Risk Management for Familial Adenomatous Polyposis

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Link to information for people and families

Familial adenomatous polyposis (FAP) due to a germline mutation in the APC gene is an autosomal dominant condition.

Target group

- known APC mutation carrier
- individuals meeting clinical criteria for FAP, who have uninformative genetic testing
- first degree relatives of an individual with proven FAP (i.e. individuals at 50% risk of FAP)

Exclusion criteria

- multiple mixed polyposis phenotype with no APC or MYH mutation identified

Lifetime risk of cancer/other neoplasms*

Tumour	Familial adenomatous polyposis	General population to age 85 yrs
Colorectal cancer	>95%[1]*	8% **
Duodenal or periampullary cancer	4.5% by age 57[2]	<2%
Other neoplasms		
Desmoid tumours	12% [3] increased risk with surgery and pregnancy and family history of desmoid tumours[4]	<1%
Duodenal adenomas	90%[2]	0.3- 4.6%
Gastric fundic gland polyps	55%[1]	6%
This data is from classical FAP *Penetrance of colorectal adenoma is almost 100% by age 40 in the Classical FAP[1]		
**This data does not take into account the impact of surveillance. Data Source: NSW Central Cancer Registry 2008 final dataset and NSW Health Outcomes Information Statistical Toolkit (HOIST).		

Cancer risk management guidelines

Tumour type	Recommendations	
Colorectal	Surgical	▪ **prophylactic colectomy** is standard of care and is strongly recommended ▪ timing of surgery: typically late teens, exact timing to be determined by patient and number of polyps present
	Surveillance	▪ prior to **prophylactic colectomy**: From age 12-15yrs, flexible sigmoidoscopy annually or biennially ▪ annual colonoscopy once polyps start developing until **prophylactic colectomy** performed
	Risk-reducing medication	▪ no evidence that Non-Steroidal Anti-Inflammatory Drugs (NSAIDs) prevent colorectal cancer (CRC) in this situation
Duodenum or periampulla	Surgical	▪ consider duodenectomy for Stage IV adenomas[5]

	Surveillance	▪ from age 25: Upper gastrointestinal endoscopy frequency dependent on **Spigelman criteria**[5] ▪ endoscopic surveillance should include side viewing endoscopy to ensure the medial wall and papilla are assessed ▪ patients with adenomatous lesions larger than 10mm to be reviewed in centres with both endoscopic and surgical expertise in duodenal and ampullary adenoma management
Desmoid tumours	Risk-reducing medication	▪ no evidence that NSAIDs prevent desmoid tumours in this situation

Evidence for risk management guidelines

Recognition of the disorder and screening of at risk relatives with sigmoidoscopy/colonoscopy and ultimately prophylactic colectomy results in a significant reduction in CRC diagnosis. CRC rates for non-screened (initial family case) versus screened at risk relatives fell from 68% to 3% in one registry study[6]

These guidelines are based on:

2008 Familial aspects of bowel cancer guidelines

2008 Clinical practice guidelines for GPs

Cancer Institute NSW FAP information

Support and information

First degree (blood) relatives (parents/brothers/sisters/children) are at 50% risk of having this condition. First degree relatives should be referred to a local Family Cancer Clinic.

Website Resources

Centre for Genetics Education NSW Health
The Association of Genetic Support of Australiasia INC (AGSA)
Hereditary Cancer Registry

References

1. **Burn, J., P. Chapman, J. Delhanty, et al. 1991. "The UK Northern region genetic register for familial adenomatous polyposis coli: use of age of onset, congenital hypertrophy of the retinal pigment epithelium, and DNA markers in risk calculations." J Med Genet 28(5):289-296.**
2. **Bulow, S, Bjork, J, Christensen, I, et al. 2004. "Duodenal adenomatosis in familial adenomatous polyposis." Gut 53: 381-386**
3. **Clark, S. K. and R. K. Phillips. 1996. "Desmoids in familial adenomatous polyposis." Br J Surg 83(11):1494-1504.**
4. **Klemmer, S., L. Pascoe and J. DeCosse. 1987. "Occurrence of desmoids in patients with familial adenomatous polyposis of the colon." Am J Med Genet 28(2):385-392.**
5. **Groves, C. J., B. P. Saunders, A. D. Spigelman, et al. 2002. "Duodenal cancer in patients with familial adenomatous polyposis (FAP): results of a 10 year prospective study." Gut 50(5):636-641.**
6. **Bulow, S. 2003. "Results of national registration of familial adenomatous polyposis." Gut. 52(5):742-746.**

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