The breast cancer risk assessment pathway in England: a systems analysis of current challenges and ways to improve, *British Journal of Cancer,* Valasaki et al. 2026. https://doi.org/10.1038/s41416-025-03329-2.

Introduction

Current pathway for identifying women at a moderate or high risk of breast cancer, eligible for additional screening, is dependent on whether women present to primary care with concerns about their family history. This can introduce inequalities and it is estimated fewer than 10% of women are aware of their increased risk.

Recent research has looked into the feasibility and acceptability of proactive risk assessment. The aims of this study were;

- To map out current breast cancer risk assessment and screening pathways for women under age of 50 in England
- Assess capacity and readiness for systemic change
- Identify potential strategies for implementing risk assessment that most likely optimise adoption of breast cancer risk assessment and subsequent uptake of risk reducing options

Methods

Qualitative interviews were conducted with 29 healthcare professionals and policy stakeholders involved in breast cancer risk assessment, management and screening in England. This was followed by an online post-interview questionnaire.

Data was collected between November 2024 and January 2025

Results

- Participants described the current pathway as fragmented due to differences in regional policies, resource allocation and service delivery. Three sub-themes arose;
    - *Postcode lottery;* they described differences in age range for eligibility for mammograms in different regions, but also lack of services. Similar situation was seen with preventive medications, with discussions around risks and benefits not taking place in all regions.
    - *Patchy;* this term was used to describe a broader, more systemic critique (e.g. issues of provider awareness, standardisation and social inequalities). Variation was particularly seen in primary care, where limited time for consultations and awareness amongst some GPs can impact whether women are offered appropriate risk assessment and referral.
    - *Opportunistic approach;* the current pathway was characterised as 'opportunistic' due to women needing to self-present with concerns about breast symptoms or risk to be assessed. During interviews, it was repeatedly mentioned that this creates a bias towards wealthier, higher-educated, English-speaking individuals who are more likely to advocate for themselves. Language barriers, socioeconomic status and ethnicity all emerged as significant determinants of access to this service.

- Standardised national service
  - Participants discussed the need for systemic reform by making fundamental, structural changes to create a more efficient, equitable and coordinated system, as a way of tackling fragmented care and unequal access.
  - Participants spoke about need for a national service which ensures a standardised approach for all and continuity of care for women, regardless of where they live
  - Several participants suggested that additional screening for moderate and high-risk women should be incorporated into the NHS Breast Screening Programme (NHS BSP)

- Developing digital tools
  - Longer term; participants suggested automated systems which could match existing patient data against risk-based protocols and direct women into the appropriate pathways, rather than relying on letters and emails.
  - Shorter term; a national hub for online risk assessment and a dataset for women above population risk was suggested, to ensure equitable access
  - Digital tools could improve access to risk assessment by enabling family history questionnaires to be completed in other settings
  - They discussed the idea of having a hotline for women to seek advice

- Towards being proactive
  - There was no consensus on the optimal age to begin risk assessment, but there was an agreement on the need for a baseline age
  - NHS health checks from age 40 were considered the best opportunity, within primary care. They acknowledged that this may still miss a lot women due to the low and sometimes inequitable uptake
  - It was agreed that there would need to be a system to include tailored approaches for undeserved groups, which can be achieved through religious and community groups
  - Outside primary care, participants accepted the idea of introducing risk assessment when women present to secondary care with breast symptoms

- Responsibilities
  - The focus was where the system risk assessment and management would be best conducted
  - When considering primary care, participants expressed the lack of capacity (time, skills and staff). Participants felt that family history clinics and CGS would have the capacity and expertise.
  - When thinking about preventive medication, there was no consensus on who should be prescribing (some supported primary care, whilst others referred to limited capacity and favoured family history or CGS)
  - The most preferred suggestion was that moderate-risk women be managed in FH clinics, whilst high-risk management was widely viewed as a shared responsibility between FH clinics and CGS

- Need for funding

    - Limited funding was felt to be the cause of many of the inconsistencies in service provision across different locations

- Strengthening skills
    - Participants highlighted the need for increased education and awareness around risk assessment, particularly within primary care, but also in secondary care.

Discussion

Current pathway was consistently described as postcode lottery, patchy and opportunistic as access to risk assessment and management was often influenced more by ethnicity, geographic and socioeconomic factors than by clinical need. Based on the challenges described, 10 recommendations were developed;

1. There should be a national strategy for risk assessment and management, ensuring a standardised approach for all.
2. Additional screening for women at moderate and high risk should be incorporated within the NHS Breast Screening Programme.
3. There is a need for a national register with centrally stored data for women at above-population-level risk.
4. There should be flexible and accessible digital tools alongside the offer of support or paper versions to improve access to risk assessment.
5. There is an emerging need for shifting towards a proactive system to improve equity of access to risk assessment.
6. Consideration should be given to offering risk assessment within NHS Health Checks, at cervical screening, via religious or community groups and when women present to secondary care with breast symptoms.
7. Moderate-risk women should be managed by specialist secondary care clinics and high-risk women should be managed between those clinics and CGS.
8. There is a need for clear shared-care pathways for counselling and initiation of preventive medication.
9. Funding should be prioritised to enhance capacity in family history clinics, screening, CGS and secondary care.
10. Ensure clinicians in primary care and those in secondary care, seeing women with breast symptoms, have access to training around risk assessment and management.

ERN GENTURIS guideline on counselling on reproductive options for individuals with a cancer predisposition syndrome (CPS) (including genturis), *European Journal of Human Genetics,* Farschtschi et al. (2026). https://doi.org/10.1038/s41431-025-02007-4

Introduction

Reproductive decision making for individuals with CPS is complex and nuanced, encompassing emotional, ethical and practical challenges. ERN GENTURIS have developed a guideline with the aim to highlight disparities, establish standards, empower HCPs and help patients uphold autonomy across diverse healthcare systems and social environments in Europe

Methods

ERN GENTURIS guideline group for counselling on reproductive options for individuals with CPS, was established by 20 experts in reproductive counselling from 10 countries and 3 patient representatives. The guideline was developed based on publications that could potentially answer the following health questions;

- What content should counselling regarding reproductive options have for individuals with a CPS?
- How can healthcare professionals aid individuals with a CPS to make informed choices
- Under what circumstances and at what time in the person's life should healthcare providers professionals refer individuals with a CPS for counselling regarding reproductive options?
- In what context should counselling for reproductive options be provided to individuals with a CPS?
- Who should perform counselling for reproductive options for individuals with a CPS?
- How should reproductive counselling be performed for individuals with a CPS?

Delphi survey was conducted which assessed the recommendations using a four-point Likert scale

Results

Recommendations

Reproductive decision making

1. Reproductive counselling should be offered to all individuals with a CPS. It is voluntary for the individual with a CPS to accept or decline counselling.
2. All individuals with a CPS and relevant family members of reproductive age should be offered information about their reproductive options.
3. Reproductive counselling must provide individuals with a CPS and relevant family members with comprehensive, balanced, and timely information.
4. Reproductive counselling should be non-directive ensuring patients can freely decline specific or all reproductive options without fear of recrimination, feelings of guilt or social pressure.
5. Couples, at risk for a child with a CPS, considering prenatal diagnosis should be encouraged to reflect on their views regarding continuation or termination of pregnancy pre-conceptionally.

6. Couples with a CPS considering pregnancy should have access to a multidisciplinary team of healthcare experts in an individualised way.
   This may include:
   a. A genetic counsellor or clinical geneticist to assess genetic risk, discuss the feasibility of both prenatal diagnosis (PND) and IVF (in vitro fertilization) with preimplantation genetic testing (PGT).
   b. A clinician experienced in performing and interpreting prenatal diagnostic tests to explain the risks, benefits, and procedures of PND options such as amniocentesis, chorionic villus sampling, and NIPT, if PND is considered.
   c. A fertility doctor to provide guidance on PGT, including PGT-M, and other assisted reproductive techniques where relevant.
   d. A psychologist trained in reproductive and genetic counselling, given the emotional and psychological impact of these decisions
   e. In difficult or unusual cases, advice should be sought from additional experts.

Timing of reproductive counselling provision

7. Reproductive counselling should be offered longitudinally to individuals with a CPS (and relevant family members) with multiple opportunities for discussion during reproductive age. At the time of diagnosis, individuals with a CPS should receive clear information about the availability of genetic and reproductive counselling services for future family planning.
8. Genetic and reproductive counselling should be available for individuals with a CPS and for parents (at risk) of an affected child, ideally beginning before family planning and continuing as needed.
9. Individuals with a CPS should be offered age- and context-appropriate genetic and reproductive counselling at the time of diagnosis.
10. Children at risk for cancer susceptibility should be offered counselling regarding predictive testing and family planning once they reach adulthood, or earlier if they express interest or the condition affects childhood.
11. Counselling regarding reproductive options is relevant for all individuals with a CPS, regardless of whether they already have children, are considering more children, or are not currently planning a pregnancy, since this may influence decisions regarding testing or informing their children/family members.

Presentation of reproductive options

12. Reproductive counselling for individuals with a CPS should provide sufficient time, follow-up opportunities, and access to psychological support.
13. Reproductive counselling should take psychological factors into account and be provided by a multidisciplinary team. This team should include professionals with expertise in reproductive genetics, oncology (when relevant), and psychological support. Access to additional specialists should be tailored to individual patient needs.
14. Reproductive counselling should be offered to both male and female individuals with a CPS independently and include their partners, if appropriate.

Range of assisted reproductive technologies

15. Female fertility preservation options, such as oocyte cryopreservation, should be included in reproductive counselling for individuals with a CPS, when there is a high risk of infertility due to cancer treatment.
16. Male fertility preservation options, such as sperm cryopreservation, should be included in reproductive counselling for individuals with a CPS, when there is a high risk of infertility due to cancer treatment.

Discussion

The broad recommendations outlined in the UK BSGM/FGG guidelines are in concordance, with each adapted to the relevant legal regulations and healthcare systems

The BSGM/FGG guidelines is more specific in the guidance relating to timing of prenatal diagnoses, whereas, the GENTURIS guidelines are more general in the timing and strategies, with less detail on the specific reproductive options. The main reason for this is that the GENTURIS guidelines aim to cover all European countries with different health care systems and options of reproductive interventions. The legal and ethical regulations across European countries are also quite diverse.

Future topics include;

- Centres providing assisted reproductive technology for CPSs to collaborate on analysing and publishing data on the success rate and safety
- Effectiveness of patient education tools to replace or supplement in-person reproductive counselling
- Role of lay organisations in development of guidelines
- Developing multinational registers
- Exerts and affected individuals working together to outline an ethical framework regarding reproduction options
- Lack of condition-specific evidence for the ideal form that reproductive counselling should take

A framework for using DNA methylation-based modelling for the clinical management of cranial meningioma, *Neuro-Oncology*, Landry et al (2025). https://doi.org/10.1093/neuonc/noaf237.

Background

- Cranial meningioma management is primarily guided by WHO grade and extent of resection (EOR), which do not consistently predict tumour recurrence or response to radiotherapy (RT). This can lead to overtreatment with adjuvant RT in some patients and undertreatment of biologically aggressive tumours classified as low grade.
- DNA methylation profiling has previously been shown to stratify meningiomas according to recurrence risk and predicted RT response. However, there is limited guidance on how these molecular models could be integrated into routine post-surgical clinical decision-making.
- This study assessed the combined clinical utility of methylation-based recurrence risk and RT response models in a large, multi-institutional cohort.

Methods

- A retrospective cohort of 1,953 cranial meningiomas with available clinical follow-up data was analysed. Tumour DNA was profiled using Illumina EPIC methylation arrays.
- Two previously validated methylation-based models were applied to each tumour:
    - a model predicting post-surgical recurrence risk
    - a model predicting sensitivity or resistance to RT
- Tumours were categorized as high- or low-risk for recurrence and as RT-sensitive or RT resistant, generating four combined molecular groups.
- Progression-free survival (PFS) was evaluated according to molecular classification, EOR and receipt of adjuvant RT.
- Molecular risk groups were compared with WHO grade and RTOG-0539 clinical risk stratification.

Results

- Four molecular subgroups were identified:
    - low-risk, RT-sensitive (61.8% of patients)
    - low-risk, RT-resistant (4.3% of patients)
    - high-risk, RT-sensitive (15.7% of patients)
    - high-risk, RT-resistant (18.2% of patients)
- Gross total resection was associated with improved PFS across all molecular groups.
- Patients with high-risk, RT-sensitive tumours derived significant benefit from adjuvant RT following both gross total and subtotal resection.
- In low-risk, RT-sensitive tumours, benefit from adjuvant RT was observed only after subtotal resection, with no clear benefit following gross total resection.
- Tumours classified as RT-resistant did not show a significant PFS benefit from adjuvant RT, regardless of recurrence risk.
- High-risk, RT-resistant tumours were associated with consistently poor outcomes.
- Molecular risk stratification showed notable discordance with WHO grading, with some WHO grade 1 tumours classified as high-risk and some grade 2–3 tumours classified as low-risk.

Conclusion

- This study demonstrates that DNA methylation-based modelling provides additional prognostic information beyond standard histopathological and clinical criteria.
- Combined assessment of recurrence risk and predicted RT response may help guide more selective use of adjuvant RT following surgery.
- Molecular profiling may reduce both overtreatment and undertreatment in meningioma management.
- Further prospective validation and evaluation of clinical implementation are required.

Monthly Journal Round-Up brought to you by:

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