Who to refer?

Based on FH e.g. in one stop

The following women should be referred to Clinical Genetics for an assessment about their eligibility for additional breast screening. A woman with:

- A first degree relative (FDR) with breast cancer under the age of 40
- A first degree relative with male breast cancer
- Two first degree relatives with breast cancer
- One first and one second degree relative with breast cancer
- One first degree relative with bilateral breast cancer
- One relative with breast cancer and one with ovarian cancer (one must be first degree)
- Two relatives with ovarian cancer
- Three or more relatives with breast or ovarian cancer

Notes

1. If the consultee has breast cancer and residual breast tissue, count as a FDR

2. An affected female second degree relative through a male relative is equivalent to an FDR

In a nutshell – one breast >40 ***OR*** *one ovarian cancer doesn't meet referral criteria – everything else does*

Who to refer?

New cancer diagnosis

Does the individual with breast cancer (<u>including high grade DCIS</u>) meet one of the following criteria?

- Breast cancer <40 years, OR
- HER2 positive breast cancer, age 31-35 years, OR
- Bilateral breast cancer, both < 60 years, OR
- Triple negative breast cancer < 60 years, OR
- Male/assigned male at birth and breast cancer at any age, OR
- High grade non-mucinous ovarian cancer at any age, OR
- Breast cancer <45 years and a first degree relative (FDR) with breast cancer < 45 years, OR
- Ashkenazi Jewish ancestry, OR
- 1 or more grandparents from Westray (Orkney) or Whalsay (Shetland)

OR MANCHESTER SCORE 15+

Risk groups

Population	Moderate	High	Very High
	family history	family history	gene carriers
	Screening; *(chemoprophylaxis)*	Screening; Chemo-prophylaxis; *(Surgical risk reduction)*	Screening; Chemo-prophylaxis; Surgical risk reduction

Aims of genetic testing after bc diagnosis

Short term

- To know whether the patient carries a 'very high' risk breast cancer gene change
- To guide risk estimates of developing a second separate breast cancer over their lifetime
- May influence primary surgical and radiotherapy decisions
- Avoidance of radiotherapy in *TP53* carriers

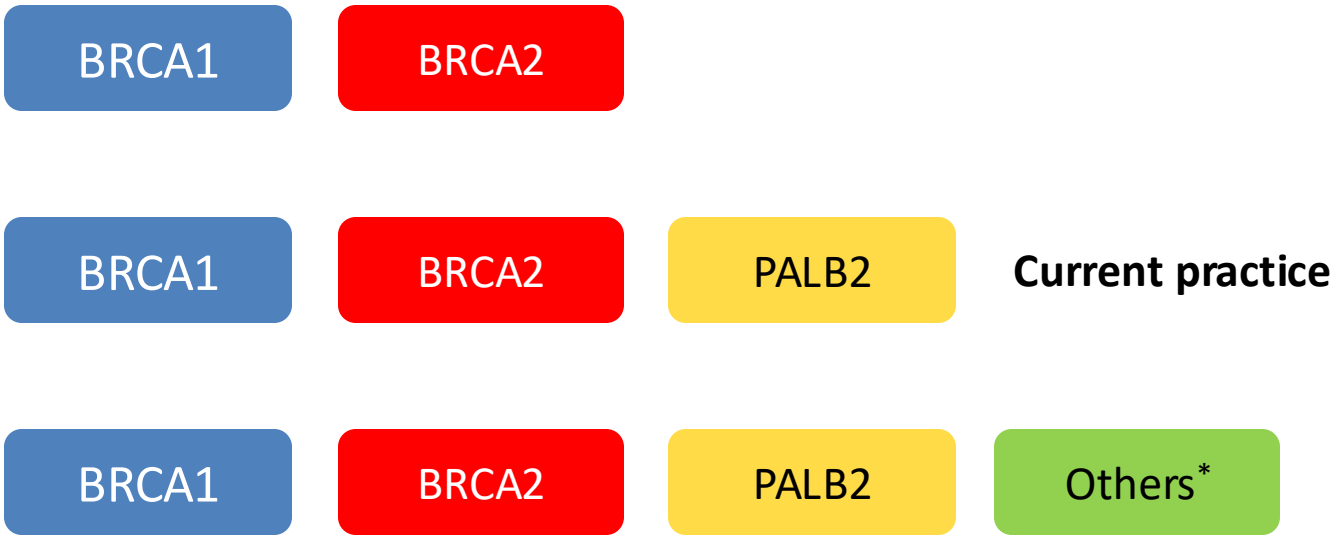
Longer term

- Is there a high risk of associated cancers for which screening or risk reducing options are available?
- To guide information and advice to other family members

Future

- To guide chemotherapeutic options

Genetic testing in breast cancer



*ATM, CHEK2, PTEN, TP53, STK11

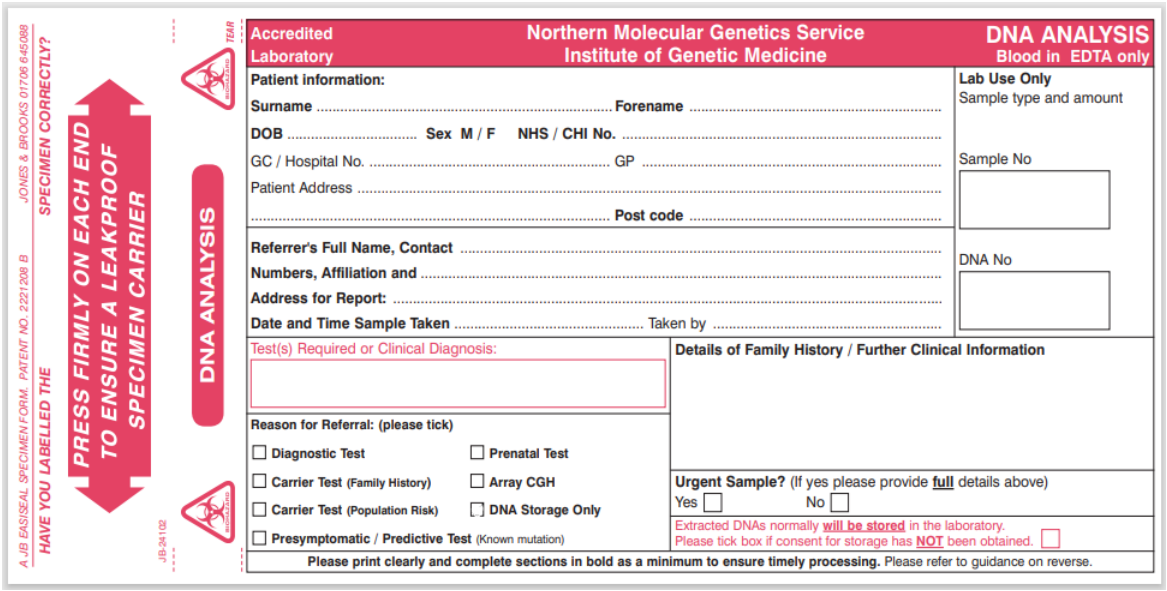
DNA storage

DNA can be extracted from a blood sample and stored for many years in our lab

Then potentially used in the future (with patient's explicit consent) to activate specific genetic tests

Useful for urgent samples and/or for patients who might prefer a telephone appointment with us; or for terminally ill patients to benefit their families.

Need approx. 3mls purple (EDTA) tube in the form below – **need to tell patient this is not being tested – just stored**



A JB EASISEAL SPECIMEN FORM. PATENT NO. 2221208 B JONES & BROOKS 01706 645088

HAVE YOU LABELLED THE SPECIMEN CORRECTLY?

PRESS FIRMLY ON EACH END TO ENSURE A LEAKPROOF SPECIMEN CARRIER

TEAR

DNA ANALYSIS

JB-2410/2

Accredited Laboratory

Northern Molecular Genetics Service
Institute of Genetic Medicine

DNA ANALYSIS
Blood in EDTA only

Patient information:

Surname ..... Forename .....

DOB ..... Sex M / F NHS / CHI No. .....

GC / Hospital No. ..... GP .....

Patient Address .....

..... Post code .....

Referrer's Full Name, Contact .....

Numbers, Affiliation and .....

Address for Report: .....

Date and Time Sample Taken ..... Taken by .....

Lab Use Only
Sample type and amount

Sample No

DNA No

Test(s) Required or Clinical Diagnosis:

Details of Family History / Further Clinical Information

Reason for Referral: (please tick)

☐ Diagnostic Test ☐ Prenatal Test

☐ Carrier Test (Family History) ☐ Array CGH

☐ Carrier Test (Population Risk) ☐ DNA Storage Only

☐ Presymptomatic / Predictive Test (Known mutation)

Urgent Sample? (If yes please provide full details above)
Yes ☐ No ☐

Extracted DNAs normally will be stored in the laboratory.
Please tick box if consent for storage has NOT been obtained. ☐

Please print clearly and complete sections in bold as a minimum to ensure timely processing. Please refer to guidance on reverse.