

APPENDIX 3. MANAGEMENT OF MUTATION CARRIERS

Genes	Interventions	Action: Initiation and Frequency
Breast CA: <i>BRCA1 & BRCA2</i>	<p>Surveillance: Breast</p> <ul style="list-style-type: none"> Clinical breast exam Mammography MRI and ultrasound <p>Surveillance: Ovarian</p> <ul style="list-style-type: none"> CA125 & transvaginal ultrasound <p>Surgery</p> <ul style="list-style-type: none"> Prophylactic mastectomy, prophylactic oophorectomy <p>Chemoprevention</p> <ul style="list-style-type: none"> Tamoxifen & raloxifene (patients are encouraged to enrol in study protocols) <p>Patient Counselling</p> <ul style="list-style-type: none"> Lifestyle: diet, exercise, alcohol intake Symptoms of endometrial cancer 	<ul style="list-style-type: none"> Every 6 months starting at age 25 or when carrier status identified, whichever is earlier (Grade I Recommendation) Annually starting at age 30 (Grade I Recommendation) Annually if surveillance required before age 30. After age 30, use as necessary to complement mammography, based on breast density (Grade A Recommendation) Not routinely recommended but, if used, every 6 months starting at age 30–35 (Grade I Recommendation) Consider (Grade A Recommendation) Consider. Discuss enrolment in clinical trials. <i>BRCA1</i>: Grade I Recommendation <i>BRCA2</i>: Grade B Recommendation Discuss (Grade I Recommendation) Educate
FAP: <i>APC</i>	<p>Surveillance</p> <ul style="list-style-type: none"> Flexible sigmoidoscopy Colonoscopy <p>Surgery</p> <ul style="list-style-type: none"> Colectomy 	<ul style="list-style-type: none"> Every 1–2 years beginning at age 10–12 (subsequent colonoscopy) Every 1–2 years once polyps detected If colectomy delayed more than a year after polyps emerge, annual colonoscopy
HNPCC: <i>MLH1, MSH2, MSH6, PMS1, PMS2</i>	<p>Surveillance</p> <ul style="list-style-type: none"> Colonoscopy <p>Surgery</p> <ul style="list-style-type: none"> prophylactic colectomy—<i>controversial</i> <p>Chemoprevention</p> <ul style="list-style-type: none"> NSAIDs <p>Patient Counselling</p> <ul style="list-style-type: none"> Symptoms of endometrial cancer 	<ul style="list-style-type: none"> Starting at age 20–25 or 5 years younger than earliest diagnosis in family (Grade I Recommendation) Subsequent colonoscopy every 1–2 years (Grade B Recommendation) May recommend if many polyps or difficult colonoscopy or recurrent colorectal cancer Studies ongoing Educate

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