Erratum

In the September 2002 issue of the *Journal,* in the article entitled, "*BRCA2* T2722R Is a Deleterious Allele That Causes Exon Skipping," by Fackenthal et al. (71:625–631), the authors reported evidence for a *BRCA2* exon-skipping allele carrying a disruption in a potential exonic splicing enhancer (ESE). The evidence for the *BRCA2* T2722R allele being deleterious per se was its cosegregation with affected members of a family exhibiting classic hereditary breast cancer phenotypes, including high-frequency breast cancer occurrence among first-degree relatives, early-onset breast cancer, bilateral breast cancer, and ovarian cancer. Using methods published elsewhere, we predicted that the base change in this allele, 8393C→G, disrupts three overlapping consensus ESE sequences, potentially resulting in skipping exon 18 during splicing. We performed RT-PCR on mononuclear blood cells from an affected mutation carrier and on control lymphoblasts from an unaffected individual, and we found that the exon skipping in the mutation carrier was far more pronounced than the background level seen in the wild type *BRCA2* (fig. 3). However, as discussed on p. 627, this result was from a potentially saturating end-point PCR (made necessary by limited patient material) and was not fully quantitative.

Since making these observations, we and others have sequenced the full-length RT-PCR product and have found that, on several occasions, both the mutant and wild-type alleles are detectable. Thus, the putative T2722R-specific exon-skipping event is not complete. The possibility of both mutant and wild-type alleles contributing to correctly spliced messages was discussed on page 629. As this new evidence suggests that exon-skipping may not be fully penetrant in blood cells, the authors feel it is important to regard the *BRCA2* T2722R allele as an unclassified variant until further analysis can provide definitive evidence of clinically deleterious behavior in breast epithelial cells.