

Gene(s)	Syndrome	Major Linked cancers	Possible Interventions to be Discussed
BRCA 1 BRCA 2	Hereditary Breast-Ovarian cancer	1. Breast 2. Ovary 3. Pancreas 4. Prostate	1. Mammogram, MRI, Preventive mastectomy (optional) 2. Transvaginal ultrasound, Serum CA-125, Preventive removal of tubes and ovaries (recommended) 3. Investigational imaging 4. Rectal examination Serum PSA
TP53	Li-Fraumeni Syndrome	1. Breast 2. Sarcoma 3. Leukemia, Brain, Adrenal and Others	1. Mammogram, MRI, Preventive mastectomy (optional) 2. Investigational total body MRI 3. Not standard screening available
PTEN	Cowden Syndrome	1. Breast 2. Uterus 3. Thyroid 4. Colon 5. Kidney	1. Mammogram, MRI, Preventive mastectomy (optional) 2. Transvaginal ultrasound, Endometrial biopsy, Preventive hysterectomy (optional) 3. Thyroid examination and ultrasound 4. Colonoscopy 5. Investigational imaging
CDH1	Hereditary Diffuse Gastric cancer	1. Stomach 2. Breast	1. Upper endoscopy, Preventive gastrectomy 2. Mammogram, MRI, Preventive mastectomy (optional)
STK11	Peutz-Jeghers Syndrome	1. Small bowel 2. Pancreas 3. Colon 4. Breast 5. Ovary	1. Small bowel imaging by X-ray or endoscopy 2. Investigational imaging 3. Colonoscopy 4. Mammogram, MRI, Preventive mastectomy (optional) 5. Transvaginal ultrasound
ATM		1. Breast 2. Pancreas (uncertain risk)	1. Mammogram, Possible MRI 2. Investigational imaging
CHEK2		1. Breast 2. Colon	1. Mammogram, Possible MRI 2. Colonoscopy
PALB2		1. Breast 2. Pancreas (uncertain risk)	1. Mammogram, Possible MRI 2. Investigational imaging
RAD51C RAD51D		1. Breast (RAD51D) 2. Ovary	Management guidelines not clearly established
BRIP1 BARD1		1. Breast	Management guidelines not clearly established
NBN		1. Breast	Management guidelines not clearly established
MLH1 MSH2 MSH6 PMS2 EPCAM	Lynch Syndrome	1. Colon 2. Uterus, Ovary 3. Stomach and small bowel 4. Urinary track	1. Colonoscopy 2. Transvaginal ultrasound, Endometrial biopsy, Serum CA-125, Preventive hysterectomy with tubes and ovaries removed (recommended) 3. Upper endoscopy 4. Urinalysis or urine cytology
APC	Familial Adenomatous Polyposis	1. colon 2. Small bowel	1. Colonoscopy, Preventive colectomy if enough polyps 2. Upper endoscopy
MUTYH	MYH-Associated Polyposis	1. Colon 2. Small bowel	1. Colonoscopy, Preventive colectomy if enough polyps 2. Upper endoscopy
SMAD4 BMPR1A	Juvenile Polyposis	1. Colon 2. Stomach, Small bowel	1. Colonoscopy, Preventive colectomy if enough polyps 2. Upper endoscopy and small bowel imaging
CDKN2A (P16) CDK4	Familial atypical mole-melanoma (FAMM)	1 Melanoma 2. Pancreas	1. Total body skin exam 2. Investigational Imaging