Screening – surveillance of women who are known to carry a breast cancer susceptibility gene mutation (eg, BRCA)

Standard 1.2	Women who are known to carry a breast cancer susceptibility gene mutation (BRCA) have annual breast MRI and consider annual mammography from 10 years prior to the age of onset for the youngest affected family relative. Mammography is not recommended before 30 years of age.

Rationale

Breast MRI is the most sensitive and specific test for early breast cancer detection in breast cancer susceptibility gene mutation carriers. Mammography and ultrasound have been shown to be relatively insensitive in this group.

Good practice points

1.17 Women with p53, PTEN and CDH-1 (E-cadherin) gene mutations are managed similarly to breast cancer susceptibility gene mutation carriers.

1.18 Mammography is not recommended under 30 years of age, because of the low sensitivity of the test and the risk of radiation-induced cancer (NHS Cancer Screening Programmes 2013; Pijpe et al 2012; expert opinion).

1.19 Women with p53 mutations are especially at risk of radiation induced malignancy so should not have mammograms. Whole body MRI may be considered in view of the complex of malignancies associated with p53 mutations.

1.20 Women are encouraged to report any breast changes (such as lumps or swelling, nipple discharge, skin puckering or discolouration) promptly to their clinician.

1.21 Women have a clinical breast examination every six to 12 months with a clinician who specialises in breast care from 10 years prior to the age of onset for the youngest-affected family relative or starting at 25–30 years of age (Genetic Health Service New Zealand 2012).

1.22 The specialist responsible for a woman's ongoing management should follow general high-risk surveillance recommendations developed by Genetic Health Service New Zealand (Genetic Health Service New Zealand 2012).

See Standard 1.4 and Genetic services section (Surveillance of women of potentially high risk but mutation status not known).