

# **Protocol 2**



## BRCA1 and BRCA2 mutation testing guidelines<sup>1</sup>

## 1) has bilateral BC and both cancers diagnosed < 50 yrs 2) has triple negative BC diagnosed < 50 yrs Woman with breast cancer who 3) has OC 4) has bilateral BC and a relative with BC < 60 yrs 5) has a relative with BC and both diagnosed < 45 yrs 6) has relatives with cancer and a Manchester Score ≥15 1) Has serous or endometrioid OC diagnosed <65 yrs<sup>2</sup> B Woman with 2) has BC ovarian cancer who 3) has a relative with OC or MBC 4) has relatives with cancer and a Manchester Score ≥ 15 1) has a relative with OC or MBC Male with 2) has relatives with cancer and a Manchester Score ≥ 15 **breast cancer** who 1) has relatives with breast and/or ovarian cancer and a Manchester Score ≥ 17 (see FAQ for full eligibility) Individual unaffected with cancer who

### Results

BRCA1/2 mutation identified: see Protocol 3

**BRCA1/2** mutation not identified: see Protocol 1 for breast screening recommendation

**BRCA1/2 variants/VUS identified:** CIGMA report will be issued for internal results\*. For external results email: vus@icr.ac.uk

#### Notes

<sup>1</sup>The above criteria relate to a full gene screen of *BRCA1* and *BRCA2*, individuals of Ashkenazi Jewish heritage may be eligible for founder mutation testing.\*

<sup>2</sup>Currently eligible for testing within Mainstreaming Cancer Genetics programme.\*

### Key

**Relative** = first degree or second degree relative only, except when calculating a Manchester score\*. Female relatives through an intervening male shift up one degree of relationship\*

**BC** = breast cancer **MBC** = male breast cancer **OC** = ovarian cancer **Triple negative breast cancer** = breast tumour negative for oestrogen receptor (ER), progesterone receptor (PR) and HER2 Expression

**CIGMA**=Clinical Impact of Genetic Mutation Analysis

Manchester Score*	;
Cancer, age at diagnosis	Score
♀ Breast Cancer, <30	11
♀ Breast Cancer, 30-39	8
♀ Breast Cancer, 40-49	6
♀ Breast Cancer, 50-59	4
♀ Breast Cancer, > 59	2
∂ Breast Cancer, <60	13
♂ Breast Cancer, > 59	10
Ovarian Cancer, <60	13
Ovarian Cancer, >59	10
Pancreatic Cancer	1
Prostate Cancer, <60	2
Prostate Cancer, >59	1