ACG GUIDELINE Highlights

Diagnosis & Management of Adenomatous Colorectal Polyposis Syndromes

Concept & Content: Carl Kay, MD & Andrew M. Moon, MD, MPH, FACG | **Reviewers:** Gautam Mankaney, MD & Carol A. Burke, MD, MACG

Informative Genetic Testing Results and Management

TRIGGERS FOR GERMLINE TESTING in individuals with adenomatous colorectal polyposis

- **≥20 cumulative adenomas** [Strong / Low]
- **Extracolonic FAP features** (Desmoid, Bilateral/multifocal CHRPE, Hepatoblastoma, CMTC thyroid) [Strong / Low]
- **10–19 cumulative adenomas** [Conditional / Low]

MULTIGENE PANEL TESTING (MGPT)

Germline MGPT is recommended with the following minimum set of genes (APC, AXIN2, BMPR1A, EPCAM, GREM1, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, POLD1, POLE, PMS2, PTEN, SMAD4, and STK11); results of MGPT and other family history guide patient management.

FAP	AFAP	MAP	PPAP	Other Rare Syndromes
APC	*APC*	*MUTYH* (biallelic)	*POLD1* / *POLE*	*NTHL1* | *AXIN2* | *MLH3* | *MBD4* | *MSH3*
Age 10–15 | q1–3 yr	Age 18–20 | q1–3 yr	Age 18–20 | q1–3 yr	Age 25–30 | q1–3 yr	Age 25–30* | q1–3 yr
Age 20–25	Age 20–25	Age 30–35	Age 25–30	No routine
Yes · By age 18	Yes · By age 18	No routine	Multisystem	Multisystem

★ **Many syndromes carry extraintestinal manifestations** requiring specialized screening and surveillance. *Consult guideline for syndrome-specific recommendations.*
*Colonoscopy start age or 5 yrs before youngest FDR polyposis dx / 10 yrs before FDR CRC diagnosis, whichever is earlier.

Negative Germline Test

NEGATIVE GPV ON MGPT = COLONIC POLYPOSIS OF UNCERTAIN ETIOLOGY (CPUE)* → Consider *APC* mosaicism testing
Manage CPUE according to cumulative number of colorectal & duodenal adenomas; If *APC* mosaic, manage according to FAP
**Also referred to as Multiple Colorectal Adenomas (MCRA), Idiopathic Adenomatous Polyposis (IAP)*

Endoscopic and Surgical Management

Gastric Polyposis – Report Polyposis Features

Finding	Management*
Number, size, and location of FGPs	Resect if ≥10 mm
Number, size, and location of gastric adenomas	Resect
High risk features: White mucosal patch (WMP), polypoid mounds, carpeting	
WMP, polypoid mounds	Resect; Consider high-risk center
Multi-focal HGD or adenocarcinoma	Gastrectomy

Duodenal Polyposis – Report Spigelman Features

Feature	1 pt	2 pts	3 pts
# Polyps	1–4	5–20	>20
Size (mm)	1–4	5–10	>10
Histology	TA	TVA	VA
Dysplasia	LGD	—	HGD

Spigelman Stage: 0 (0 pt); I (1–4 pts); II (5–6 pts); III (7–8 pts); IV (9–12 pts)
Interval based on Stage; consider duodenectomy for Stage IV

★ **Upper GI Surveillance:** Interval determined by severity of gastric or duodenal polyposis, whichever is more advanced. *See guideline for details.

Colorectal Polyposis

Colonoscopy	Resect adenomas ≥10 mm, ≥4 mm if feasible. Multi-focal HGD, endoscopically unmanageable burden, symptoms associated with polyposis, or CRC warrant surgical referral.
Surgery	Surgical approach individualized based on rectal polyp burden, colon polyp burden, desmoid risk, and patient preferences—Consult guideline.

AFAP = attenuated FAP
CHRPE = congenital hypertrophy of retinal pigment epithelium
CMTC = cribriform-morular thyroid carcinoma
FAP = familial adenomatous polyposis
FDR = first-degree relative
FGP = fundic gland polyp
GPV = germline pathogenic variant
HGD = high-grade dysplasia
LGD = low-grade dysplasia
MAP = *MUTYH*-associated polyposis
PPAP = polymerase proofreading-associated polyposis
SS = Spigelman stage
TA = tubular adenoma
TVA = tubulovillous adenoma
VA = villous adenoma

Gautam Mankaney, MD; Gregory E. Idos, MD; Elena M. Stoffel, MD, MPH; Jennifer M. Weiss, MD; Shailja Shah, MD, MPH; Emad Qayed, MD, MPH, FACG; Mary Pat Harnegie, MLIS; Carol A. Burke, MD, MACG | *The American Journal of Gastroenterology* | DOI: 10.14309/ajg.0000000000004105

AJG The American Journal of GASTROENTEROLOGY

→ READ THE GUIDELINE: **bit.ly/acg-polyposis-syndromes-26**