identifying affected women at higher risk for inherited forms are promising.

Key messages:

- BRCA testing programs are effective public health programs that must be systematically implemented in all developed countries to prevent avoidable cancers.
- BRCA screening programs of unaffected and affected individuals based on family and personal history, and followed by cascade testing, are promising public health strategies.

From bench to bedside: a Health Technology Assessment of BRCA genetic testing programs.

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Background:

The evidence generated on the incremental benefit of the appropriate use of BRCA genetic testing for preventing breast and ovarian cancers suggested that delivery programs for this testing must be systematically implemented in all developed countries. However, it is still unclear which are the programs that could be a best fit for the European health care systems.

Methods:

To answer this issue, we performed a comprehensive evaluation of all BRCA testing programs potentially ready for implementation using a Health Technology Assessment approach.

Results:

Published information on the analytic validity of BRCA testing estimated analytic sensitivity, specificity and accuracy at 99%. Clinical validity results, stratified by prevalence and penetrance, showed high and consistent positive and negative predictive values. In clinical utility, the most effective procedure was the prophylactic surgery. Bilateral mastectomy and salpingo-oophorectomy could reduce breast, ovarian cancers and mortality up to 100%. Evidence of personal utility were also provided. The main BRCA programs identified and evaluated were: (i) population-based genetic screening of individuals without cancer; (ii) family history (FH)-based genetic screening, on women without cancer but with a suggestive FH; (iii) familial mutation-based genetic screening, on women without cancer but with carriers in the family; and (iv) cancer-based genetic screening, on women with BRCA-related cancers.

Conclusions:

Population-based screening is a good investment among specific ethnicities only. FH-based screening is potentially the best fit for most European countries, however further studies on the identification of unaffected high-risk women are needed. More evidence should be provided for the screening of all newly diagnosed cases of breast/ovarian cancers, followed by cascade testing, but programs that include tools for

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