

Gene	Syndrome	Society	Summary of Societal Guidelines (age to begin) ¹⁵ <i>Management could include any of the following. Please refer to published guidelines for complete management recommendations</i>
<i>BRCA1</i>	Hereditary Breast and Ovarian Cancer Syndrome (HBOC)	NCCN, CAPS	breast awareness (18); clinical exams (25); breast MRI (25); mammogram (30); mastectomy (individualized); salpingo-oophorectomy (35-40 <i>BRCA1</i> or 35-45 <i>BRCA2</i>); male breast surveillance (35); CA-125 and transvaginal ultrasound (30); breast/ovarian risk reduction agents (individualized); prostate screening (40); consider whole-body skin exams (<i>BRCA2</i> only, individualized) ¹ ; pancreatic surveillance endoscopic ultrasound/magnetic resonance cholangiopancreatography (EUS/MRCP) and/or other clinical trials for screening (<i>BRCA2</i> only, individualized) ³
<i>BRCA2</i>		NCCN, CAPS	
<i>MLH1</i>	Lynch Syndrome / Hereditary Non-Polyposis Colorectal Cancer (HNPCC)	NCCN, CAPS, USMSTF	colonoscopy (20-25), hysterectomy, salpingo-oophorectomy (after completion of child-bearing); endometrial sampling, CA-125 and transvaginal ultrasound, EGD (30-35), urinalysis (25-35), use of aspirin as a risk reduction agent ^{2,4} ; colorectal surgical evaluation ² ; pancreatic surveillance (EUS/MRCP) and/or other clinical trials for screening ³
<i>MSH2</i>			
<i>MSH6</i>			
<i>PMS2</i>			
<i>EPCAM</i>			
<i>APC</i>	Familial Adenomatous Polyposis (FAP)/ Attenuated FAP (AFAP)	NCCN	colonoscopy/sigmoidoscopy (10-15), colorectal surgical evaluation, use of aspirin as a risk reduction agent, upper endoscopy and MRI/CT (20-25), thyroid exam/ultrasound (late teens), abdominal palpation with consideration of MRI/CT ²
<i>MUTYH</i> (2 copies)	<i>MUTYH</i> -Associated Polyposis (MAP) Cancer Risk	NCCN	colonoscopy (25-30), colorectal surgical evaluation, upper endoscopy (30-35) ²
<i>MUTYH</i> (1 copy)	<i>MUTYH</i> -Associated Cancer Risk	NCCN	colonoscopy (40) ²
<i>CDKN2A</i> (<i>p16INK4a</i>)	Melanoma-Pancreatic Cancer Syndrome (M-PCS)	MGC, CAPS	skin protection, skin exams(10), ^{5,6} pancreatic surveillance (EUS/MRCP) and/or other clinical trials for screening ³
<i>CDKN2A</i> (<i>p14ARF</i>)	Melanoma Cancer Syndrome (MCS)	MGC	skin protection, skin exams(10) ^{5,6}
<i>CDK4</i>			
<i>TP53</i>	Li-Fraumeni Syndrome (LFS)	NCCN	general comprehensive clinical exam (childhood), breast awareness (18), clinical breast exams (20-25), whole body MRI (individualized), breast MRI (20), mammography (30), mastectomy, colonoscopy (25), skin exams, physical/neurological exams, additional individualized organ-targeted surveillance ¹
<i>PTEN</i>	PTEN Hamartoma Tumor Syndrome (PHTS)	NCCN	breast awareness (18), clinical exams (25), breast MRI (30-35), mammography (30-35), mastectomy (individualized), endometrial biopsy/ultrasound (30-35), hysterectomy, thyroid ultrasound (at time of PHTS diagnosis), colonoscopy (35), renal ultrasound (40) ¹

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American Urological Association (AUA), International Cancer of the Pancreas Screening (CAPS), Hereditary Haemorrhagic Telangiectasia Foundation (HHT), Melanoma Genetics Consortium (MGC), National Comprehensive Cancer Network (NCCN), US Multi-Society Task Force on Colorectal Cancer (USMSTF)

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STK11	Peutz-Jeghers Syndrome (PJS)	NCCN, CAPS	mammography and breast MRI (25), colonoscopy and upper endoscopy (late teens), CT or MRI enterography (8-10), pancreatic surveillance (EUS/MRCP) and/or other clinical trials for screening (30-35), pelvic exam/pap smear/transvaginal ultrasound (18-20), testicular exams (10) ^{2,3}
CDH1	Hereditary Diffuse Gastric Cancer (HDGC)	NCCN	breast awareness (18); clinical exams (25); breast MRI (25); mammogram (30); mastectomy (individualized), endoscopy with biopsy, gastrectomy (18), colonoscopy (40), treat for Helicobacter pylori infection if present ^{7,8,9}
BMPR1A	Juvenile Polyposis Syndrome (JPS)	NCCN	colonoscopy and upper endoscopy (15), monitor for rectal bleeding and/or anemia ^{2,10}
SMAD4	Juvenile Polyposis Syndrome (JPS) & Hereditary Hemorrhagic Telangiectasia (HHT)	NCCN, HHT	colonoscopy and upper endoscopy (15), ² monitor for rectal bleeding and/or anemia, ¹⁰ brain MRI, contrast echocardiogram, and chest CT may be recommended ¹¹
PALB2	PALB2-Associated Cancer Risk	NCCN	mammography and breast MRI (30), ¹ consider mastectomy based on family history, ¹ male breast screening, ¹ pancreatic surveillance (EUS/MRCP) and/or other clinical trials for screening, ³ mastectomy ¹
CHEK2	CHEK2-Associated Cancer Risk	NCCN	mammography and breast MRI (40), ¹ consider mastectomy based on family history, ¹ male breast screening, ¹ colonoscopy (40) ²
ATM	ATM-Associated Cancer Risk	NCCN, CAPS	mammography and breast MRI (40), ¹ pancreatic surveillance (EUS/MRCP) and/or other clinical trials for screening ³
NBN	NBN-Associated Cancer Risk	NCCN, AUA	individualized breast screening, ¹ prostate screening (45) ^{13,14} , mammography and consider breast MRI with contrast (40) ¹
BARD1	BARD1-Associated Cancer Risk		individualized breast screening/risk reduction ^{1,12}
BRIP1	BRIP1-Associated Cancer Risk	NCCN	bilateral salpingo-oophorectomy, ¹ individualized breast and ovarian screening/risk reduction ^{1,12}
RAD51C	RAD51C-Associated Cancer Risk	NCCN	bilateral salpingo-oophorectomy, ¹ individualized ovarian screening/risk reduction ¹
RAD51D	RAD51D-Associated Cancer Risk	NCCN	bilateral salpingo-oophorectomy, ¹ individualized ovarian screening/risk reduction ¹
POLD1	Polymerase Proofreading-associated Syndrome (PPAS)	NCCN	colonoscopy (25-30), colorectal surgical evaluation ²
POLE	Polymerase Proofreading-associated Syndrome (PPAS)	NCCN	colonoscopy (25-30), colorectal surgical evaluation ²
GREM1	Hereditary Mixed Polyposis Syndrome (HMPS)	NCCN	colonoscopy (25-30), colorectal surgical evaluation ²

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- In many cases, the ages to begin an intervention should be younger if indicated by the patient's family history.



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