Further development and evaluation of a breast/ovarian cancer genetics referral screening tool

To the Editor:

This letter is being written in follow-up to our original article "Evaluation of a breast/ovarian cancer genetics referral screening tool in a mammography population" published in the November 2009 issue of *Genetics in Medicine*.[1] The tool is designed to quickly identify individuals who are appropriate to refer for cancer risk consultation regarding *BRCA1/2* testing.

We modified the original paper-based tool in an attempt to address some of its limitations and have created an interactive web version (breast cancer genetics referral screening tool [B-RST]) to enhance accessibility. The tool now distinguishes between maternal and paternal lineages when identifying second-degree relatives. We have added nieces and nephews to the list of included second-degree relatives. In addition, the parameters of bilateral breast cancer, and breast and ovarian cancer in the same individual, have been added to the algorithm.

We evaluated the new web-based version of the tool using the pedigree data collected on the subjects in the original study.[1] As before, the tool score (positive or negative) was compared with analysis of the full pedigrees using BRCAPRO, Myriad II, BOADICEA, and family history assessment tool (FHAT), with a ≥10% *BRCA1/2* mutation probably or an FHAT score of 10 as the true measure of "high" risk. Table 1 provides the validation statistics of the revised tool (Table 1). Sensitivity increased for all model comparisons (+8.2% to +11.1%), whereas specificity decreased slightly (−0.4% to −2.3%). As hypothesized in the original article, the addition of bilaterality accounted for the majority of the reduction in specificity. How-

ever, we chose to retain this parameter in the web version, because it is included in most *BRCA1/2*-related guidelines and models, and maximizing sensitivity is desired.

The revised tool (B-RST) is now available (open access) on the web at: http://www.brcagenscreen.org. There is both a health care provider and a consumer portal. Information entered by users is not stored, but a printable report is provided, which includes the screen result, interpretation, disclaimers, family history survey responses, and resources. Future studies to access the clinical utility of the tool are planned.

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Disclosure: The author declares no conflict of interest.

REFERENCE

1. Bellcross C, Lemke A, Pape L, Tess AL, Meisner LT. Evaluation of a breast/ovarian cancer genetics referral screening tool in a mammography population. *Genet Med* 2009;11:783–788.

Table 1 B-RST validation measures

Measure	BOADICEA	BRCAPRO	Myriad II	FHAT	Overall[a]
Sensitivity (%)	100	100	96.9	90.0	89.4
Specificity (%)	75.1	74.0	76.1	89.8	91.5
ROC AUC (95% CI)	0.88 (0.83–0.92)	0.87 (0.83–0.91)	0.87 (0.81–0.92)	0.90 (0.85–0.94)	0.90 (0.85–0.95)

[a]High risk by at least one model = actual positive state.
B-RST, breast cancer genetics referral screeningtool; ROC AUC, area under the receiver operator characteristic curve; CI, confidence interval.