

ASCS, KL - 5th Mar 2010

Surveillance Recommendations for Patients with FAP

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Reference

COLORECTAL CANCER SCREENING

Guidance on gastrointestinal surveillance for hereditary non-polyposis colorectal cancer, familial adenomatous polyposis, juvenile polyposis, and Peutz-Jeghers syndrome

M G Dunlop

Gut 2002;**51**(Suppl V):v21-v27



Large bowel surveillance for FAP family members

In a minority of FAP families a mutation cannot be identified and so annual flexible sigmoidoscopy should be offered to at risk family members from age 13–15 years until age 30, and at three to five year intervals thereafter until age 60 years. Surveillance might also be offered as a temporary measure for people with documented APC gene mutations but who wish to defer prophylactic surgery for personal reasons. Such people should be offered six monthly flexible sigmoidoscopy and annual colonoscopy but surgery should be strongly recommended before 25 years. After colectomy and ileorectal anastomosis, the rectum must be kept under review at least annually for life because the risk of cancer in the retained rectum is 12%–29%.^{16 61 62} The anorectal cuff after restorative proctocolectomy should also be kept under annual review for life.

Recommendation Grade: B



Upper GI surveillance in FAP

- To combat the substantial risk of upper gastrointestinal malignancy in FAP after prophylactic colectomy, upper gastrointestinal surveillance is recommended. While the presence of gastroduodenal polyposis is well recognised, there are few published studies on which to gauge the potential benefit of surveillance. However, the approach seems reasonable and three yearly upper gastrointestinal endoscopy is recommended from age 30 years with the aim of detecting early curable cancers. Patients with large numbers of duodenal polyps should undergo surveillance yearly.
Recommendation Grade: B

Extracolonic manifestations

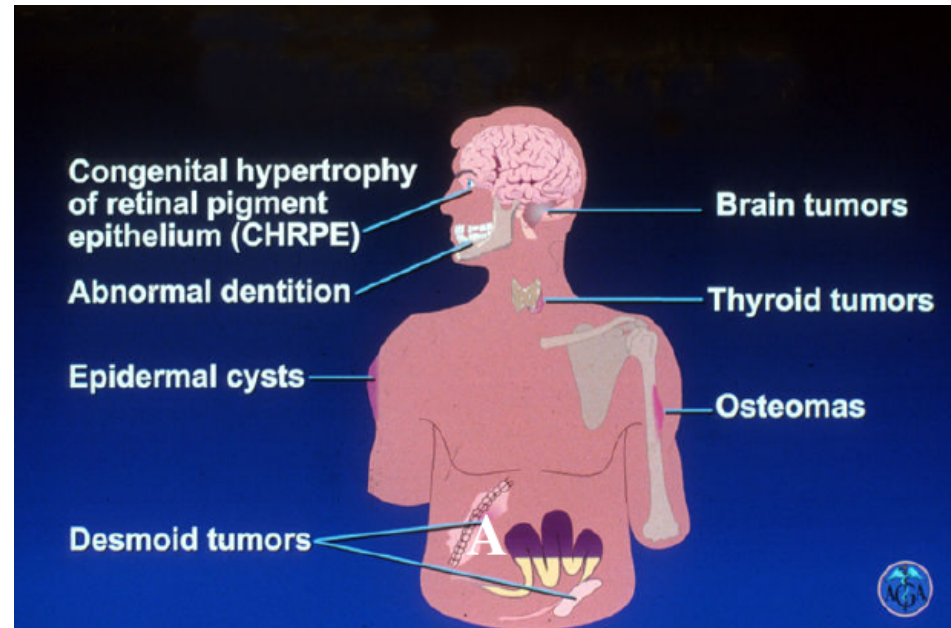
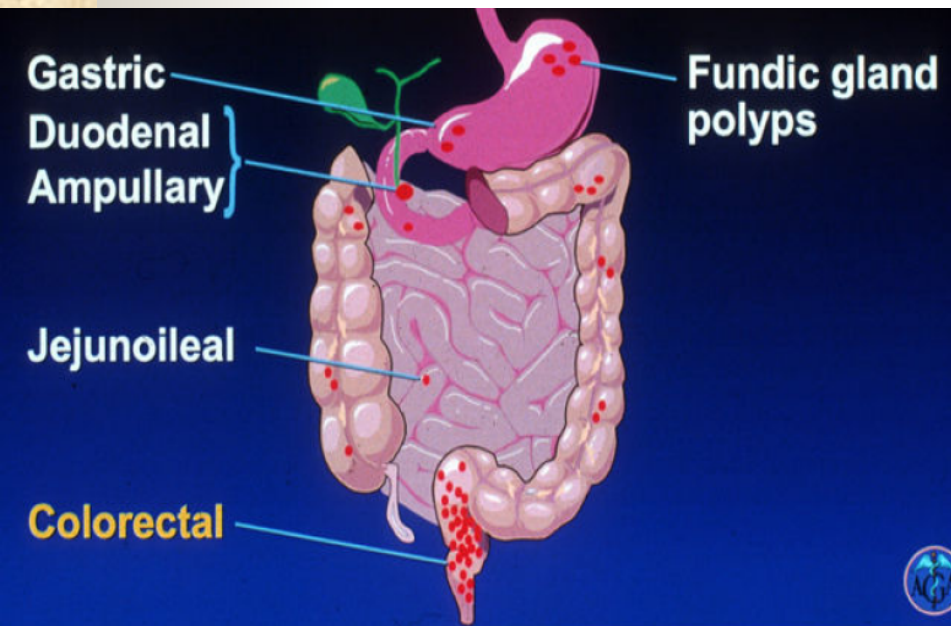


Table 1 Extracolonic Manifestations of Adenomatous Polyposis Coli (APC) Mutations

	Incidence	Significance
Duodenal adenomas	80–90%	12% Risk of malignancy
Gastric hamartomas	50–70%	Must exclude gastric adenoma
Desmoids	12–35%	27% Risk of complications
Hepatoblastoma	1%	Most common before age 2 years
Osteomas	80%	Usually < 1 cm in size
CHRPE	60%	Marker for screening purposes
Thyroid cancer (papillary)		Predilection for women

CHRPE, congenital hypertrophy of the retinal pigment epithelium.



Surveillance Recommendations

- Surveillance for FAP patients and family members
- Post-colectomy surveillance



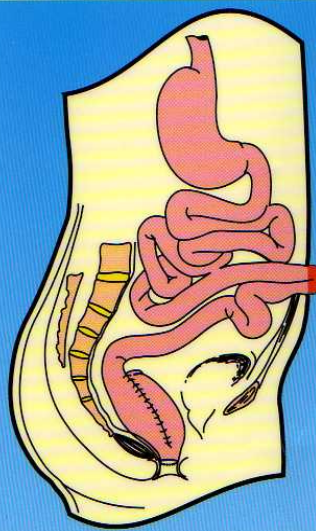
Reference

CLINICS IN COLON AND RECTAL SURGERY
VOLUME 21, NUMBER 4 2008

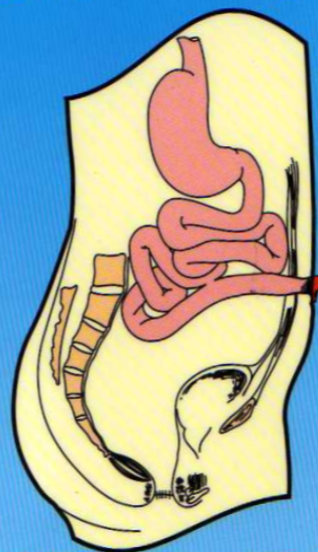
Table 5 Surgical Options for Polyposis Syndromes

Procedure	Indications
Abdominal colectomy with ileorectostomy	aFAP by family history, clinical exam, or genetic testing; mild disease (FAP or MAP) as manifested by fewer than 20 polyps in the rectum and less than 1000 overall
Proctocolectomy with ileal-pouch anal anastomosis	Severe disease (FAP or MAP) by family history, clinical exam, or genetic test results; cancer in the colon or mid-upper rectum; risk of desmoid tumor by personal or family history or <i>APC</i> mutation between codons 1403–1578.
Proctocolectomy with ileostomy	When ileal-pouch anal anastomosis contraindicated because of anal sphincter dysfunction or technical problems; low rectal cancer which precludes sphincter preservation.

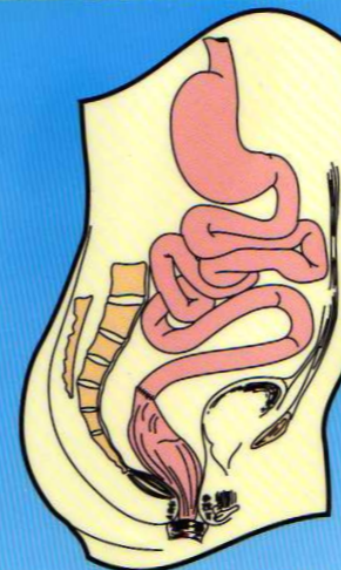
aFAP, attenuated familial adenomatous polyposis; FAP, familial adenomatous polyposis; MAP, *MYH* associated polyposis; *APC*, adenomatous polyposis coli.



Restorative procto-colectomy.
With or without loop ileostomy.



Procto-colectomy with permanent
ileostomy



Colectomy with ileo-rectal
anastomosis



Post-colectomy surveillance

- Routine postop – physical and psychological adaptation
- Oncologic issues – if cancer has developed
- TAC/IRA – rectal remnant
- RPC/IPAA – ileal pouch
- PC/ileostomy – distal ileum
- No one surveillance strategy is appropriate in all patients.
- At least once yearly endoscopy appears reasonable



J Gastrointest Surg

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ORIGINAL ARTICLE

Long-Term Outcome of Metachronous Rectal Cancer Following Ileorectal Anastomosis for Familial Adenomatous Polyposis

Tomohiro Yamaguchi & Seichiro Yamamoto &
Shin Fujita & Takayuki Akasu & Yoshihiro Moriya

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IRA – metachronous rectal cancers

- 59 patients underwent IRA
- 17 (28.8%) developed metachronous rectal cancers, average 8.8 years (range 1.3 -23.3 years)
- 11(65%) required radical surgery
- 8 with Tis-T1 invasion had undergone endoscopic surveillance at intervals of 6 months to 1 year after IRA, but the other three T3 patients did not undergo surveillance for more than 2 years



Pouch surveillance

- Persistent rectal mucosa distal to IPAA
- Risk of adenomas and carcinomas within ileal pouch
- Initial endoscopy anywhere from 3 months to 1 year after pouch creation; continued yearly or every other year for life.
- ?Indigocarmine
- Endoscopic polypectomy for polyps >5mm.

Upper GI surveillance in FAP


Spigelman's Staging System for Duodenal Polyposis and Current Surveillance Recommendation

Variable	Scoring Criteria		
	Points		
	1	2	3
Number of polyps	1-4	5-20	> 20
Size of polyps (mm)	1-4	5-10	> 10
Histology	Tubular	Tubulo-villous	Villous
Dysplasia	Low-grade	-	High-grade

Staging and Surveillance Recommendations

Stage	Points	Surveillance Recommendations*
0	0	Endoscopy every 4 years
I	1-4	Endoscopy every 2-3 years
II	5-6	Endoscopy every 2-3 years
III	7-8	Endoscopy every 6-12 months. Consider surgical consultation
IV	9-12	Endoscopy every 6-12 months. Surgical treatment indicated

*Endoscopy with direct and side-viewing scope is advocated.



High resolution endoscopy and the additional value of chromoendoscopy in the evaluation of duodenal adenomatosis in patients with familial adenomatous polyposis. Dekker et al.

Endoscopy 2009 Aug;41(8):666-9.

N=43

Results: Upstaged 12%

CONCLUSION:

Ampullary adenomas are commonly found in FAP and are best visualized using a side-viewing endoscope. Therefore, a combination of forward-viewing HRE and chromoendoscopy with side-viewing endoscopy for the periampullary region seems useful for surveillance of duodenal adenomatosis in FAP.



Desmoid Tumours

- Intraabdominal 80%, abdominal wall 10 to 15% and extraabdominal 5%.
- Risk factors
 - Previous surgery, tend to develop within 5 years.
 - Female
 - Family history (> 30% of affected family members)
 - Presence of osteomas
 - Patients with mutations toward the 3' end of the APC gene, mutations beyond codon 1444 specifically
- MRI may be helpful in predicting active growth when high-signal intensity is seen on the T2-weighted images.



Desmoid Tumours

- MRI may be helpful in predicting active growth when high-signal intensity is seen on the T2-weighted images.⁴⁶



Table 2 Screening Recommendations for Patients at Risk for a Polyposis Syndrome

For the patient whose family medical history suggests FAP or whose <i>APC</i> mutation is between codons 200–2500	Flexible sigmoidoscopy or colonoscopy begin at puberty, then flexible sigmoidoscopy annually
For the patient whose family medical history suggests aFAP or MAP, or whose <i>APC</i> mutation is before codon 157 or after codon 2500	Colonoscopy beginning at age 18–21 years; Every 1–2 years
For all patients with a known or suspected <i>APC</i> mutation	Esophagogastroduodenoscopy every 1–5 years according to Spigelman stage, beginning at 20 years

FAP, familial adenomatous polyposis; APC, adenomatous polyposis coli; aFAP, attenuated familial adenomatous polyposis; MAP, *MYH* associated polyposis.



Small bowel adenomas

- Small bowel adenomas occur and are rarely a problem, although cancers have been reported.
- Prevalence of small bowel polyps related to the duodenal polyposis stage and subject's age. The location, size and number of polyps progress as duodenal polyposis stage advances.
- Capsule endoscopy (CE) surveillance of asymptomatic FAP patients is presently unproven and should not replace upper endoscopy.
- CE underestimated the number of small bowel polyps and did not reliably detect large polyps (especially periampullary) but identified polyps in areas not accessible by standard endoscopy.



Other extra-gastrointestinal tumors

- Rare
- Papillary Thyroid Cancer (2%) - 20-30y.o.
- Pancreatic adenocarcinoma (2%)
- Gastric adenocarcinoma (0.5%) - 3x higher in Asians
- Hepatoblastoma in children less than 5 years of age (<1%).
- These should be searched for during routine clinical and imaging follow-up.



Effectiveness of screening

- Screening asymptomatic FAP patients for all of its possible manifestations is unproven.
- For children, to identify hepatoblastoma, some recommend annual alpha-fetoprotein and abdominal ultrasound from birth until the age of 10 years.
- For all FAP patients, an annual physical examination should include an evaluation for soft tissue or bone lesions, and a thorough thyroid examination with a low threshold for performing an ultrasound of any suspicious lesion.



Symptomatic patients

- Abdominal pain, new onset diabetes mellitus or acute pancreatitis
- CT abdomen to rule out desmoid tumors of the mesentery or pancreatic adenocarcinomas (or IPMT).
- MRI indicated if CT not diagnostic - outline vascular involvement of a desmoid tumor and may predict its growth
- CT brain can also be used in symptomatic patients to search for a medulloblastoma.



Long-term compliance with endoscopic surveillance advice for FAP. Colorectal Disease

(epub 10 Jul 2009)

N=328

Result: One in five at-risk for FAP and one in four with a retained rectum are under-compliant with screening advice. Low self-efficacy, non-use of sedatives during surveillance and pain after surveillance are negatively associated with compliance behavior in at-risk individuals.

Recommendation: Sedatives should be patient-tailored for FAP-individuals undergoing colorectal cancer surveillance and adequate pain medication provided after endoscopy, based on experiences and needs of the patients.