upon an obstructing stone or inspissated bile (sludge). Although the stereotyped higher-risk person is female, 40, fat, fair and fertile, it can occur from adolescence to old age and in both sexes.

Clinical features

See [FIGURE 24.13](#).

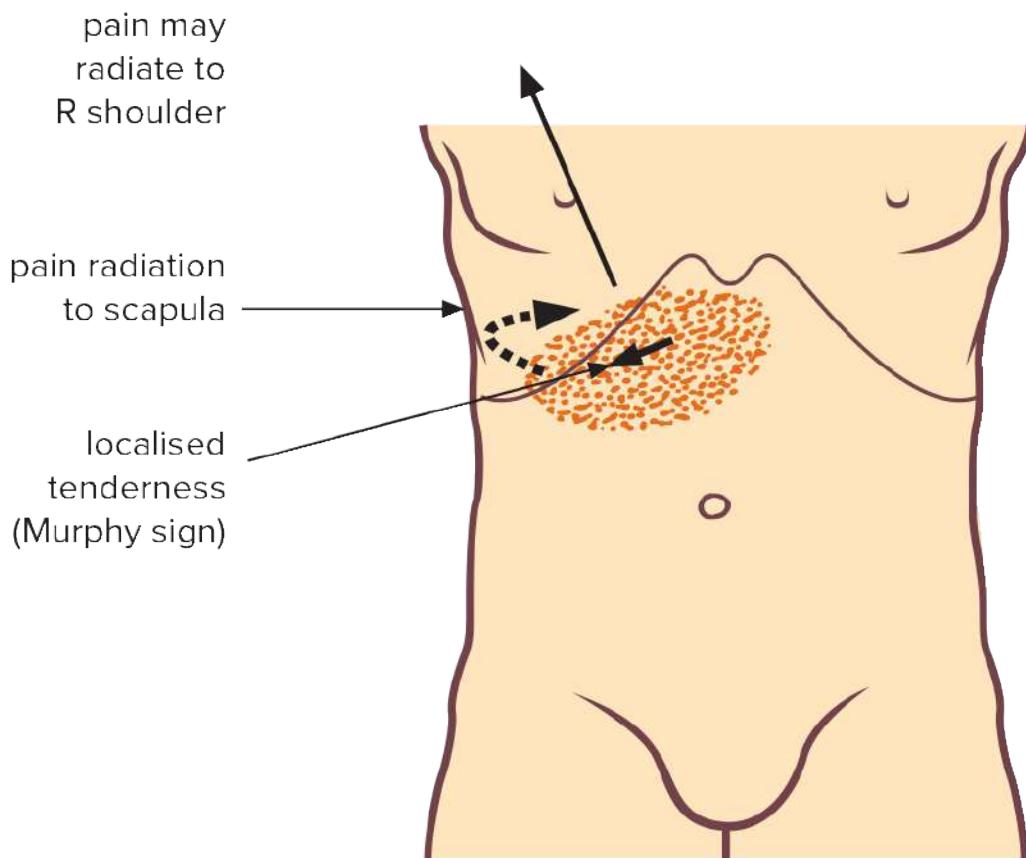


FIGURE 24.13 Typical site of pain of biliary colic and acute cholecystitis

Typical clinical features are:

- acute onset severe pain
- postprandial or at night (often wakes 2–3 am)
- constant pain (not colicky)
- lasts 20 minutes to 2–6 hours

- maximal RUQ or epigastrium
- may radiate to tip of right shoulder or scapula
- painful episode builds to a crescendo for about 20 minutes; may recede or last for hours
- some relief by assuming flexed posture
- ± nausea and vomiting with considerable retching
- often a history of biliary pain (may be mild) or jaundice
- often precipitated by a fatty meal

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DxT severe pain + vomiting + pain radiation → biliary colic

Signs

- Patient anxious and restless, usually in a flexed position or rolling in agony
- Localised tenderness (Murphy sign) over fundus of gall bladder (on transpyloric plane)
- Slight rigidity

Diagnosis

- Abdominal ultrasound (to diagnose gallstones)
- Helical CT
- Intravenous cholangiography if previous cholecystectomy
- LFTs may show elevated bilirubin and alkaline phosphatase

Management

- Pain relief:¹⁸

morphine 2.5–5 mg IV statim then titrate to effect (age-dependent; use lower end of range if ≥70 years)

or

fentanyl 50–100 mcg IV statim then titrate to effect

or

ketorolac 10–30 mg IM 4–6 hourly (max. 90 mg daily)

- Gallstone dissolution with ursodeoxycholic acid or lithotripsy (in those unable to have surgery)
- Laparoscopic cholecystectomy (main procedure)

Gallstone facts¹⁸

- Gallstones form from bile in the gall bladder and sometimes in the bile duct (especially post-cholecystectomy)
- Two main types—cholesterol and pigment (bilirubin)
- Lifetime risk in first world countries is 12–20%
- 70% of people with gall bladder stones are asymptomatic, but risk of developing symptoms is about 15% over 20 years
- Cholecystectomy almost never indicated for asymptomatic stones
- Complications: acute cholecystitis (may lead to empyema, perforation, cholecystoenteric fistula), obstructive jaundice, cholangitis (infection with pain) and acute pancreatitis (pancreatic duct obstruction)

Microlithiasis (biliary ‘sludge’)

This condition, which causes biliary ‘colic’ due to spasm of the sphincter of Oddi, often follows prolonged fasting. Cholecystectomy may be necessary.

⌚ Acute cholecystitis

Cholecystitis is associated with gallstones in over 90% of cases¹⁸ and there is usually a past history of biliary pain. It occurs when a calculus becomes impacted in the cystic duct and inflammation develops. It is very common in the elderly. The acute attack is often precipitated by a large or fatty meal. The causative organisms are usually aerobic bowel flora (e.g. *E. coli*, *Klebsiella* species and *Enterococcus faecalis*).

Clinical features

- Steady severe pain and tenderness
- Localised to right hypochondrium or epigastrium
- May be referred to the right infrascapular area
- Anorexia, nausea and vomiting (bile) in about 75%

- Aggravated by deep inspiration

Signs

- Patient tends to lie still
- Localised tenderness over gall bladder (positive Murphy sign)
- Muscle guarding
- Rebound tenderness
- Palpable gall bladder (approximately 15%)
- Jaundice (approximately 15%)
- ± Fever

Diagnosis

- Ultrasound: gallstones but not specific for cholecystitis
- HIDA scan: demonstrates obstructed cystic duct—the usual cause
- WCC and CRP: can be elevated

Treatment

- Bed rest
- IV fluids
- Nil orally
- Analgesics
- Antibiotics
- Cholecystectomy

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If evidence of sepsis, use amoxi/ampicillin 1 g IV, 6 hourly plus gentamicin 4–6 mg/kg IV daily.¹⁹

Change to amoxicillin + clavulanate 875 + 125 mg (o) 12 hourly when afebrile.

⌚ Acute pancreatitis

With acute pancreatitis there may be a past history of previous attacks or a past history of

alcoholism (35%) or gallstone disease (40–50%). It is commonly precipitated by fatty foods and alcohol, mumps, hypertriglyceridaemia and some antidiabetic medications, e.g. gliptins.

Clinical features

See [FIGURE 24.14](#).

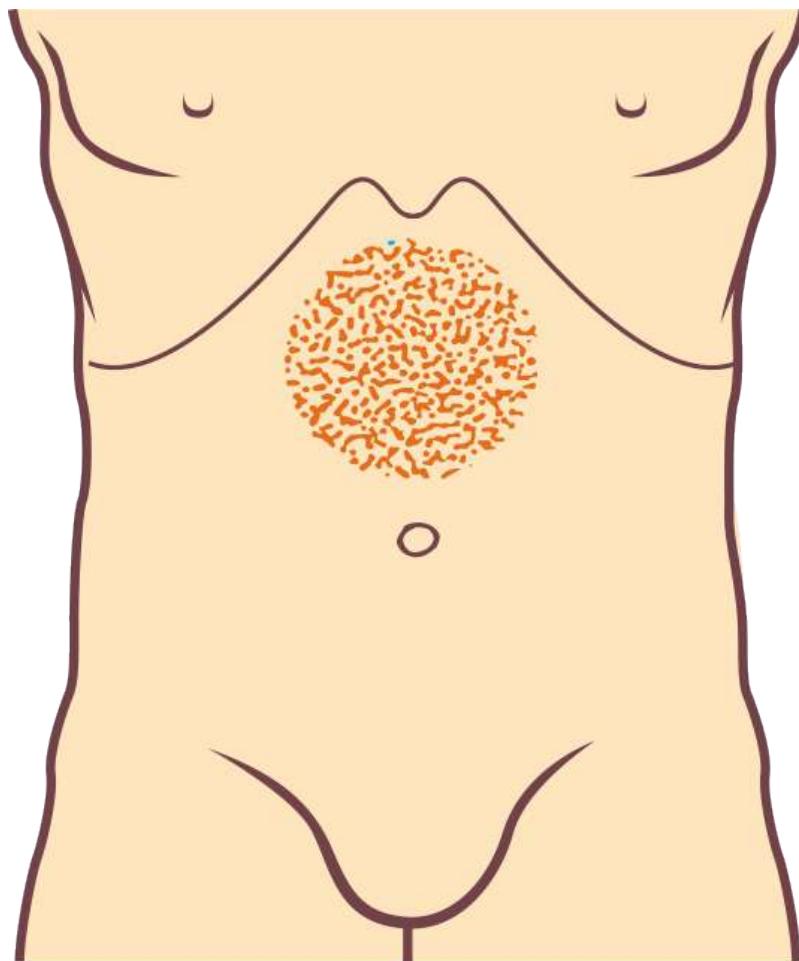


FIGURE 24.14 Typical pain distribution of acute pancreatitis

Typical clinical features are:

- sudden onset of severe constant deep epigastric pain but onset can be steady
- lasts hours or a day or so
- pain may radiate to back
- pain may be relieved by sitting forwards

- nausea and vomiting
- sweating and weakness



DxT severe pain + nausea and vomiting + relative lack of abdominal signs → acute pancreatitis

Signs

- Patient is weak, pale, sweating and anxious
- Tender in epigastrium
- Lack of guarding, rigidity or rebound
- Reduced bowel sounds (may be absent if ileus)
- ± Abdominal distension
- Fever, tachycardia ± shock

Diagnosis

- WCC—leucocytosis
- Serum lipase (preferred as more sensitive and specific) or serum amylase
- CRP—elevated
- Serum glucose ↑, calcium ↓
- Blood gases: P_aO_2 (?pulmonary complications)
- LFTs: ?obstructive pattern
- Plain X-ray, may be sentinel loop
- CT scan, best after 48 hours (especially for complications)
- Ultrasound better for detecting cysts and unsuspected gallstones

Management¹⁹

- Arrange admission to hospital (but many cases are mild).
- Basic treatment is bed rest, nil orally, nasogastric suction (if vomiting), IV fluids and analgesics (morphine). Treat hyperglycaemia or hypocalcaemia.

- Use morphine 2.5–5 mg IV or fentanyl 30–100 mcg IV statim then titrate to effect.
- May require ERCP if obstructive LFTs.

Autoimmune pancreatitis¹⁸

This IgG4-related disorder presents with abdominal pain, jaundice and weight loss. Diagnosis is by a pancreatic mass or enlargement on imaging and serology (IgG4). Treatment is with corticosteroids.

Chronic pancreatitis

In comparison to acute pancreatitis, the pain of chronic pancreatitis is milder but more persistent. There may be epigastric pain boring through to the back. Symptoms may relapse and worsen. Investigate with CT scan and ultrasound and faecal elastase ($N > 200 \mu\text{g/g}$ stool: pancreatic exocrine insufficiency $< 200 \mu\text{g/g}$ stool). MRCP is the most sensitive imaging study. The person with this problem is often labelled as ‘gastritis’, ‘ulcer’ or even ‘neurotic’ because of the indeterminate nature of the pain. Malabsorption and diabetes may result from pancreatitis. Weight loss and steatorrhoea become prominent features.

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Pain associated with pancreatic cancer is indistinguishable from that of chronic pancreatitis but generally tends to be more severe and more prominent in the back. Use paracetamol for pain. Give pancreatic enzyme supplements (e.g. pancrelipase) for malabsorption.

Acute diverticulitis

The person with acute diverticulitis is usually over 40 years of age, with longstanding, grumbling, left-sided abdominal pain and constipation, but can have an irregular bowel habit. It occurs in less than 10% of those with diverticular disorder.⁴ (Refer to [CHAPTER 34](#) .)

Clinical features

See [FIGURE 24.15](#) .

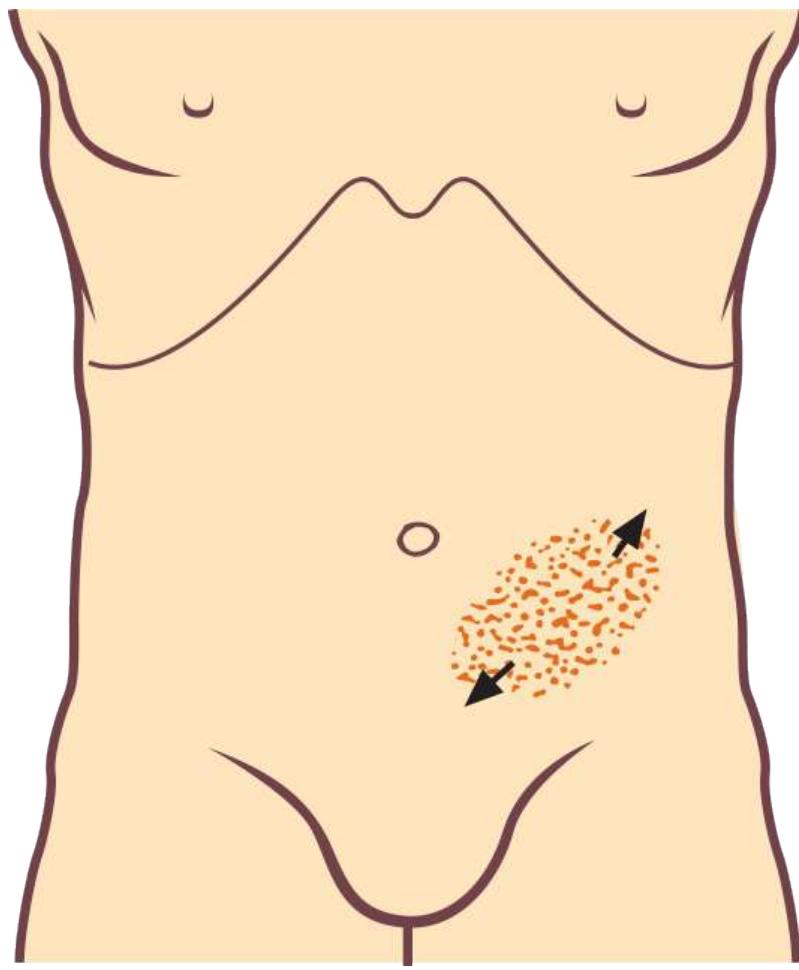


FIGURE 24.15 Typical pain distribution of acute diverticulitis

Typical clinical features are:

- acute onset of pain in the left iliac fossa
- pain increased with walking and change of position
- usually associated with constipation



DxT acute pain + left-sided radiation + fever → acute diverticulitis

Signs

- Tenderness, guarding and rigidity in LIF
- Fever

- May be inflammatory mass in LIF

Investigations

- FBE: leucocytosis
- Elevated ESR
- Pus and blood in stools
- Abdominal ultrasound/CT scan (especially—can detect fistula, abscess or perforation)
- Erect chest X-ray
- Erect and supine abdominal X-ray

Complications

- Bleeding (can be profuse, especially in elderly)
- Perforation (high mortality)
- Abscess
- Peritonitis
- Fistula (bladder, vagina, small bowel)
- Intestinal obstruction

Treatment¹⁹

- Hospital admission (unless mild)
- Rest the GIT: nil orally, drip and suction
- One landmark study showed that a ‘wait and see’ approach without antibiotics can be considered appropriate in patients with an uncomplicated initial attack of diverticulitis²⁰
- Analgesics
- Antibiotics:

mild cases: amoxicillin + clavulanate 875/125 mg (o) 12 hourly for 5 days

or

metronidazole + cephalexin

severe cases: amoxi/ampicillin 1 g IV 6 hourly

+

gentamicin 5–7 mg/kg IV/day

+

metronidazole 500 mg IV 12 hourly

or

metronidazole + ceftriaxone 1 g IV/day

- Surgery for complications
- Screening colonoscopy after acute episode

Peritonitis

Can be generalised due to intra-abdominal sepsis following perforation of a viscus, e.g. peptic ulcer, appendix, diverticulum. Typical signs are as for perforated peptic ulcer. Key investigations are peritoneal fluid culture and CT scan. Surgical intervention is usually required. Usual antibiotic treatment is IV cephalosporins or amoxi/ampicillin + gentamicin + metronidazole.²¹ Spontaneous bacterial peritonitis can occur in any patient with ascites.

Abdominal ‘stitch’

The common ‘stitch in the side’ is the experience of a sharp, stabbing pain in the epigastric or hypochondrium regions of the abdomen, usually during running. The sufferer should:

- stop and rest, then walk—don’t run
- apply deep massage to the area with the palps (fleshy tips) of the middle three fingers
- perform slow or deep breathing

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Chronic or recurrent abdominal pain

Advances in technology, particularly the use of ultrasound, CT scanning and endoscopy, have increased the opportunities for diagnosing chronic or recurrent abdominal pain in adults.

If there are ‘red flag’ symptoms (see box) and the above investigations are unavailable, consider the possibility of conditions such as pancreatic cancer, ovarian cancer, small bowel tumours, mesenteric ischaemia, Crohn disease, metabolic disorders such as lactase deficiency, and rarer conditions as outlined in TABLE 24.2 .

Other investigations that may help:

- MRI
- laparoscopy—this may allow the identification of chronic adhesive obstruction, small bowel tumours or inflammation, or intra-abdominal malignancy

Red flags for organic disease¹²

- Older person
- Nocturnal pain or diarrhoea
- Progressive symptoms
- Rectal bleeding
- Fever
- Anaemia
- Weight loss
- Abdominal mass
- Faecal incontinence or urgency (recent onset)

⌚ Chronic appendicitis

It is possible to have recurrent episodes of subacute inflammation of the appendix. If suspected, laparoscopy performed during or soon after an attack is diagnostic.

⌚ Adhesions

There is no firm evidence that intra-abdominal adhesions are painful apart from complications such as bowel obstruction. Sometimes patients are ‘cured’ by laparoscopic divisions of adhesions.

⌚ Peptic ulcer (gastric or duodenal)

See [CHAPTER 36](#).

Clinical features

- Usually central epigastric pain
- Burning pain
- Relieved by antacids or food or milk
- DU: usually 2–3 hours after meals or wakes from sleep
- GU: may occur after meals but inconsistent relationship to eating

When to refer

- All cases of acute abdominal pain where urgent surgical intervention is required
- Special urgency and early diagnosis is important with: ruptured ectopic pregnancy, ruptured AAA, mesenteric artery occlusion, ruptured viscus, perforated peptic ulcer, strangulated obstructed bowel, intussusception
- All cases where surgery is necessary
- Complex medical causes, such as diabetic ketoacidosis and porphyria
- Have a low threshold for referral if acute cause is not apparent

Practice tips

- Special caution is required at the extremes of age when the symptoms and signs do not often reflect the seriousness of the underlying pathology.
- If an elderly person presents with intense acute abdominal pain, inadequately relieved by strong parenteral injections, likely causes include mesenteric artery occlusion, acute pancreatitis and ruptured or dissecting aortic aneurysm.
- When an inflamed appendix ruptures, the abdominal pain improves for a significant period of time.
- Consider gallstones and duodenal ulcer if the person is woken (e.g. at 2–3 am) with abdominal pain.
- Pus cells and red cells may be present in the urine with appendicitis when a pelvic appendix involves the bladder and a retrocaecal appendix involves the ureter.
- Consider diabetic ketoacidosis in a person with abdominal pain, tenderness and rigidity and deep sighing respiration.

Patient education resources

Hand-out sheets from *Murtagh's Patient Education* 8th edition:

- Appendicitis
- Diverticular disease
- Gallstones
- Infant colic
- Irritable bowel
- Kidney stones
- Pancreatitis

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25 Arthritis

Rheumatic disorders are common in old age: much of rheumatology is geriatric and much of geriatrics is rheumatology.

DR FRANK DUDLEY HART¹ 1983

The clinical evaluation of the presentation of arthralgia (painful joints) or arthritis (inflammation of the joints) can be a difficult and challenging exercise because it can be a presentation of many systemic disorders, some of which are rare. Important considerations are sex, age, the pattern of joint involvement (monoarticular or polyarticular), immediate and more remote history, family history and drug use—all of which may provide important diagnostic clues. Polyarthritis, which implies the active inflammation of five or more joints, presents a more challenging diagnostic problem.

Key facts and checkpoints

- In a UK National Morbidity Survey, joint symptoms composed just over 7% of all morbidity presenting to the family doctor.²
- The commonest cause was osteoarthritis (OA), which affects 5–10% of the population.
- Almost 2% of Australians report having rheumatoid arthritis (RA).³
- There should be no systemic manifestations with OA.
- One-quarter of disability in elderly people is due to severe joint disease.
- Systemic diseases that may predispose to, or present with, an arthropathy include the connective tissue disorders, diabetes mellitus, a bleeding disorder, previous tuberculosis, the spondyloarthropathies such as psoriasis, SBE, hepatitis B, rheumatic fever, the various vasculitic or arteritic syndromes (the vasculitides) such as Wegener granulomatosis, HIV infection, lung cancer, haemochromatosis, sarcoidosis, hyperparathyroidism, Whipple disease and

Paget disease.

- The pain of inflammatory disease is worse at rest (e.g. on waking in the morning, also with stiffness) and improved by activity.
- Early diagnosis and management of RA results in considerably better outcomes.
- Causes of monoarthritis include crystal deposition disease, sepsis, osteoarthritis, trauma and spondyloarthritis.
- Gout and septic arthritis have a recognised cause and cure.
- Acute gout is 4–6 times more prevalent in men than women; however, it becomes more common in postmenopausal women, particularly those on thiazide diuretics.⁴

A diagnostic approach

A summary of the diagnostic strategy model is presented in TABLE 25.1 .

Table 25.1 Arthralgia: diagnostic strategy model

Probability diagnosis

Osteoarthritis

Viral polyarthritis (e.g. parvovirus)

Serious disorders not to be missed

Infection:

- rheumatic fever
- endocarditis
- tuberculosis
- brucellosis
- pyogenic (septic) arthritis: e.g. gonococcus, *Staphylococcus*, *Kingella kingae*
- HIV arthropathy
- dengue fever

Cancer:

- bronchogenic carcinoma
- leukaemia/lymphoma
- secondary malignancy

Other:

- rheumatoid arthritis (RA)

- connective tissue disorders: SLE, scleroderma, polymyositis and dermatomyositis, psoriasis, other

Pitfalls (often missed)

Fibromyalgia syndrome

Polymyalgia rheumatica

Crystal deposition:

- gout
- pyrophosphate (pseudogout)

Haemarthrosis

Dengue fever

Lyme disease

Ross River virus

Avascular necrosis

Rarities:

Other vasculitides (e.g. polyarteritis nodosa)

Haemochromatosis

Sarcoidosis

Whipple disease

Hyperparathyroidism

Familial Mediterranean fever

Amyloidosis

Pigmented villonodular synovitis

Seven masquerades checklist

Depression (unlikely)

Diabetes (?arthropathy)

Drugs (esp. narcotics)

Endocrine disorder (thyroid storm, Addison disease)

Spinal dysfunction (possible spondyloarthropathies)

Is the patient trying to tell me something?

Always a consideration with pain. Psychogenic factors aggravate chronic arthritic conditions.

Probability diagnosis

The probability diagnoses for the person presenting with arthritis are:

- osteoarthritis (mono- or polyarthritis)

- viral arthritis (if acute and polyarthritis)

OA is very common in general practice. It may be primary, which is usually symmetrical, and can affect many joints. This clinical pattern is different from secondary OA, which follows injury and other wear-and-tear causes.

Viral polyarthritis is more common than realised. It presents usually within 10 days of the infection, and is usually mild.

Clinical features (viral arthritis)

- Acute onset
- Polyarthritis
- Symmetric inflammation
- Mainly hands and feet
- Rash—persists for 24 hours minimum
- Terminates rapidly—over days
- FBE: lymphopaenia, lymphocytosis, ± atypical lymphocytes

It tends to terminate quickly and spontaneously without permanent damage to joints. It is caused by many viruses, including those causing influenza, mumps, rubella, varicella, hepatitis B and C, infectious mononucleosis (more muscle aching), cytomegalovirus, parvovirus, Australian epidemic polyarthritis due to the alphaviruses, Ross River virus and Barmah Forest virus.

Adenovirus is common in children. COVID-19 causes arthralgia in around 15% of cases.⁵

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Serious disorders not to be missed

These include rheumatoid arthritis (RA), which can start as a monoarthritis; pyogenic arthritis, including gonococcus, *Staphylococcus*, *Kingella* sp. and *Streptococcus* infections; tuberculosis; rheumatic fever; and bacterial endocarditis. Early diagnosis of septic arthritis is very important as it can destroy a hip in 24 hours.

It is important to be forever watchful for rheumatic fever (RF). It presents typically as a migratory polyarthritis involving large joints sequentially, one becoming hot, red, swollen and very painful as the other subsides. It rarely lasts more than 5 days in any one joint.

A flitting polyarthritis can also occur with endocarditis in addition to a systemic upset and a cardiac murmur. Gonococcal infection may present in a single joint or as flitting polyarthritis, often accompanied by a rash. Brucellosis can cause arthritis and sacroiliitis and can be confused with the spondyloarthropathies.

HIV infection can present as a chronic oligoarticular asymmetrical arthritis.⁶ It can also involve a rash very similar to psoriasis. Tuberculosis can also present as an arthritis.

Connective tissue disorders may be involved. They include SLE, progressive systemic sclerosis (scleroderma) and dermatomyositis. Extra-articular features include rash, sicca symptoms, red eyes, chronic diarrhoea, oral ulcers and urethral discharge. It is most inappropriate to settle with a general diagnosis such as ‘rheumatism’ or ‘arthritis’ and where doubtful it is important to find the specific entity causing the problem.

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In respect to malignant disease, arthralgia is associated with acute leukaemia, lymphoma and neuroblastoma in children and with bronchial carcinoma, which may cause hypertrophic osteoarthropathy, especially of the wrist and ankle (not a true arthritis but simulates it). Occasionally, polyarthritis may be the first feature of an occult neoplasm. Monoarticular metastatic disease may involve the knee (usually from lung or breast).

Red flag pointers for polyarthritis

- Fever
- Weight loss
- Profuse rash
- Lymphadenopathy
- Cardiac murmur
- Severe pain and disability
- Malaise and fatigue
- Vasculitic signs
- Two or more systems involved

Pitfalls

A common pitfall is gout, particularly in older women taking diuretics, whose osteoarthritic joints, especially of the hand, can be affected. The condition is often referred to as nodular gout and does not usually present as acute arthritis.

Fibromyalgia syndrome is a real puzzle (see CHAPTER 27) as it can mimic the connective tissue disorders in its early presentation—typically a woman in the third or fourth decade.

Another ‘trap’ is haemarthrosis in a patient with a bleeding disorder.

Infective causes that may be overlooked are dengue fever, especially in travellers returning from a tropical or subtropical area, and Lyme disease, which is now surfacing in many countries, especially where ticks are found.

There are many rare causes of arthritis. Sarcoidosis causes two forms: an acute benign form, usually in the ankles and knees, and a chronic form with longstanding sarcoidosis that involves joints (large or small) adjacent to underlying bone disease.

Then there are the uncommon vasculitides, which can cause confusion in diagnosis. This group includes polyarteritis nodosa, hypersensitive vasculitis, polymyalgia rheumatica/giant cell arteritis, Wegener granulomatosis, Henoch–Schönlein purpura and Behçet syndrome.

Haemochromatosis can present with a degenerative arthropathy that characteristically affects the second or third metacarpophalangeal joints.⁶

Other rare causes of arthritis are erythema nodosum, serum sickness and Sjögren syndrome.

General pitfalls

- Not searching beyond RA when an RA pattern polyarthritis may be part of another systemic disease
- Failing to search for some cause of arthritis other than OA, especially in an elderly patient (i.e. underdiagnosing); an important example of this is polymyalgia rheumatica
- Failing to consider the various drug interactions between NSAIDs, over-the-counter medications and other drugs used by the elderly
- Underdiagnosing and misdiagnosing through lack of appreciation of the many causes of arthritis, especially those presenting as part of a systemic disease

Seven masquerades

Drug-induced arthritis usually affects the hands and is generally symmetrical. The drugs may induce autoantibodies (e.g. ANA, ANCA). Those that induce a lupus syndrome include the anti-epileptics, chlorpromazine and some cardiac drugs. Various antibiotics have been associated with arthralgia, e.g. minocycline, while diuretics, especially frusemide and thiazides, can precipitate gout. The problem usually resolves promptly after withdrawal of the agent.⁷

Intravenous drug abuse may be associated with septic arthritis, hepatitis B and C, HIV-associated arthropathy, SBE with arthritis and serum sickness reactions.

Hyperthyroidism can uncommonly cause acropathy (clubbing and swelling of the fingers) and may present as pseudogout, while hypothyroidism can present with an arthropathy or cause proximal muscle pain, stiffness and weakness. Diabetes mellitus may cause an arthropathy that can be painless or mild to moderately painful.

The spondyloarthropathies may be a causative factor. They often present with an acute monoarthritis, particularly in teenagers, some time before causing sacroiliitis and spondylitis.

Psychogenic considerations

Although ‘arthralgia’ is an uncommon complaint in psychoneurotic disorders, any pain syndrome can be a significant manifestation. The usual cause of arthralgia is inflammation in the joint—that is, arthritis—but a functional cause is encountered from time to time.

Furthermore, some patients who are unfortunate enough to acquire arthritis, especially the more serious disorders, certainly develop ongoing emotional and psychological problems that appear to aggravate their total problem.

So-called ‘growing pains’ of the lower limb are common in children, and the physical examination and investigations are normal. Parents need to be reassured that it is a benign condition, while recognising that emotional factors may be quite significant. As Apley pointed out, ‘physical growth is not painful, but emotional growth can hurt like hell’.⁸

The clinical approach

A priority is to determine whether or not the arthritis is caused by a primary rheumatic disorder or whether it is part of an underlying systemic disorder.

History

Very careful enquiry about the exact onset of the arthritis is important. This includes whether it was acute or insidious, and confined to specific joints or flitting as in rheumatic fever and sometimes in infective endocarditis. Is it a true polyarthritis or monoarthritis? Symmetrical or asymmetrical? It is also important to differentiate between arthralgia (pains in or around the joints) and arthritis (inflammation of the joints). Not all arthralgia is arthritis.

A family history is important because a positive family history is associated with conditions such as RA (rarely), ankylosing spondylitis, connective tissue disorders (rarely), psoriasis, gout, pseudogout and haemophilia.

A very hot, red, swollen joint suggests either infection or crystal arthritis.

Key questions

- Can you carefully point out exactly where you feel the pain?
- Does the pain move from joint to joint or stay in the same joint?
- Are you aware of anything that brought on the pain?

- Does the pain disturb you at night?
- Do your joints feel very sore or stiff when you wake up in the morning?
- What effect does exercise or activity have on the pain or stiffness?
- Have you had an injury in the past to your painful joint(s)?
- Have you got a skin rash? Is it new?
- Have you had a fever, sweats or chills?
- Do you get very tired, weak or out of sorts?
- Have you noticed any change in the colour of your urine?
- Have you had a sore throat?
- Have you had acute pain in your big toe or in other joints before?
- Do you have a history of psoriasis?
- Do you have a history of rheumatic fever?
- Do you have pain in your neck or lower back or in other joints?
- Have you had any diarrhoea?
- Are you at risk of getting an STI? Have you had a vaginal/penile discharge?
- Have you had any problems with your eyes?
- What drugs are you taking? Are you taking fluid tablets (diuretics)?
- How much alcohol would you drink a day?
- Have you travelled overseas recently?
- Have you been drinking untreated milk recently?
- Have you had cats as pets, especially as a child (associated with RA)?⁹

Examination

A systematic examination of the affected joint or joints should be performed, looking for signs of inflammation, deformity, swelling and limitation of movement. Tenderness and warmth indicates inflammatory activity. Erythema indicates gouty arthritis or other crystallopathy, rheumatic fever or septic arthritis.

Joint swelling:

- acute (1–4 hours) with intense pain = blood infection or crystals (e.g. gout)
- subacute (1–2 days) and soft = fluid (synovial effusion)
- chronic and bony = osteoarthritis
- chronic and soft/boggy = synovial proliferation

A coarse crepitus suggests OA. Each joint should be examined specifically. Inspection should note the presence of lumps or bumps such as Heberden nodes on the osteoarthritic DIP joints of the hands, Bouchard nodes on the osteoarthritic PIP joints of the hands and rheumatoid nodules, which are the only pathognomonic finding of RA and gouty tophi. Signs that may be of diagnostic help are presented in [FIGURE 25.1](#) .

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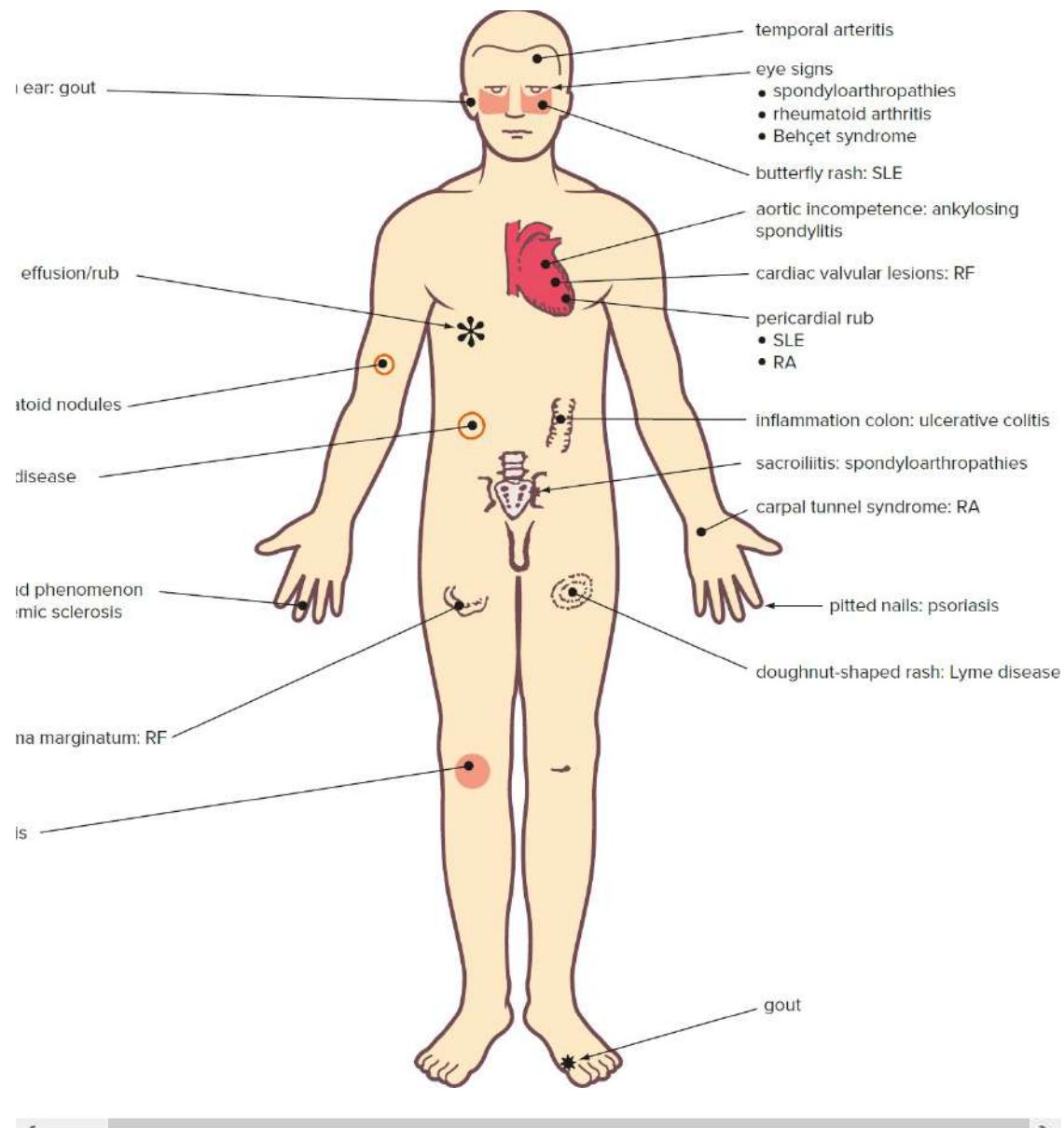


FIGURE 25.1 Physical examination: possible findings to consider in diagnosis

The specific inflamed joint or joints may give an indication of the disease process. Typical joints affected by various arthropathies are illustrated in [FIGURE 25.2](#).

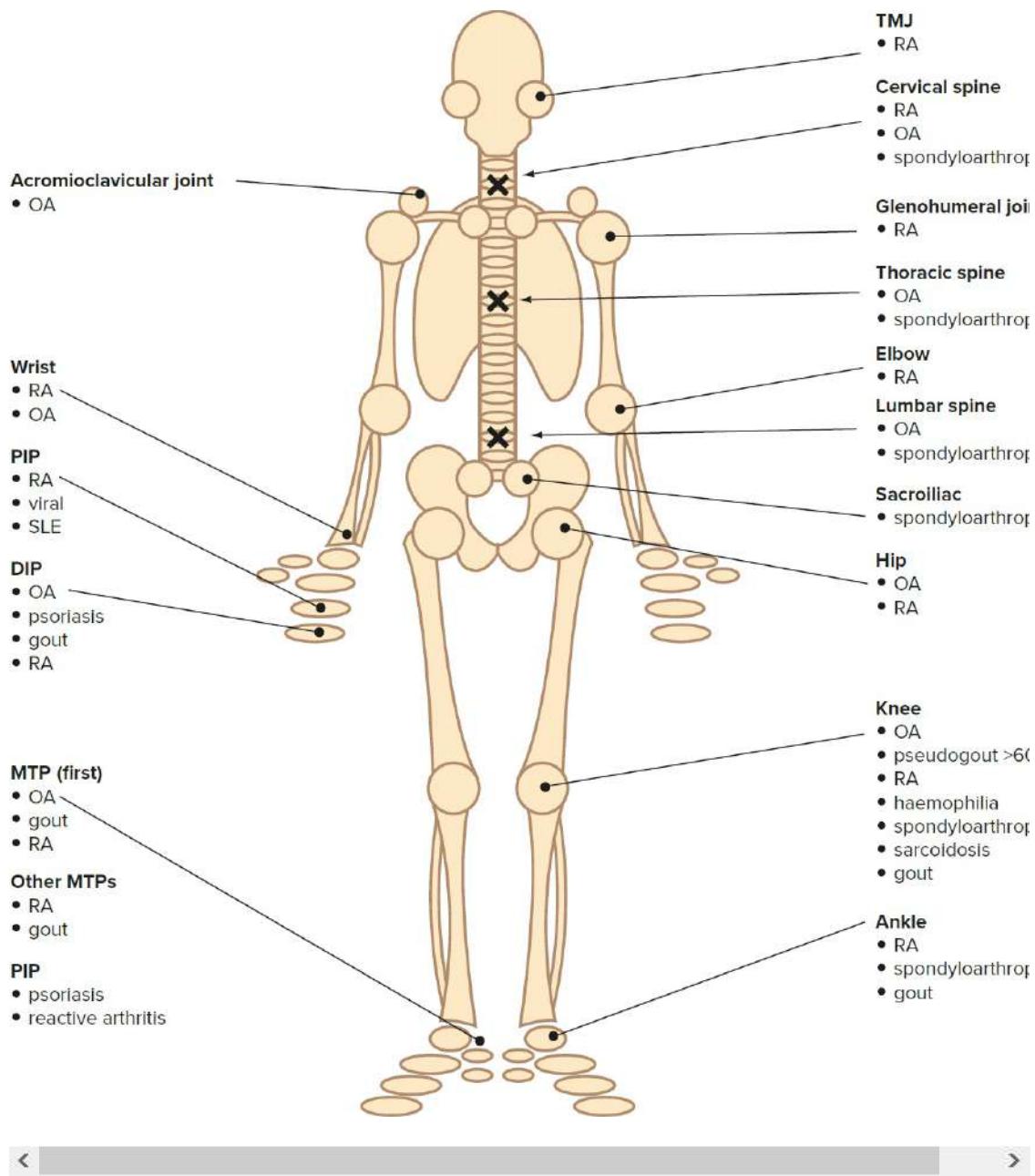


FIGURE 25.2 Joints typically affected by various arthropathies

Investigations

TABLE 25.2 lists the many possible investigations that are used to reach a diagnosis. Clinical acumen permits a judicious selection of particular tests, which is preferable to ordering an expensive battery of tests in a scattergun approach.

Table 25.2 Investigations for arthritis

Appropriate tests can be selected (thoughtfully) from the following:

- urine analysis: blood, protein, sugar
 - synovial fluid: analysis, culture
 - radiology—plain X-ray*
 - blood and other cultures
 - haemoglobin and differential WCC
 - ESR*
 - C-reactive protein
 - serum uric acid, creatinine*
 - 24-hour urinary uric acid
 - rheumatoid factor
 - anti-CCP (cyclic citrullinated peptide) antibody*
 - antinuclear antibody (screening test for SLE)*
 - dsDNA antibodies
 - extractable nuclear antigen (ENA) antibodies
 - HLA-B₂₇ (poor predictive value)
 - various specific serological tests (e.g. Australian epidemic polyarthritis, rubella, hepatitis B, Barmah Forest virus, parvovirus)
 - HIV serology
 - antistreptolysin O titre
 - streptococcal anti-DNase B
 - arthroscopy and biopsy
 - bone scan
-

*Key tests

It is important to keep in mind the many specific serological tests to detect infective causes of arthralgia. These include Australian epidemic polyarthritis, rubella, *Brucella*, hepatitis B, gonococcus, mycoplasma, HIV tests, parvovirus and Barmah Forest virus. Lyme disease is not endemic in Australian ticks,¹⁰ but testing may be relevant for those who have travelled, particularly to Europe.

In reference to viral serology, a positive immunoglobulin M (IgM) antibody test is presumptive evidence of recent infection and is likely to be of diagnostic significance in this clinical context. However, sometimes IgM antibodies can persist for months or years.¹¹ A positive IgG antibody result indicates previous exposure to the virus but a single positive titre is of no diagnostic significance. Seroconversion or at least a fourfold rise on paired sera confirms recent infection (see FIG. 25.3).

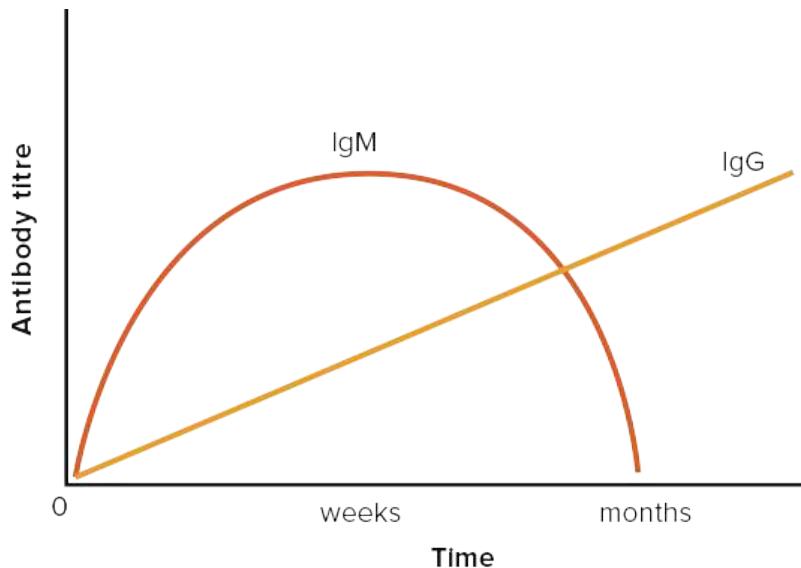
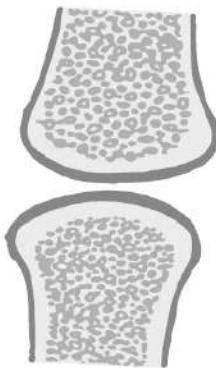


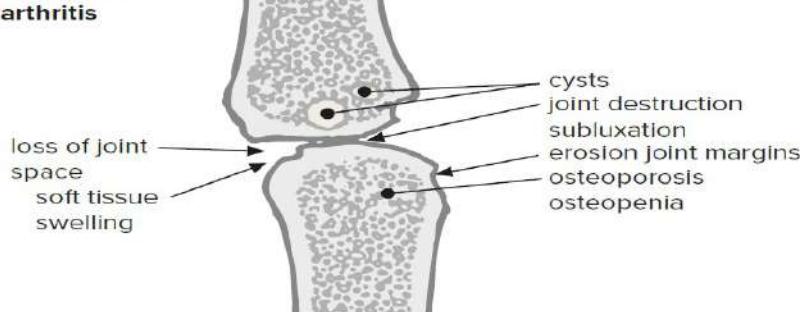
FIGURE 25.3 Time course of IgG and IgM antibodies in viral arthritis

Plain X-ray is invaluable, although in some conditions radiological changes may be apparent only when the disease is well established. Typical X-ray changes for common conditions are presented in [FIGURE 25.4](#). Arthrography has limited value in the diagnosis of polyarthritis but is very useful for specific joints such as the shoulder and the knee. Ultrasound examination for joints such as the shoulder and the hip can be very useful.

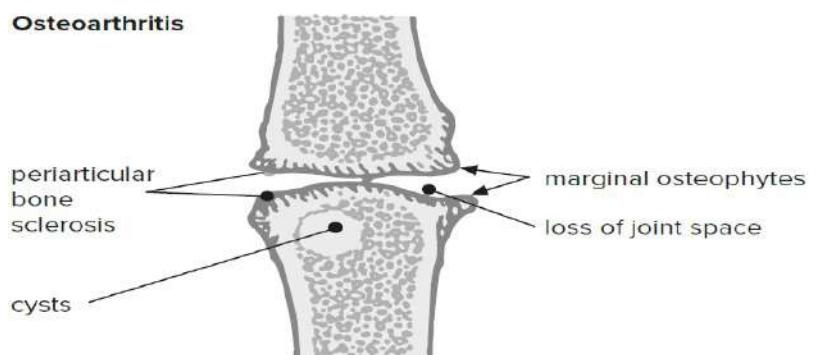
Normal joint



Rheumatoid arthritis



Osteoarthritis



Gout

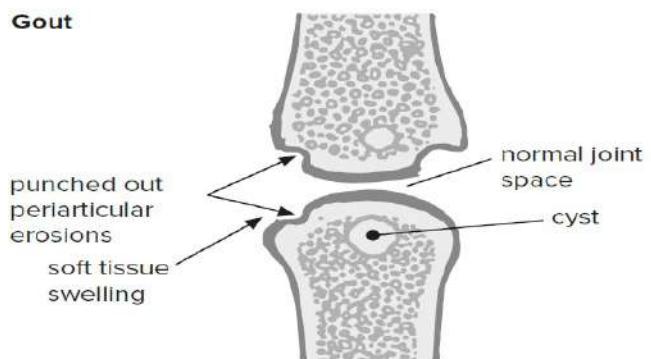


FIGURE 25.4 X-rays for common arthritic conditions: typical changes

HLA-B₂₇ should not be used for arthritis screening. It has a high sensitivity for ankylosing spondylitis, but low specificity, and should rarely be ordered.¹¹

The various immunological tests for diagnosis of the connective tissue disorders are outlined with the description of each condition. Such screening tests include:

- rheumatoid factor and anti-CCP
- antinuclear antibodies
- dsDNA antibodies

The LE cell test has been superseded by the antinuclear, dsDNA and ENA (especially Sm) antibody tests but the latter should only be performed if there is an elevated ANA test.¹¹

Arthritis in children

Arthralgia (joint pain) is a common problem in childhood and, although arthritis is rare, the complaint demands considerable respect because of the many serious problems causing it. Particular consideration should be given to rheumatic fever, septic arthritis, osteomyelitis and meningoccaemia. Rheumatic fever typically occurs in children and young adults, the first attack usually occurring between 5 and 15 years of age.

Arthritis may be part of an infectious disease such as rheumatic fever, rubella, mumps, varicella, cytomegalovirus infection, erythema infectiosum (human parvovirus), influenza, COVID-19 or other viral infection, and is occasionally encountered with Henoch–Schönlein purpura. Actually, viral arthritis is very common in children. An FBE is helpful as it may show lymphopaenia, lymphocytosis or atypical lymphocytes.¹ It is worth noting that underlying bone tumours can present as joint pain if the tumour is adjacent to the joint.

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Note: Acute-onset monoarticular arthritis associated with fever is septic until proven otherwise.

⌚ Juvenile idiopathic arthritis

JIA, also known as juvenile chronic arthritis and juvenile rheumatoid arthritis (US), is defined as a chronic arthritis persisting for a minimum of 6 weeks (some criteria suggest 3 months) in one or more joints in a child younger than 16 years.⁸ It is rare, affecting only about 1 in 1000 children, but produces profound medical and psychosocial problems.

The commonest types of JIA are oligoarticular (pauciarticular) arthritis, affecting four or fewer joints (about 50%), and polyarticular arthritis, affecting five or more joints (about 40%). Systemic onset arthritis, previously known as Still syndrome, accounts for about 10% of cases. It

is usually seen in children under the age of 5 but can occur throughout childhood. The child can present with a high remittent fever and coppery red rash, plus other features, including lymphadenopathy, splenomegaly and pericarditis. Arthritis is not an initial feature but develops ultimately, usually involving the small joints of the hands, wrists, knees, ankles and metatarsophalangeal joints.

These children should be referred once the problem is suspected or recognised. JIA is not a benign disease—50% have persistent active disease as adults.

Arthritis in perspective

Five per cent of all children complain of recurrent lower limb pain, which often awakens them from their sleep. There may be emotional factors involved and parents need appropriate reassurance. A careful history and physical examination are essential, and perhaps simple basic investigations may be appropriate. As Rudge⁸ points out, we have to be vigilant against underdiagnosis, misdiagnosis and overdiagnosis. Refer to growing pains ([CHAPTER 84](#)) and post-activity musculoskeletal pain ([CHAPTER 55](#)).

Rheumatic fever

RF is an inflammatory disorder that typically occurs in children and young adults following a group A *Streptococcus pyogenes* infection. It is common in developing countries and among Aboriginal and Torres Strait Islander people (see [CHAPTER 127](#)) but uncommon in first world countries.¹²

Clinical features

- Age 5–15 years (can be older)
- Acute-onset fever, joint pains, malaise
- Flitting arthralgia mainly in leg (knees, ankles) and arm (elbows and wrists)
- One joint settles as the other is affected
- May follow a sore throat

However, the symptoms depend on the organs affected and arthritis may be absent.

Diagnosis

Based on clinical criteria:

2 or more major criteria

or

1 major + 2 or more minor criteria
in the presence of supporting evidence of preceding Group A streptococcal (GAS) infection.

Major criteria

- Carditis
- Polyarthritis
- Chorea (involuntary abnormal movements)
- Subcutaneous nodules—in crops on elbows, wrists, knees or ankles
- Erythema marginatum—spread in a circular fashion

Minor criteria

- Fever ($\geq 38^{\circ}\text{C}$)
- Previous RF or rheumatic heart disease
- Monoarthralgia
- Raised ESR $>30 \text{ mm/hr}$ or CRP $>30 \text{ mg/L}$
- ECG—prolonged PR interval

Investigations

A selective combination of:

- FBC
- throat swab for GAS
- ESR/CRP
- streptococcal ASOT
- streptococcal anti-DNase B (repeat in 10–14 days)
- plus ECG and echocardiogram (if ↑ PR) and CXR

Treatment

- Rest in bed (if carditis, for up to 2 weeks)

- GAS sensitive antibiotics, e.g. benzathine penicillin 900 mg IM (450 mg in child <20 kg) statim or phenoxymethylenicillin 500 mg (o) bd 10 days
- Paracetamol 15 mg/kg (o) 4 hourly (max. 60 mg/kg/day); aspirin or naproxen for arthritis
- Diuretics for carditis (may be ACE inhibitor and corticosteroids)
- Prophylactic long-term penicillin

Septic arthritis

Acute sepsis (see FIG. 25.5) can affect any joint at any age, although it is more common in children. It evolves over hours or days and can rapidly destroy a joint structure. It is an emergency in the hip joint of children. Check for IV drug use. The commonest organisms are *S. aureus*, *Streptococci*, *Kingella kingae* and *N. gonorrhoea*. Diagnosis is by blood culture and synovial fluid analysis and culture. Treatment is with drainage and washout of the joint and IV followed by oral antibiotics, e.g. di/flucloxacillin. Orthopaedic referral recommended.



FIGURE 25.5 Septic arthritis in a young girl who presented with a painful swollen left knee and difficulty walking. A knee aspiration revealed turbid fluid with elevated leukocytes. The joint fluid culture grew *Staphylococcus aureus*.

Arthritis in the elderly

OA is very common with advancing age and for this reason care has to be taken not to simply attribute other causes of arthritis to OA. Other musculoskeletal conditions that become more prevalent with increasing age are:

- polymyalgia rheumatica
- Paget disease of bone
- avascular necrosis
- gout
- pseudogout (pyrophosphate arthropathy)
- malignancy (e.g. bronchial carcinoma)

Pseudogout

This crystal deposition arthropathy (chondrocalcinosis) is noted by its occurrence in people over 60 years. It usually affects the knee joint but can involve other joints.

Rheumatoid arthritis

Although it usually begins between the ages of 30 and 40 it can occur in older people, when it occasionally begins suddenly and dramatically. This is called ‘explosive’ RA and fortunately tends to respond to small doses of prednisolone and has a good prognosis.¹³ RA in the elderly can present as a polymyalgia rheumatica syndrome.

Osteoarthritis

OA is the most common type of arthritis, occurring in about 10% of the adult population and in 50% of those aged over 60.¹² It is a degenerative disease of cartilage and may be primary idiopathic or secondary to causes such as trauma and mechanical problems, septic arthritis, crystallopathy or previous inflammatory disorders, or structural disorders such as SCFE and Perthes disorder. OA of the hips and knees has a strong association with being overweight or obese.

The arthritis

Primary OA is usually symmetrical and can affect many joints. Unlike other inflammatory disease the pain is worse on initiating movement and loading the joint, and eased by rest. OA is usually associated with stiffness, especially after activity, in contrast to RA.

Joints involved

In primary OA all the synovial joints may be involved, but the main ones are:

- first carpometacarpal (CMC) joint of thumb
- first metatarsophalangeal (MTP) joint of great toe

- distal interphalangeal (DIP) joints of hands

Other joints that are affected significantly are the proximal interphalangeal joints, the knees, hips, acromioclavicular joints and joints of the spine, especially the facet joints of the cervical (C5–6, C6–7) and lumbar regions (L3–4, L4–5, L5–S1) (see FIG. 25.6).

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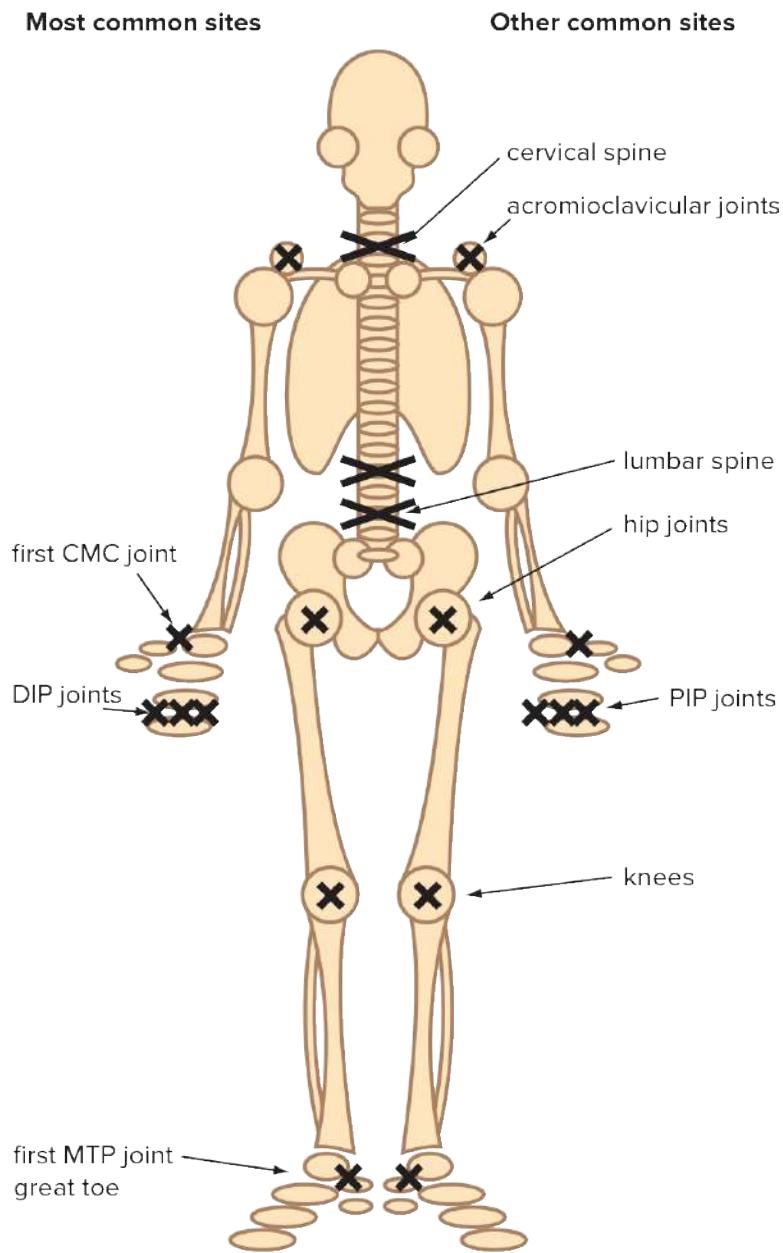


FIGURE 25.6 Osteoarthritis: typical joint distribution

Clinical features

- Pain: worse by the end of the day, aggravated by use, relieved by rest, worse in cold and damp
- Variable morning stiffness
- Variable disability

Signs

(See FIG. 25.7 .)

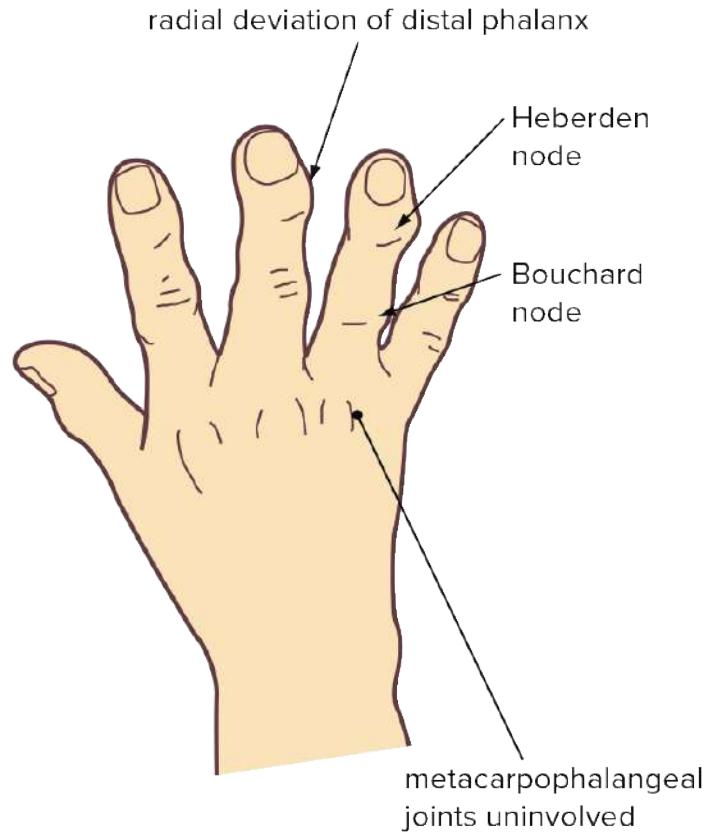


FIGURE 25.7 Typical clinical features of osteoarthritis of the hand

- Hard and bony swelling
- Crepitus
- Signs of inflammation (mild), warmth, pain
- Restricted movements; inability to weight bear
- Joint deformity

Note: There should be no systemic manifestations.

Crystal arthropathy can complicate OA, especially in the fingers of people taking diuretics (e.g. nodular gout).

Differentiation from an inflammatory arthropathy

OA does not exhibit the typical inflammatory pattern. The clinical diagnosis is based on:

- gradual onset of pain after activity (worse towards the end of the day)
- the pattern of joint involvement
- the lack of soft tissue swelling
- the transient nature of the joint stiffness or gelling
- takes <30 minutes to settle after rest while inflammatory arthritis takes at least 30 minutes

Diagnosis

The diagnosis is clinical and radiological but the degree of changes on X-ray do not always parallel levels of symptoms.¹²

X-ray findings

- Joint space narrowing with sclerosis of subchondral bone
- Formation of osteophytes on the joint margins or in ligamentous attachments
- Cystic areas in the subchondral bone
- Altered shape of bone ends

Principles of management¹⁴

- Provide explanation and reassurance, including patient education hand-outs
- Correct modifiable risk factors: obesity, injury, overuse
- Control pain and maintain function with appropriate drugs
- Suggest judicious activity, exercise and physical therapy: regular exercise has strong evidence of benefit for hip and knee OA; discourage exercise avoidance¹⁴
- For weight-bearing joints, there is evidence for weight loss of at least 5–7.5% for those with BMI >25 kg/m²
- Consider factors lowering the coping threshold (e.g. stress, depression, anxiety, overactivity)

Referral for surgery should be used judiciously, as surgeons differ in their enthusiasm for following the best independent evidence for surgical interventions. For example, the Australian Knee Society's arthroscopy 'position statement'¹⁵ recommends against arthroscopy for knee OA except in a small number of circumstances (particularly knee locking), which goes against the vast majority of those having had debridement or meniscectomies in the age of arthroscopies. Refer for consideration of joint replacement and for advice on joints causing intractable pain or disability. Hand surgery can offer good pain relief and functional improvement. Osteotomies have a limited place for a varus or valgus deformity of the knee.

Treatment (least useful interventions towards the end)¹⁴

- *Explanation.* Provide patient education and reassurance that arthritis is not the crippling disease perceived by most patients.
- *Exercise.* A graduated exercise program is essential to maintain joint function. Aim for a good balance of relative rest with sensible exercise. It is necessary to stop or modify any exercise or activity that increases the pain. Systematic reviews have found that both exercise and education may help reduce the pain and disability in people with OA of the hip or knee.¹⁶
- *Diet.* If overweight it is important to reduce weight to ideal level. Obesity increases the risk of OA of the knee approximately fourfold and weight loss may slow progression,¹⁷ otherwise, no specific diet has been proven to cause or improve OA.
- *Rest.* Prolonged bed rest is contraindicated, and exercise is important. However, rest during an active bout of inflammatory activity is reasonable as a pain reduction strategy.
- *Heat.* Recommended is a hot-water bottle or other heat pack, warm bath or electric blanket to soothe pain and stiffness. Advise against getting too cold. Do not advise using local cold packs, as they have been shown not to help.
- *Physiotherapy.* Referral should be made for specific purposes such as:
 - correct posture and/or leg length disparity (but beware of the increasing unscientific use of this term)
 - supervision of a hydrotherapy program
 - heat therapy and advice on simple home heat measures
 - teaching and supervision of isometric strengthening
 - exercises (e.g. for the neck, back, quadriceps muscle)
 - therapeutic ultrasound, kinesio taping and electrical or laser stimulation have not been demonstrated to work; discourage such practices.
- *Occupational therapy.* Refer for advice on aids in the home, more efficient performance of daily living activities, protection of joints and on the wide range of inexpensive equipment and