Ashkenazi Jewish genetic disease carrier screening**Reply to letter from Begleiter et al.:**

We thank the authors for their comments regarding testing of the non-Ashkenazi Jewish partner of a known (Ashkenazi Jewish) carrier. They are quite right that some of the disorders included in the panel are rarely, if ever, found in individuals who are not of Ashkenazi Jewish heritage. In fact, if it was known with certainty that the partner did not have any Ashkenazi Jewish ancestors, the authors' conclusion not to offer testing would have merit. However, it has been our experience, particularly those of us who practice in very diverse populations, that self-reported ethnicities can be mistaken. In the case of Ashkenazi Jewish reporting, there are other factors to take into account as the concept of "ethnicity" overlaps with religious, cultural, and historical considerations. In addition, some of the other disorders (e.g., cystic fibrosis) are not limited to the Jewish population. It is also worth bearing in mind that with respect to cost, it is not necessary to test the non-Ashkenazi Jewish partner for the entire carrier screening panel because carrier testing is only recommended for the specific disease gene in question. Clearly, we have an obligation to counsel such individuals that the chance of a positive result is low and that the residual carrier risk may not be able to be calculated. However, molecular testing does offer the option of potentially detecting a disease-causing mutation in someone who may not be fully aware of his or her ethnic heritage. Such

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a result would have a significant impact on the couple's reproductive options and possibly other family members.

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