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)
- MUTHY/MYH-associated polyposis syndrome
- AXIN2-related oligodentia-colorectal cancer syndrome

<10 polyps

Refer to: Lynch Syndrome Testing Algorithm

Order appropriate single gene test:
- APCZ / APC Gene, Full Gene Analysis
- MYHZ / MUTHY Gene, Full Gene Analysis
- AXINZ / AXIN2 Gene, Full Gene Analysis

≥10 polyps

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

Nonadenomatous (ie, hamartomas, serrated, hyperplastic, juvenile, etc) or mixed (adenomatous and nonadenomatous) polyps

Patient's clinical and/or family history* is suggestive of multiple differential diagnoses
- 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)
- MUTHY/MYH-associated polyposis syndrome

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)
- MUTHY/MYH-associated polyposis 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
- MYHZ / MUTHY 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.