

NICE: Familial Breast Cancer draft scope consultation

Yorkshire Cancer Research response, September 2025

Yorkshire Cancer Research comments:

Yorkshire Cancer Research agree that the 'Genetic testing for BRCA1, BRCA2 and TP53 mutations within 4 weeks of diagnosis of breast cancer' section should be considered for updating. It is vital that testing is carried out as soon after diagnosis as possible to be able to best inform treatment decisions. Timely testing can also ensure that people understand their risk of developing other cancers and can initiate cascade testing.

There is no specific mention within the scope document of cascade testing. Yorkshire Cancer Research recommend that the guideline update should review the evidence for cascade testing both male and female relatives after discovering a genetic mutation. This could enable a better understanding of carrier probability and improve the early detection of various cancer types.

The evidence around population based genetic screening is currently growing. For example, Yorkshire Cancer Research are currently funding PROTECT-C, a research study offering genetic testing to women, regardless of whether they or their families have had cancer. Increased evidence generation in this area could be critical for understanding carrier probability and enable more women to make informed decisions about their health. Studies such as this will provide a greater depth of understanding on population level rates of BRCA carriers. This guidance update should engage with population level studies and consider the value of recommending further research in this area.

There is no mention within this scope of the use of PARPi as a risk reduction strategy. In a recent position statement, Yorkshire Cancer Research along with six other organisations have called for research to explore the viability of whether poly (ADP-ribose) polymerase (PARP) inhibitors can be used as a chemo preventative treatment for people at high risk of hereditary cancers as part of a drive toward a future of precision cancer prevention. At this stage there is limited evidence for the use of PARPi within primary prevention. However, given the emerging evidence in terms of secondary prevention, the updated guideline should back this call for more research into the use of PARPi in risk reduction.

Yorkshire Cancer Research recommends that 'Family history-taking and initial assessment in primary care' is added to the scope of this guidance update to consider broadening the information gathered for referral decisions to include other Founder populations alongside those with Jewish ancestry.