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2 Diagnosis, Investigations and Screening

Investigation of colorectal cancer may be considered in three distinct groups:

a) Initial diagnosis
b) Staging of colorectal cancer
c) Surveillance following completion of treatment

2.1 Initial diagnosis

2.1.1 Process of referral and investigation

Concern over a diagnosis of colorectal cancer should prompt urgent referral, in most cases within general practice through a ‘two week wait rule’. Criteria for two-week wait referral (National Institute for Health and Clinical Excellence, 2015) should be satisfied and in many institutions, patients are offered a ‘direct to test service’. Initial investigations are planned on the basis of presenting symptoms (anaemia, change in bowel habit, rectal or abdominal mass), age and co-morbidity, as well as available local expertise and resources. A typical investigation algorithm is shown in Table 2.1. Patients presenting via a non-urgent route should also be investigated along similar lines. The NICE guidance (NG 12) for suspected colorectal cancer (National Institute for Health and Clinical Excellence, 2015) recommends using faecal occult blood testing for a subset of patients without rectal bleeding. This remains contentious due to equivocal evidence in symptomatic patients. Its practical implementation remains challenging in most regions.

Patients with suspected colorectal cancer should be referred urgently to a team specializing in the management of colorectal cancer.

Recommendation grade D

2.1.2 Choice of investigations

The recent UK Special Interest Group in Gastrointestinal and Abdominal Radiology (SIGGAR) trials (Atkin et al., 2013; Halligan et al., 2013) provides level I evidence on the choice for whole colon imaging in patients with symptoms suggestive of colorectal cancer. The trial comprised a composite single endpoint incorporating:

1. CT colonography (CTC) vs double contrast barium enema (DCBE), with detection rates of cancer and large polyps (≥1 cm).
2. CTC vs optical colonoscopy (OC), powered to measure the requirement for additional colonic tests following the randomized investigation. The rationale is

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<table>
<thead>
<tr>
<th>Symptom</th>
<th>Age</th>
<th>2WW Indication</th>
<th>Please tick*</th>
<th>Direct to test</th>
<th>Results</th>
</tr>
</thead>
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<tr>
<td>Change in bowel habit</td>
<td>Over 55</td>
<td><strong>Looser and/or more frequent stools</strong>, persistently for more than 6 weeks without rectal bleeding</td>
<td></td>
<td>Colonscopy</td>
<td>Positive test (cancer) – direct referral to MDT via colorectal nurse specialists</td>
</tr>
<tr>
<td></td>
<td>Over 55</td>
<td></td>
<td></td>
<td></td>
<td><strong>Y { } N { }</strong></td>
</tr>
<tr>
<td>Rectal bleed</td>
<td>Over 40</td>
<td><strong>Change in bowel habit</strong> – rectal bleeding with a change in bowel habit to looser and/or more frequent stools persistently for 6 weeks</td>
<td></td>
<td>Colonscopy</td>
<td><strong>Y { } N { }</strong></td>
</tr>
<tr>
<td></td>
<td>Over 55</td>
<td><strong>No change in bowel habit</strong> Rectal bleeding persistently for &gt;6 weeks without a change in bowel habit &amp; without anal symptoms</td>
<td></td>
<td></td>
<td><strong>Y { } N { }</strong></td>
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<td>Mass</td>
<td>All ages</td>
<td><strong>Rectal</strong> – A definite palpable rectal (not pelvic) mass</td>
<td></td>
<td>Flexible sigmoidoscopy</td>
<td></td>
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<tr>
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<td>All ages</td>
<td><strong>Abdominal</strong> – A definite palpable abdominal mass</td>
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<td>CT colonography or colonoscopy</td>
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<td>Anaemia</td>
<td>All ages</td>
<td><strong>Men</strong> – of any age with unexplained iron deficiency 110 g/l or below</td>
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<tr>
<td></td>
<td>All ages</td>
<td><strong>Women</strong> – non-menstruating women with unexplained iron deficiency anaemia 100 g/l or below</td>
<td></td>
<td>CT colonography or colonoscopy + OGD</td>
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<tr>
<td>Any of above</td>
<td>&gt;75 years</td>
<td>As above (please tick) BUT aged &gt;75 years or symptoms not covered in above</td>
<td></td>
<td>2WW Outpatient clinic Clinic then Direct-to-Test and as above</td>
<td></td>
</tr>
</tbody>
</table>
that subsequent OC with biopsy will usually be required for CTC-suspected cancer or significant polyp, and that any benefit in avoiding an endoscopic procedure would be cancelled out if a CTC precipitates a large number of subsequent unnecessary colonoscopies. Conversely, OC is more often suboptimal, limited or incomplete in the more elderly symptomatic population, which may lead to further colonic investigations.

The principal finding was that CTC detected significantly more cancers and large polyps than DCBE (7.3% vs 5.6%; \( P = 0.039 \)). Patients randomized to CTC were diagnosed with fewer post-test colorectal cancers than DCBE during a 3-year follow up period (3/45 vs 12/85). No significant difference was found in detection rates between CTC and OC (10.7% vs 11.4%) but CTC generated more colonic investigations than OC (relative risk 3.65). Post-test colorectal cancers during 3-year follow up were 1/29 for CTC and 0/55 for OC, confirming prior meta-analysis suggesting no significant difference in sensitivity of the two tests for colorectal cancer (Pickhardt et al., 2011).

Patients with suspected colorectal cancer should be offered either optical colonoscopy or CT colonography.

**Recommendation grade A**

CT colonography should replace double contrast barium enema.

**Recommendation grade A**

### 2.1.3 Quality of investigations

Regardless of whether OC or CTC is used, certain levels of Quality Assurance should be achieved by these investigations.

- **Colonoscopy**
  Colonoscopy should be performed by an experienced colonoscopist, or a colonoscopist who has been certified by the Joint Advisory Group (JAG), or by trainees supervised by one of the above. National quality and safety standards for endoscopy have been set by the Department of Health using a Global Rating Scale (GRS) (www.grs.nhs.uk) and by the British Society for Gastroenterology (www.BSG.org.uk). The quality and safety indicators underpin the respective items using GRS.

- **CT Colonography (CTC)**
  There are several international consensus recommendations regarding acquisition of CTC images (Burling, 2010; McFarland et al., 2009; Neri et al., 2013). The examination may be performed following full purgative bowel preparation and administration of a faecal tagging agent (oral contrast medium administered several hours prior to the investigation). Some faecal tagging agents such as sodium amidotrizoate/meglumine amidotrizoate (Gastrografin) also have a laxative effect and can be used as a combined cleansing/tagging agent. Although some oral faecal tagging agents have a laxative effect, others do not, and in particularly frail patients a combination of dietary manipulation and non-laxative faecal tagging (such as low-dose iso-osmolar iodinated contrast media, or certain barium sulphate preparations) may achieve adequate image quality to exclude clinically relevant neoplasia. In all cases (with or without purgation), faecal tagging is now regarded as mandatory when performing CTC.

Mechanical insufflation with CO\(_2\) reduces patient discomfort and improves colonic distension, when compared with room air. Intravenous antispasmodic, typically hyoscine butylbromide (Buscopan), also improves distension and may reduce pain. The use of intravenous contrast medium is not essential for colonic lesion detection, but may be an efficient strategy in patients in whom the pre-test probability of either colorectal cancer or important extracolonic pathology is high, and is invariably given when whole colon imaging and staging is required following detection of a cancer where OC failed to evaluate the proximal colon.

All reporting radiologists must meet the recommended standards for competency, and be actively involved in audit of their practice and performance (British Society of Gastrointestinal and Abdominal Radiology (BSGAR), The Royal College of Radiologists, 2014). There are specific stipulations for radiologists involved in the National Bowel Cancer Screening Program (NHS Bowel Cancer Screening Programme, 2012). Radiological and endoscopic investigations should meet national standards and be subjected to regular Quality Assurance.

**Recommendation grade C**

### 2.1.4 Diagnosis of colorectal cancer

Colon cancer should ideally be confirmed histologically, but when a lesion with high probability of malignancy has been detected by CTC, histology prior to surgery is not essential if the segment cannot be reached endoscopically. Conversely, histological confirmation of malignancy should be considered mandatory in all rectal cancers when surgery might result in either a permanent stoma or an ultra-low anterior resection, or when neoadjuvant therapy is being considered. Exception to this may be a large endoluminal lesion where biopsies have been inconclusive and the lesion is not amenable to endoscopic surgery.
Preoperative histological confirmation of colonic cancer is desirable but not essential in cases with high probability of malignancy based on clinical presentation and optimal imaging.

**Recommendation grade D**

Pre-treatment histological confirmation of rectal cancer is considered essential. Exceptions should be rare and are usually large lesions not amenable to endoscopic removal and inconclusive biopsies.

**Recommendation grade D**

### 2.2 Staging of Colorectal Cancer

Staging investigations for colorectal cancer should address three areas:

a) Assessment of local disease

b) Metastatic disease

c) Synchronous colonic disease

#### 2.2.1 Assessment of local disease

Local extent of colonic disease is assessed by abdominal and pelvic computed tomography (CT) to provide information on extent of spread in relation to the bowel wall and adjacent organs. This is essential to the planning of surgery (e.g. non-anatomical resection) and to allow consideration of neoadjuvant therapy when the disease is locally very advanced or unresectable.

CT has limited ability to differentiate the individual layers of the bowel wall, making the distinction between T1 and T2 disease challenging. Although certain tumour morphology features seen on CT colonography, namely an arc-shaped or trapezoidal tumour configuration, may suggest T1 or T2 status, these are yet to be validated in a prospective, multicentre setting. Similarly, the distinction between T3 and T4 disease can be challenging because the latter may be contingent only on sub-millimetre spread through the serosa for peritonealised colon (T4a by TNM 7th edition; T4b by TNM 5th edition).

Conversely, because of the naturally high contrast at CT between the bowel wall and the adjacent fat, T3 disease can usually be discriminated from earlier-stage lesions, making the distinction between T3 and T4 disease can be challenging. Although certain tumour morphology features seen on CT colonography, namely an arc-shaped or trapezoidal tumour configuration, may suggest T1 or T2 status, these are yet to be validated in a prospective, multicentre setting. Similarly, the distinction between T3 and T4 disease can be challenging because the latter may be contingent only on sub-millimetre spread through the serosa for peritonealised colon (T4a by TNM 7th edition; T4b by TNM 5th edition).

#### 2.2.2 Metastatic disease

Colorectal cancers commonly metastasize to the liver, lungs and peritoneum. Routine staging by CT chest, abdomen and pelvis provides adequate staging in most cases. Further characterization of equivocal liver lesions may be assisted by MRI. 18-Fluorodeoxyglucose positron emission tomography/CT (18-FDG PET/CT) may be used to investigate suspicious lesions seen on CT or MRI. In addition, PET/CT provides valuable assessment to help exclude occult metastatic disease in
patients being considered for non-anatomical resection of locally advanced cancers or surgical management of liver and lung metastases, to facilitate appropriate patient selection aiming to reduce futile procedures (Adams et al., 2013).

2.2.3 Synchronous colonic disease
Synchronous colorectal cancers are present in 2–3% of patients and a further 20% have synchronous significant benign lesions (diverticular disease, colitis or polyps). Synchronous lesions should be diagnosed at colonoscopy or CTC, as this is crucial in planning complete surgical resection. The most distal significant lesion in the colon should be tattooed (Williams et al., 2013) with the proviso that the endoscopist should ensure they are in the sigmoid and not the rectum. The tattoo should be placed into a saline bleb 2–5 cm from the base of the lesion on the distal (anal) side and the estimated distance clearly documented. More than one tattoo may be applied but the endoscopist should document very carefully the number and position of tattoos in relation to the polyp or tumour. Rectal lesions should not be tattooed.

All patients with suspected colonic cancer should be staged with a CT thorax, abdomen and pelvis.

Recommendation grade B

All patients with suspected rectal cancer should be staged with a CT thorax, abdomen and pelvis. Patients being considered for curative locoregional treatment should have MRI of the pelvis.

Recommendation grade B

Synchronous lesions should be identified using colonoscopy or CTC prior to colorectal cancer resection. If complete colonoscopy is not possible, CTC should be considered. CT of the thorax can be combined with CTC to permit staging at the same hospital visit.

Recommendation grade B

2.3 Colorectal Cancer Screening

2.3.1 Screening for average risk population

2.3.1.1 NHS Bowel Cancer Screening Program (BCSP)
The NHS BCSP offers screening every 2 years to the population aged between 60–69, as over 80% of newly diagnosed colorectal cancers occur at this age and beyond. Since 2008, the screening age group has been extended to 74 years old, in accordance with the government’s strategy to improve cancer outcomes.

Screening with guaiac faecal occult blood test (FOBT) has been shown to reduce colorectal cancer mortality in four large population-based randomized control trials after more than 10 years of follow up (Table 2.2) (Hewitson et al., 2008). The results from the UK and Denmark were directly comparable. The apparently superior results in the US trial were ascribed to a far higher colonoscopy rate, which in turn was due to more frequent FOBT testing and a lower positive test threshold.

The guaiac FOBT is being replaced with the more accurate faecal immunochemical test (FIT) for colorectal cancer screening programs (Halloran et al., 2012). It has shown improved uptake and the ability to detect significantly more colorectal cancers and advanced adenomas (Digby et al., 2016; Moss et al., 2016). However, the higher uptake and test positivity is likely to stretch colonoscopy services even further (Tinmouth et al., 2015).

People with positive faecal occult blood tests (5 or 6 samples tested positive; or 1–4 samples positive, followed by any positive result on two subsequent screening kits) are offered a screening colonoscopy. This is carried out in endoscopy units by a JAG accredited BCSP colonoscopist. For individuals unwilling to have or who cannot tolerate colonoscopy, CTC could be considered. Follow up colonoscopies will be determined by the results of the index colonoscopy.

The NHS BCSP has had a positive impact on elective treatment of colorectal cancers by early cancer detection, increased use of minimally invasive techniques and reducing the need for emergency colorectal

<table>
<thead>
<tr>
<th>Study</th>
<th>Country</th>
<th>Screening frequency</th>
<th>Age range (Yr)</th>
<th>Follow up period (Yr)</th>
<th>RR</th>
<th>95% CI</th>
</tr>
</thead>
<tbody>
<tr>
<td>Funen</td>
<td>Denmark</td>
<td>Annual</td>
<td>45–75</td>
<td>17</td>
<td>0.84</td>
<td>0.73–0.96</td>
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<td></td>
<td></td>
<td>Biennial</td>
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<td></td>
<td>0.89</td>
<td>0.78–1.01</td>
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<td>Annual</td>
<td>50–80</td>
<td>18</td>
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<td>0.51–0.83</td>
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<td></td>
<td></td>
<td>Biennial</td>
<td></td>
<td></td>
<td>0.79</td>
<td>0.62–0.97</td>
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<td>Biennial</td>
<td>45–74</td>
<td>11.7</td>
<td>0.87</td>
<td>0.78–0.97</td>
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<tr>
<td>Gotheberg</td>
<td>Sweden</td>
<td>Biennial</td>
<td>60–64</td>
<td>19</td>
<td>0.84</td>
<td>0.71–0.99</td>
</tr>
</tbody>
</table>
cancer resections. It also has had a positive impact on
30-day postoperative mortality and 5-year survival rates.

2.3.1.2 NHS Bowel Scope Program
A randomized controlled trial in the UK has shown that
a ‘one-off’ screening flexible sigmoidoscopy reduces
CRC incidence and CRC-related mortality by 23% and
31% respectively (Atkin et al., 2010) after a median fol-
low up of 11 years. Results from the UK trial have
prompted the NHS to roll out a population-based pro-
gram using flexible sigmoidoscopy as screening offered
at age 55 with the option to opt in up to aged 60
(http://www.cancerscreening.nhs.uk/bowel/).

Population-based NHS Bowel Cancer Screening
Program will increase detection of early stage dis-
ease, improve cancer survival and reduce the need
for emergency surgery. Establishment of flexible sig-
moidoscopy screening is likely to reduce incidence of
distal cancers as well as reducing mortality.

Recommendation grade A

Patients diagnosed through the National Bowel
Cancer Screening Program should have a defined
pathway into their local colorectal cancer MDT.

Recommendation grade D

2.3.2 Screening for at risk population
Approximately 5% of colorectal cancers are due to the
high-risk familial conditions, Lynch syndrome and the
polyposis syndromes. It is important that these familial
syndromes are identified so that the index case and fam-
ily members can be offered appropriate management.
In a further 30% there is some familial contribution to acti-
ology, but the genetic factors involved remain unclear
and the level of risk is heterogeneous.

Families deemed to be at high risk of colorectal can-
cer should be managed through clinical genetic services,
which should provide a robust surveillance program
ensuring appropriate use of genetic testing, timely
surveillance and counseling.

2.3.2.1 Risk stratification according to family history
An accurate family history gives an empirical assessment
of risk. The site and age at diagnosis of all cancers in
family members should be documented, as well as the
presence of related features such as colorectal adeno-
as. The value of risk assessment on the basis of family
history is limited in small families and those with poorly
defined paternity. Current BSG guidelines (Cairns
et al., 2010) give a detailed evidence base for stratifying
risk level according to family history, and provide
screening recommendations (summarized in Table 2.3):

<table>
<thead>
<tr>
<th>Risk</th>
<th>Relative risk of CRC</th>
<th>Colonoscopy screening</th>
</tr>
</thead>
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<tr>
<td>Low</td>
<td>&lt;2</td>
<td>None</td>
</tr>
<tr>
<td>Low moderate</td>
<td>3–6</td>
<td>One off age 55 years</td>
</tr>
<tr>
<td>High moderate</td>
<td></td>
<td>5 yearly from 50 years</td>
</tr>
</tbody>
</table>

Low risk
Individuals in this group have one of the following:
- No confirmed family history of colorectal cancer
- No first-degree relative (i.e. parent, sibling or child) with colorectal cancer
- Only one first-degree relative with colorectal cancer, diagnosed at age 50 years or older

Low risk individuals should be reassured,
informed of the symptoms of colorectal cancer and
encouraged to participate in the NHS Bowel Cancer
Screening Program.

Recommendation grade C

Low-moderate risk
This group comprises those with one of:
- One affected relative diagnosed with colorectal cancer under 50 years
- Two affected first-degree relatives diagnosed at age 60 years or older

Low-moderate risk individuals should be offered
‘one-off’ screening colonoscopy at the age of 55 years.

Recommendation grade C

High-moderate risk
This group comprises those with one of:
- Three or more relatives with colorectal cancer in a
  first degree kinship* (but none under 50 years; a fac-
  tor which confers high risk)
- Two relatives diagnosed with colorectal cancer under
  60 years (or with a mean age at diagnosis under
  60 years) in a first degree kinship*
- Both parents diagnosed with colorectal cancer under
  60 years

* A first-degree kinship is a family group in which each
affected individual is a first degree relative of the others.
The individual seeking screening should be a first degree
relative of one of these affected relatives.

High-moderate risk individuals should be offered
colonoscopy every 5 years from the age of 50 to
75 years.

Recommendation grade C
High risk
This category encompasses Lynch syndrome and the polyposis syndromes. Criteria for inclusion include:
- Member of a family with known familial adenomatous polyposis (FAP) or other polyposis syndrome
- Member of a family with known Lynch syndrome
- Pedigree suggestive of autosomal dominantly inherited colorectal (or other Lynch syndrome-associated) cancer (Amsterdam criteria Table 2)
- Pedigree indicative of autosomal recessive inheritance, suggestive of MYH associated polyposis (MAP)

2.3.2.2 Lynch syndrome
This dominantly inherited condition is due to germline mutation in one of the mismatch repair (MMR) genes, and carries a 50–70% lifetime risk of colorectal cancer (often developing at an early age), as well as increased incidence of other cancers. The cancers are characterized by microsatellite instability (MSI) and loss of mismatch repair (MMR) protein, detectable by immunohistochemical staining. There is a preponderance of mucinous and right sided cancers compared to sporadic colorectal cancer populations. Indications for MSI testing are increasing with recent evidence supporting its use in all patients with colorectal cancer (Vasen et al., 2013). Further analysis of methylation status and BRAF mutation can distinguish sporadic cases of MSI from those with a genetic predisposition.

Full guidelines for the management of individuals with Lynch syndrome are available (Vasen et al., 2013), and anyone suspected of having it should be referred to a clinical genetics unit or specialist polyposis registry. Important points for the clinician managing these patients are:
- Genetic testing (or endoscopic screening if no mutation has been identified) should begin at around 12 years of age, unless the child is symptomatic.
- Prophylactic surgery is the mainstay of management, the type and timing determined by genotype, phenotype and personal circumstances.
- Lifelong follow-up is required, with annual surveillance of the retained rectum or ileo-anal pouch.
- Endoscopic examination of the duodenum using a side-viewing scope should start at age 30 years and continue at intervals determined by disease severity.

Annual colonoscopy should be offered to mutation carriers from diagnosis until polyp load indicates a need for surgery.

Recommendation grade B

Indicators from families where no mutation can be identified and genetic linkage analysis is not possible, should have annual surveillance from the age of 13–15 until age 30 years, and every 3–5 years thereafter until age 60.

Recommendation grade B

For individuals who have undergone either total colectomy and ileorectal anastomosis or proctocolectomy and ileal pouch-anal anastomosis, lifelong endoscopic annual surveillance of the retained rectum or anorectal cuff is recommended.

Recommendation grade B

Upper gastrointestinal endoscopy (using a side-viewing endoscope) at intervals determined by...
Spigelman stage for mutation carriers is recommended from age 30 years.

Recommendation grade C

2.3.2.4 MYH-associated polyposis
This recently recognized polyposis syndrome has considerable overlap with FAP, although the polyp burden is often considerably less, and extra-intestinal manifestations are less frequent (Cairns et al., 2010). Management of the colon and rectum is essentially the same as FAP, although some individuals with a light polyp burden can be managed endoscopically, without the need for prophylactic surgery.

Colonoscopic surveillance is recommended every year from age 25 years for individuals who are bi-allelic MYH carriers (or homozygous carriers of other BER gene defects).

Recommendation grade C

Upper gastrointestinal endoscopy at 3–5 year intervals is recommended from age 30 years.

Recommendation grade C

2.3.2.5 Serrated polyposis
This poorly understood condition is characterized by the development of multiple hyperplastic polyps, sessile serrated polyps and serrated adenomas (Edelstein et al., 2013). There appears to be an inherited element, but the gene(s) responsible have not yet been identified and genetic testing is not available.

Annual surveillance colonoscopy is recommended. First-degree relatives should have five yearly screening from the age of 30 years.

Recommendation grade C

2.3.2.6 Peutz-Jeghers syndrome
This dominantly inherited syndrome results in characteristic gastrointestinal polyps, particularly in the small bowel, and peri-oral pigmentation (Cairns et al., 2010). The risk of gastrointestinal, and other cancers, is substantial. These patients should be referred to a clinical genetics unit or polyposis registry.

A baseline colonoscopy and upper GI endoscopy (OGD) is recommended at age 8 years. If significant polyps are detected, endoscopy should be repeated every 3 years. If no significant polyps are present at baseline endoscopy, routine surveillance is repeated at age 18, or sooner should symptoms arise, and then every 3 years.

Recommendation grade C

Small bowel screening using video capsule endoscopy (VCE) should be performed every 3 years, from age 8 years, or earlier if the patient is symptomatic. Magnetic resonance enterography (MRE) or CT enterography are reasonable alternatives in adult patients but CT enterography is not favoured in children due to radiation exposure.

Recommendation grade C

2.3.2.7 Juvenile polyposis
This is a rare syndrome due to mutation in the SMAD4 or BMPR1A genes, which results in typical polyps, particularly in the large bowel (Cairns et al., 2010). The risk of colorectal cancer is in the range 10–40%, and that of gastric cancer around 25%. Management is usually by endoscopic polypectomy, with surveillance intervals of 1–3 years, depending on polyp burden. Surgery (colectomy, proctocolectomy or gastrectomy) is sometimes required.

Those with a SMAD4 mutation frequently also have hereditary haemorrhagic telangectasia, so bleeding or anaemia may not be due to the juvenile polyps, and appropriate investigations to exclude associated arteriovenous malformations should be done before any general anesthetic. These patients should be referred to a clinical genetics unit or polyposis registry.

Colonoscopic surveillance for at risk individuals and mutation carriers is recommended every 1–2 years from age 15–18 years, or earlier if the individual is symptomatic up to age 70 years.

Recommendation grade C

Upper gastrointestinal surveillance is recommended every 1–2 years from age 25 years.

Recommendation grade C

Conflicts of interest
None of the authors have any conflicts of interest to declare.

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References