



Response to Faulkner et al.

We appreciate Faulkner et al.'s thoughtful letter¹ in response to our article.² As a general matter, we agree that different clinical genetic testing situations create different challenges with regard to reinterpretation of test results (especially variants of uncertain significance [VUS]) and subsequent communication with patients.

With no pretense of offering a complete analysis here, we note that many of the considerations that incline us to endorse a responsibility for reinterpretation in the diagnostic testing context are present in carrier screening as well. Patients may benefit from more accurate interpretations of their carrier status both for their current pregnancy, if ongoing, and in future pregnancies. In the former case, even if termination is no longer a consideration, knowledge that a fetus may be at risk for a genetic disorder may permit couples and their medical providers to determine the fetus's status and to better prepare for the infant's care. With the future in mind, patients will not be left with a misleading impression of either being or not being a carrier when reinterpretation changes that status (which will be especially relevant if their partner is a carrier), allowing appropriate adjustment of their reproductive plans. In addition, the failure to notify patients of changes in their carrier status when reinterpretation of previous testing is called for may lead to a failure of trust in the medical system generally and in genetic testing in particular.

However, we do not underestimate the complexity of conveying new information about carrier status to couples during pregnancy, including the possibility of evoking considerable—and perhaps needless—anxiety. In keeping with the suggestion in our article, this consideration underscores the

importance of including patients in the development of approaches to reinterpretation, communication of results, and follow-up, so that the most beneficial and least harmful procedures can be developed. We look forward to participating with Faulkner et al. and others in ongoing discussions as to how this can best be accomplished.

DISCLOSURE

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