Colonic Polyposis Syndromes Testing Algorithm

Adenomatous polyps

Patient’s clinical and/or family history* is suggestive of a specific syndrome:
- Lynch syndrome (MLH1, MSH2, MSH6, and PMS2 genes)
- Familial adenomatous polyposis syndrome (APC gene)
- MYH-associated polyposis syndrome

Non-adenomatous or mixed (adenomatous and non-adenomatous) polyps

Patient’s clinical and/or family history* suggests multiple differential diagnoses

Consider a multi-gene panel: HCRC / Hereditary Colon Cancer Multi-Gene Panel

<10 polyps

Refer to: Hereditary Nonpolyposis Colorectal Cancer Testing Algorithm

≥10 polyps

Order appropriate single gene test:
- APCZ / APC Gene, Full Gene Analysis
- MUTYH / MYH Gene Analysis for Multiple Adenoma, Y165C and G382D

Patient’s clinical and/or family history* is suggestive of a specific syndrome:
- PTEN hamartoma tumor syndrome (PTEN gene)
- Cowden syndrome (PTEN gene)
- Bannayan-Riley-Ruvalcaba syndrome (PTEN gene)
- Proteus syndrome (PTEN gene)
- Proteus-like syndrome (PTEN gene)
- Peutz-Jeghers syndrome (STK11 gene)
- Juvenile polyposis syndrome (SMAD4 and BMPR1A genes)
- AXIN2-related oligodontia-colorectal cancer syndrome

Order appropriate single gene test:
- PTENZ / PTEN Gene, Full Gene Analysis
- STKZ / STK11 Gene, Full Gene Analysis
- SMADZ / SMAD4 Gene, Full Gene Analysis
- BMPRZ / BMPR1A Gene, Full Gene Analysis
- AXINZ / AXIN2 Gene, Full Gene Analysis

*For patients with a positive family history, refer to: Full Gene Analysis/Multi-Gene Panels versus Familial Mutation Targeted Testing Algorithm.