Interactive pathway for gBRCA testing in breast cancer

Knowing the BRCA status of breast cancer patients helps to inform their surgery and treatment options. A pathway, produced and funded by AstraZeneca as part of a steering committee, was reviewed and agreed in collaboration with UK Breast Cancer Group (UKBCG), Association of Breast Surgery (ABS), and UK Clinical Genetics Group (UKCGG). This interactive version has been adapted from that resource.

Click on each of the five steps to follow the testing process:

Imaging/biopsy | Pre-surgical MDT meeting | Genetic/molecular testing | Post-surgical MDT meeting | Treatment decision

Take action | Hover for more information

Imaging/biopsy

Imaging/biopsy +/- family history and two-week diagnosis and staging (Day 0–14)

Breast unit referral received

Appointment at breast clinic
- Imaging +/- biopsy
- Take family history

Patient investigates and documents family history if not sufficiently known

Imaging +/- biopsy results analysed

Patient provides information on family history

No cancer - patient discharged

Refer to clinical genetics if R208 criteria met

Pre-surgical MDT meeting

Treatment planning and genetic/molecular testing (Day 15–17)

Pre-surgical MDT: discuss imaging/biopsy results *(and family history if provided by patient at this stage)* and **agree treatment**

R208 eligibility identified

Appointment to inform patient of diagnosis/treatment

Testing and treatment proceed in parallel

R208 eligibility NOT identified

Appointment to inform patient of diagnosis/treatment

Possible post-MDT investigations

Neoadjuvant treatment and/or surgery if eligible

NHSE target TAT: 42 days from receipt at GLH to result for non-urgent standard results[1]

Genetic/molecular testing

Genetic/molecular testing and treatment initiation *(neoadjuvant or surgery)* (Day 17)

Identify R208- or R444.1- eligible patient

Inform patient

Second appointment *(as needed, patient-dependent)*

Patient receives and reviews information on genetic testing

Document informed consent

Request test

Blood draw

In England - regional GLH lab or designated LGL

In Scotland / Wales - regional genomics labs

Contact your local GLH for details of test request forms and where to send samples here

Blood sent to appropriate genomics laboratory

Blood sample received by laboratory

Test completion

Analysis and confirmation by clinical scientist

Report genetic testing result to:
- **Clinician** (likely to be surgeon/CNS),
- **Medical oncologist** (consultant), and
- **Patient** (if agreed during informed consent)

Germline pathogenic variant identified (e.g. g*BRCA*m)

No germline pathogenic variant identified

Referral to clinical genetics if relevant family history or variant of uncertain significance identified requiring further assessment

Post-surgical MDT meeting

Adjuvant treatment initiation and genetic/molecular testing

Post-surgical MDT

End of treatment - patient discharged to open-access follow-up

R208 or R444.1 eligibility identified

R208 or R444.1 eligibility NOT identified

Adjuvant treatment

Follow-up

End of treatment - patient discharged to open-access follow-up

R208 or R444.1 eligibility identified

R208 or R444.1 eligibility NOT identified

Continue with standard of care

Treatment decision

Treatment decision, monitoring and genetic counselling

Discuss result/treatment at next scheduled appointment

Appointment with requesting and/or treating clinician to discuss result

Treating clinician to make treatment and prescribing decision

Refer to clinical genetics

Monitor therapy

Scan or click the QR codes below to explore the Genomic Test Directory for Rare and Inherited Disease, where you'll find patient eligibility criteria for R208, R444.1, and the Scottish criteria:

NHS England[2]

National Genomic Test Directory website: check out the **Rare and inherited disease eligibility** criteria to find which patients to test under R208 and R444.1 criteria

NHS Scotland[3]

Scottish Genomic Test Directory for Rare and Inherited Disease

Note, testing criteria in Wales and Northern Ireland are similar but not identical to those listed. Please refer to local guidance.

Did you know?

AstraZeneca has a promotional *Expert on Demand* programme, where national experts can present on topics such as the 'Practicalities of *BRCA* Testing in Breast Cancer' during 30 minute sessions scheduled for you and your colleagues.

Please speak to your local AstraZeneca account manager to register your interest and hear about the other topics available.

References: 1. NHS East Genomics. Service turnaround times. Available at: https://www.eastgenomics.nhs.uk/about-us/quality/service-turnaround-times/ Accessed May 2025; 2. NHS England. National genomic test directory. Available at: https://www.england.nhs.uk/publication/national-genomic-test-directories/ Accessed May 2025; 3. NHS Scotland. Genomic Test Directory: Rare & Inherited Disease. Available at: https://www.genomics.nhs.scot/test-directories/rare-and-inherited-disease/ Accessed May 2025.

CNS=clinical nurse specialist; (g)*BRCA*(m)=(germline) breast cancer susceptibility gene (mutated); GLH=Genomic Laboratory Hub; HER2=human epidermal growth factor receptor 2; LGL=local genomics laboratory; MDT=multidisciplinary team; NHSE=NHS East; TAT=turnaround time.

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