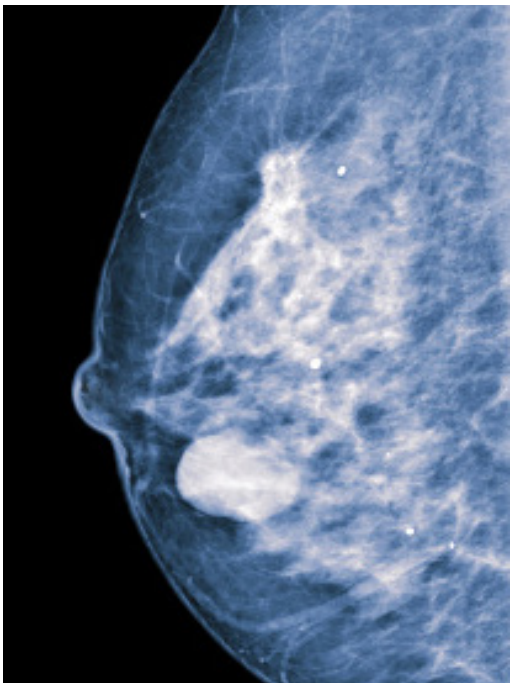
The early detection of breast cancer: An update from the Red book



Danielle Mazza, Jon Emery

Background
Breast cancer is the most common cancer affecting women and a leading cause of cancer-related deaths in Australia. Early detection through regular screening significantly improves survival rates and treatment outcomes.

Objective
This article provides an updated overview of breast cancer screening recommendations from The Royal Australian College of General Practitioners' (RACGP) 'Guidelines for preventive activities in general practice' (Red book), emphasising the critical role of general practitioners (GPs) in assessing individual risk and guiding preventive strategies.

Discussion
GPs should undertake an individualised risk assessment with each patient. Understanding risk levels enables personalised screening and prevention recommendations. While women with dense breasts are more likely to benefit from supplemental screening modalities, there is currently insufficient evidence to recommend routine supplemental screening for this patient group. Lifestyle factors significantly impact breast cancer risk and patients should be counselled about this. Technological advancements and personalised medicine will continue to shape breast cancer detection and management moving forward.

BREAST CANCER remains the most common cancer among women in Australia and is the second leading cause of cancer-related deaths in women. In 2022, approximately 20,640 new cases were expected, including 20,428 in women and 212 in men.[1] For Australian women, the lifetime risk of developing breast cancer by age 85 is one in eight (13%), while the risk is significantly lower for men at one in 668 (0.15%).[1]

Why screen for breast cancer?

Given its prevalence, early detection of breast cancer through regular screening is paramount. Early detection allows for intervention at an earlier stage when the disease is more treatable, leading to better survival rates, less aggressive treatments, and improved quality of life. Breast cancer screening, particularly through mammography, plays a vital role in reducing mortality rates by identifying malignancies before clinical symptoms arise.

What are the risk factors for breast cancer?

A large number of risk factors have been found to have convincing evidence for their association with breast cancer (see Box 1).[2] Age is one of the most significant with a person aged 50 years having 10 times the risk of a person aged 30 years.[3] Carriers of a pathogenic variant in the *BRCA* genes also have significant risk when compared to non-carriers (the relative risk estimate for *BRCA1* is 5.91 [95% CI: 5.25–6.67] and *BRCA2* is 3.31 [95% CI: 2.95–3.71]) as do carriers of pathogenic variants in the *PTEN* gene (5.83 [95% CI: 2.43–14.0]) and the *PALB2* gene (3.39 [95% CI: 2.79–4.12]).[4]

How should general practitioners approach breast cancer screening and risk assessment?

General practitioners (GPs) should undertake an individualised risk assessment to guide their patients effectively. This involves reviewing family history, genetic predisposition and other relevant factors. It is important to

note that breast cancer risk is not normally distributed and that most women have a low (<4%) lifetime risk.[5] Several risk calculators exist that account for these multiple risk factors and provide more accurate assessment of breast cancer risk than using age and family history alone.[6,7] However, they can take time to use in a consultation, given the complexity of the information needed to assess risk.

Table 1 outlines breast cancer risk levels for women, based on family history and other genetic factors.[2] These levels range from average or slightly increased risk to moderate and potentially high risk. Understanding these risk levels enables personalised screening and prevention recommendations.

Women with a family history placing them at potentially high risk should be offered referral to a familial cancer centre to consider genetic testing for high-risk single gene variants. Polygenic risk scores for breast cancer, which are applicable to women regardless of their family history, are available commercially; however, their precise role in risk assessment and tailored prevention is still under consideration.

What are the current screening recommendations for average-risk women?

BreastScreen Australia recommends biennial mammography for asymptomatic women aged 50–74 years, as evidence suggests this age group benefits most from routine screening. Women aged 40–49 years might choose to begin screening, although the evidence for benefit in this age group is less robust.[8] Screening mammograms are not routinely recommended for women aged ≥75 years due to insufficient evidence on the balance of benefits and harms.[8]

Reviews of evidence from randomised controlled trials of mammography estimate rates of overdiagnosis of breast cancers that would not have affected mortality at between 11% and 19%.[9] More recent modelling data from the US estimate that biennial screening from ages 40–74 years would result in 14 overdiagnosed cases of breast cancer per 1000 women screened over the lifetime of screening (estimated range 4–37 overdiagnosed cases).[10] While screening mammography in women aged 40–49 years reduces the risk of dying of breast cancer, the number of deaths averted is much smaller than in older women and the number of false-positive tests and unnecessary biopsies is larger.[10]

There is insufficient evidence that clinical breast examination reduces breast cancer mortality and so it is not recommended for screening of average-risk women in general practice.[11]

What screening should be provided for women at moderate risk?

Women at moderate risk may be advised to undergo annual mammography starting at age 40.[8] This group includes those with a family history of breast cancer but without known high-risk variants in genes such as *BRCA1* and *BRCA2* or high-risk family history features.

What about women at high risk?

Women at high risk, such as carriers of *BRCA1* or *BRCA2* high-risk variants or those with a significant family history, might benefit from annual MRI screening, which has greater sensitivity than mammography, especially in younger women and those with dense breasts. A Medicare rebate is available for MRI in asymptomatic high-risk women under 60 years of age if their lifetime breast cancer risk is >30% or 10-year risk >5%.[8] Additional risk-reduction strategies might include chemoprevention (eg selective oestrogen receptor modulators) and prophylactic surgery (mastectomy or oophorectomy).

What should be recommended for women with dense breasts?

Fewer than 10% of women have breasts in the lowest and highest quartiles of breast density; and breast cancer risk among women with dense breasts is more usefully compared to women with average breast density.[12] Accordingly, women with moderately dense breasts have approximately 1.5 times the risk of breast cancer and women with moderately non-dense breasts have approximately 0.6 times the risk of breast cancer compared with women with averagely dense breasts.[12] Breasts become less dense as women age.[12] BreastScreen in several Australian states will be routinely reporting breast density to women and suggesting discussion with their GP.

Box 1. Factors with convincing evidence of association with risk of breast cancer[2]

- Age (risk increases in older women)
- Geographic location and residence
- Urbanisation
- High socioeconomic status
- Height (risk increases with height)
- High mammographic breast density
- Family history of breast cancer
- Family history of other cancers
- *ATM* gene mutation
- *BRCA1* gene mutation
- *BRCA2* gene mutation
- *CDH1* gene mutation (lobular breast cancer)
- *CHEK2* gene mutation
- *PALB2* gene mutation
- *PTEN* gene mutation
- Polygenic risk score (single nucleotide polymorphisms)
- *STK11* gene mutation
- Previous benign breast disease (proliferative)
- LCIS
- DCIS
- Previous primary invasive breast cancer
- Earlier age at menarche
- Nulliparity (risk)/parity (protective)
- Later age at first birth
- Later age at menopause
- Circulating hormones – oestrogen (postmenopausal), testosterone, IGF-1
- Current use combined hormonal contraception
- Current use combined menopausal hormone therapy
- Maternal exposure to diethylstilboestrol
- Adiposity (eg BMI) in adulthood (postmenopausal)
- Weight gain (postmenopausal)
- Alcohol consumption
- Ionising radiation—radiotherapy

BMI, body mass index; DCIS, ductal carcinoma in situ; IGF-1, insulin-like growth factor; LCIS, lobular carcinoma in situ.

Table 1. Breast cancer risk levels[2]

Risk level	Average or slightly higher	Moderately increased (<4% of the female population)	Potentially high risk[A] or carrying mutation (<1% of the female population)
Risk in relation to the population average	Approximately 1.5 times the population average	Approximately 1.5–3 times the population average	More than three-fold times the population average
Lifetime prevalence of breast cancer up to age 75 years	9–12.5%	12–25%	25–50%
Relevant history	• No confirmed family history of breast cancer • One first-degree relative diagnosed with breast cancer at age ≥50 years • One second-degree relative diagnosed with breast cancer at any age • Two second-degree relatives on the same side of the family diagnosed with breast cancer at age ≥50 years • Two first- or second-degree relatives diagnosed with breast cancer at age ≥50 years, but on different sides (ie on each side) of the family	• One first-degree relative diagnosed with breast cancer at age <50 years (without the additional features of the potentially high-risk group) • Two first-degree relatives, on the same side of the family, diagnosed with breast cancer (without the additional features of the potentially high-risk group) • Two second-degree relatives, on the same side of the family, diagnosed with breast cancer, at least one at age <50 years (without the additional features of the potentially high-risk group)	• Two first- or second-degree relatives on one side of the family diagnosed with breast or ovarian cancer, plus one or more of the following features on the same side of the family: – additional relative(s) with breast or ovarian cancer – breast cancer diagnosed before age 40 years – bilateral breast cancer – breast and ovarian cancer in the same woman – Ashkenazi Jewish ancestry – breast cancer in a male relative • One first- or second-degree relative diagnosed with breast cancer at age <45 years, plus another first- or second-degree relative on the same side of the family with sarcoma (bone/soft tissue) at age <45 years • Member of a family in which the presence of a high-risk breast cancer gene mutation (eg *BRCA1*, *BRCA2*) has been established

[A]Individual risk might be higher or lower if genetic test results are known.

In addition to increasing the risk of future breast cancer, dense breast tissue can obscure mammographic findings, potentially delaying diagnosis.[8] Women with dense breasts are more likely to benefit from supplemental screening modalities such as ultrasound or MRI.[8] However, evidence remains insufficient to recommend routine supplemental screening for dense breasts.[8]

What other preventive advice can be given?

Lifestyle factors significantly impact breast cancer risk. GPs should counsel patients on the following modifiable risk factors:

- Physical activity: regular exercise is associated with a lower risk of postmenopausal breast cancer.
- Weight management: maintaining a healthy body weight reduces the risk of postmenopausal breast cancer.
- Alcohol consumption: minimising alcohol intake is recommended, as alcohol is a known risk factor.
- Reproductive factors: having children and breastfeeding are protective factors.

Educating patients about these behaviours empowers them to reduce their risk proactively.

Should women undertake self-examination?

Formal self-examination is no longer routinely recommended but breast awareness is important to ensure women seek advice about symptoms of breast cancer.[8] It is recommended that all women, whether or not they undergo mammographic screening, are aware of how their breasts normally look and feel. They should promptly report any new or unusual changes (eg a lump, nipple changes, nipple discharge, change in skin colour, skin texture, pain in a breast) to their GP. No one method for women to use when checking their breasts is recommended over another.

What lies ahead?

Technological advancements and personalised medicine continue to shape breast cancer detection and management. Future developments might include the following:

- Enhanced imaging technologies: improvements in mammography such as 3D tomosynthesis and artificial intelligence (AI) analyses of mammographic images might offer greater accuracy in detecting breast cancer and predicting future risk.
- Risk-based screening models: incorporating genetic, lifestyle and breast density data could enable more tailored screening recommendations.

- Biomarker research: advances in understanding molecular biomarkers might lead to earlier detection and personalised treatment strategies.

Conclusion

Breast cancer remains a significant public health challenge. Early detection through regular screening and risk-based prevention strategies offers the best chance of improving outcomes. GPs play a vital role in guiding patients through evidence-based screening, lifestyle modifications and, when necessary, genetic testing and specialised care. Continued research and advancements in screening technology promise further improvements in breast cancer management and survival.

Key points

- Age and lifestyle factors such as physical activity, weight, alcohol consumption and reproductive factors significantly impact breast cancer risk.
- An individualised risk assessment reviewing family history, genetic predisposition and other relevant factors should guide screening advice.
- People with a strong family history of breast cancer, as well as other cancers associated with high-risk genetic variants (eg *BRCA1* and *BRCA2*), should be offered referral to a familial cancer service.
- Women with dense breasts are more likely to benefit from supplemental screening but evidence remains insufficient to recommend this routinely.
- Enhanced imaging technologies and risk-based screening models incorporating genetic, lifestyle and breast density data could enable more tailored screening recommendations.

Authors

Danielle Mazza AM, FAHMS, MD, MBBS, FRACGP, DRANZCOG, Grad Dip Women's Health, GAICD, CF Head, Department of General Practice, Monash University, Melbourne, Vic; Director, SPHERE NHMRC Centre of Research Excellence in Women's Sexual and Reproductive Health in Primary Care, Melbourne, Vic; Chair, Red book Executive Committee and Co-clinical Lead – women's health, breast cancer, cervical cancer chapters, The Royal Australian College of General Practitioners, Melbourne, Vic

Jon Emery MA, MBBCh, FRACGP, MRCGP, DPhil, Herman Professor of Primary Care Cancer Research, University of Melbourne, Melbourne, Vic; Director of Cancer Australia Primary Care Collaborative Cancer Clinical Trials Group (PC4), Melbourne, Vic; Primary Care Research and Education Lead, Victorian Comprehensive Cancer Centre (VCCC), Melbourne, Vic; Red book Clinical Lead – cancer and genetics, The Royal Australian College of General Practitioners, Melbourne, Vic

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Correspondence to:
danielle.mazza@monash.edu

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correspondence **ajgp@racgp.org.au**