

Response to Zlotogora and Meiner

We appreciate the thoughtful letter “Ashkenazi carrier screening for reproductive planning: is this what we planned for?” by Joël Zlotogora and Vardiella Meiner.¹ We answer with a resounding “Yes.” It was our intent to present an updated panel that could be used not only for reproductive planning but also for early diagnosis and intervention. “Expanded Genetic Screening Panel for the Ashkenazi Jewish Population”² responds to what our patients have told us that they want³ as well as to the recommendation of the American College of Medical Genetics and Genomics: to provide information about actionable variants when such information becomes available from genetic testing.⁴ Patients receiving genetic counseling can choose the conditions for which they want testing and those for which they do not.

To address the accuracy of annotation and the severity of phenotypes, we performed our own independent consensus review because we observed that ClinVar annotations were often internally discordant. We acknowledge that future revisions will be made because some conditions are found to be exceedingly rare or because clinical utility is not observed. As more sequencing

and improved annotation is achieved, this expanded panel will be expanded even more.

DISCLOSURE

The authors declare no conflict of interest.

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