Patient name: CGU Number:

Breast Guidelines

For risk categorisation: calculate 10 year risk using age at beginning of decade of true age eg. from 40 for 46yr old, 50 for 53yr old, 30 for <39yr old. Use lifetime risk (calculate from current age) for information: not for screening recommendations.

Risk category	Lifetime risk from 20 yrs	10 yr risk	*Gene change probability*	Age and frequency
Population	*<17%*	**<3% (40-50)**		3yrly mammogram 50-70, >70 on request
Moderate	*17-30%*	**3-8% (40-50)**		Annual mammogram 40-49
High	*>30%*	**8-20% (40-50)** **>8% (50-60)**		Annual mammogram 40-59
Very high **BRCA/PALB2 at risk (untested)**	*>40%*		*>30%* *BRCA/PALB2*	Annual MRI 30-49, (from 25; if 8%10 year risk from 25) Annual mammogram 40-49 Proof of mutation needed for increased screening after 50
BRCA, PALB2, STK11, CDH1, PTEN carrier			*100%*	Annual MRI 25-49 Annual mammogram 40-71 Annual mammogram after 71 requested annually (self referral) Refer for screening whether affected or not, and if patient opts for risk reducing mastectomy
TP53 at risk			*>30% TP53*	Annual MRI 20-70 (no mammography)
TP53 carrier			*100% TP53*	Annual MRI 20-70 (no mammography)
ATM homozygote				Annual MRI 25+ (no mammography)

For probability and risk estimate calculations DCIS should be considered as a breast cancer 10 years later. Risk should be assessed using IBIS or Boadicea/CANRISK models preferably.

TP53 at risk/carriers: Discuss each case with Consultant Geneticist. Non-breast cancer screening subject to local availability; to be guided by TP53 consensus screening guidelines.

Genetic testing for breast and ovarian cancer families

Refer to the test directory R207 (ovarian only) or R208 (breast and ovarian cancer). Testing can be offered by mainstream colleagues.

If testing on tissue undertaken this will not include MLPA; please discuss families with a negative result at round table.

Chemoprevention

Risk	Menopausal	Uterus	Offer	Exclusions/Advice
High/Very high	pre	yes	Tamoxifen 5 yrs	Cease 2 months before conceiving Not suitable if had BSO
High/Very high	post	yes	Raloxifene 5yrs or Anastrazole 5 yrs	Suitable if post-menopausal BSO
High/Very high	post	no	Tamoxifen/Raloxifene 5yrs or Anastrozole 5yrs	Suitable if post-menopausal BSO

Tamoxifen/Raloxifene should not be prescribed if history or risk of endometrial cancer or thromboembolic disease.
Should be stopped after 5 years, 2 months before trying to conceive and 6 weeks before elective surgery.
Anastrozle not suitable if personal history of osteoporosis.
Risks and benefits should be discussed, including side effects and alternatives eg. surveillance and risk reducing surgery.
Local arrangements for starting chemoprevention will vary.

Ovarian guidelines

If BRCA1/2, BRIP1 or RAD51C/D alteration carriers, discuss TAHBSO; no proven benefit to screening. Refer to local Gynae Oncologist to discuss options.

Prostate guidelines

If BRCA2 alteration carrier, or BRCA1 with significant family history of prostate cancer; refer to urologist from age 40 for co-ordination of prostate screening; unless different local arrangement.
BRCA1 carriers refer to GP from age 40 for co-ordination of prostate screening.

Pancreatic guidelines

For diagnostic genetic testing refer to test directory R367.
If patients have Peutz Jeghers syndrome discuss with consultant.
If BRCA1/BRCA2/PALB2, Lynch Syndrome or CDKN2A mutation plus at least one FDR with confirmed pancreatic cancer refer to EUROPAC.
(NICE guidelines for risk management are controversial, under review)

Patient name: CGU Number:

Bowel Guidelines

When calculating risk FDR can be related to each other in sequence, rather than each being a FDR of the proband. i.e. an affected mother and an affected maternal aunt would be equivalent to an affected mother and an affected brother of the proband. 2 FDR can also include both parents of a proband.

Risk category	Screening	Age and frequency
Population/Average	National Bowel Screening	FIT/FOB testing 2 yearly from 60 Bowel endoscopy once age 55
Moderate - One FDR diagnosed with CRC under 50 years, or - Two FDRs diagnosed with CRC at any age	Colonoscopy	Once age 55
High At least three affected FDRs with CRC diagnosed at any age, across at least two generations~~,~~	Colonoscopy	5 Yearly from age 40-75
Lynch-like based on MMR deficient IHC on a bowel tumour but no germlne mutation	Colonoscopy	2 yearly from 25 Bring to round table to discuss testing options

If multiple polyps but no causative mutation; refer to BSG guidelines for screening recommendations.

For diagnostic testing guidelines refer to test directory R211

Bethesda Criteria: To guide testing on retrospective samples. If IHC not previously done on colorectal or endometrial tumours where one of the below criteria is met, offer IHC

- Colorectal or uterine cancer diagnosed below 50 years of age
- More than one primary Lynch spectrum tumour in an individual, any age
- Colorectal cancer in more than two FDRs, any age

For multiple polyps offer R211 according to test directory criteria.
If polyps are hamartomatous, juvenile, Peutz-Jegher or serrated/multiple-hyperplastic, send to consultant for review.

Gene Carrier Management Guidelines:

For mutation carriers refer to BSG guidelines and for Lynch Syndrome refer to CGG Lynch gene specific guidelines.
(Note that BSG guidelines give unclear advice regarding MUTYH screening; heterozygote 'carriers' should not be offered increased screening)
Gene carriers anywhere in the West Midlands can be offered referral to specialist MDT clinic at QE led by Prof Andrew Beggs. If GP-referred then referral can be made direct to Prof Beggs, if referred by a colorectal specialist then ask referrer to consider referral-on to Prof Beggs.