

Clinical practice guideline of BRCA1/2 testing for patients with breast cancer: Chinese Society of Breast Surgery (CSBrS) practice guideline 2021.

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There were 78 voting committee members for the guideline: 65 breast surgeons (83.3%), four medical oncologists (5.1%), three radiologists (3.9%), two pathologists (2.6%), one radiation therapist (1.3%), and two epidemiologists (2.6%).

Clinicians specializing in breast diseases in China.

Recommendation 1: Indications for BRCA 1/2 testing.

* Indications for high-risk healthy women refer to “Clinical Practice Guidelines for Risk Assessment to Identify Women at High Risk of Breast Cancer: Chinese Society of Breast Surgery (CSBrS) Practice Guidelines 2021.” † Close blood relatives include first-, second-, and third-degree relatives. ‡ Includes fallopian tube and primary peritoneal cancers.

Recommendation 2: Risk control for BRCA1/2 mutation carriers.

* Individualized risk control strategies should be developed for BRCA1/2 mutation carriers over 75 years of age. † Including nipple-areolar complex (NAC)-sparing or skin-sparing mastectomy. MRI: Magnetic resonance imaging.

Recommendation 3: Surgical options for breast cancer patients with BRCA1/2 mutation.

* Including nipple-areolar complex (NAC)-sparing or skin-sparing mastectomy.

Indications for BRCA1/2 testing were recommended by considering national conditions and referring to relevant guidelines and expert consensus. [6 , 8 – 12] BRCA1/2 gene variants were devised into five classifications for quantitative assessment of variant pathogenicity [Supplementary File 1].

In BRCA1/2 mutation carriers who are younger than 30 years old, breast magnetic resonance imaging screening is preferred over mammography due to the potential radiation exposure risk and less sensitivity for detection of tumors associated with mammography. Ultrasound is also an appropriate primary imaging examination. Ultrasound combined with mammography may be considered in the condition of unavailable enhanced magnetic resonance [Supplementary File 2]. Chemoprevention is not recommended for BRCA1/2 mutation carriers [Supplementary File 3].

Studies have indicated that bilateral risk-reduction mastectomy (RRM) decreased the risk of developing breast cancer by at least 90% in moderate- and high-risk women and in known BRCA1/2 mutation carriers. [13 , 14] However, this risk-reducing surgery was not significantly associated with reduced all-cause mortality. [13 , 14] Carrying a BRCA1/2 mutation is not significantly associated with nodal metastasis, [15] so sentinel lymph node biopsy is not recommended when RRM is performed.

Whether there is an increased risk of ipsilateral breast cancer recurrence in breast cancer patients with BRCA1/2 mutation after breast-conserving treatment has not been confirmed [Supplementary File 4]. The guidelines panel recommend that patients with BRCA1/2 mutation who are first diagnosed with breast conserving indications receive breast conserving surgery with the assurance of adjuvant therapy. [6 , 9 – 12] Contralateral prophylactic mastectomy for breast cancer with BRCA1/2 mutations could be considered. If prophylactic mastectomy was performed, immediate breast reconstruction is recommended [Supplementary File 5].

The expert committee for these guidelines declares no conflict of interest.

These guidelines are a reference for breast disease specialists in clinical practice. However, the guidelines are not to be used as the basis for medical evaluation, and do not play an arbitrating role in the handling of any medical disputes. The guidelines are not a reference for patients or non-breast specialists. The Chinese Society of Breast Surgery assumes no responsibility for results involving the inappropriate application of these guidelines, and reserves the right to interpret and revise the guidelines.

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