Response to Mendelsohn and Sabbadini

To the Editor,

I thank Drs. Mendelsohn and Sabbadini for their thoughtful letter[1] in response to my commentary on secondary findings in virtual panels[2] and agree with their central points. We agree that secondary findings in virtual panels constitute a challenge to the practice of genetic testing, and we agree that there is a mismatch between the availability of testing (and secondary findings) and pre- and posttest counseling. We also agree that a broader, organized approach to genomic screening would be desirable—but that must wait for an evidence base to support it. We also agree that the current approach of simply pretending secondary findings are not being detected is unacceptable—in no other area of medicine would we pretend we did not do a test because we didn't want to address the consequences of it.

It is worth pointing out that Mendelsohn and Sabbadini conflate opportunistic screening with population screening. "The ACMG believes that there are genes in which abnormalities are actionable and should be communicated to patients, but the evidence, funding, and will to systematically examine these genes in the population is lacking." This framing is just wrong enough to engender that confusion. What the ACMG said, is that *if* you detect a variant that is highly predictive of an actionable disorder in one of these 59 genes, you should tell the patient. What they are *not* saying is that you should detect the variant in the first place. Secondary findings are not a public health endeavor and there is no reason to expect the health-care system to organize the response to them as such.

What to do about secondary findings in virtual panels and exome slices is challenging. Mendelsohn and Sabbadini propose an add-on clinical charge to support pre-test and posttest education and counseling. Indeed, we and others are generating an evidence base to support online or telephone genetic services, which may be a cost-effective alternative to in-person education and counseling.[3,4] We need to demand that nongenetics providers provide this service for patients through in-person care or alternative means.

In the long run, the challenges created by testing laboratories that are performing virtual panels, exome slices, and anticipatory sequencing are good for our field. The plain fact is that secondary findings are an inescapable component of clinical genomics, and we just need to get over it and take care of the patients as best we can. We actually do want as many people who need testing to actually undergo testing and we need more doctors than just geneticists to order such tests and they need to be doing it correctly—including secondary findings. For the most part, patients are not afraid of secondary findings and we should not patronize them by thinking we are protecting them from such findings. But most importantly, this approach has the notable benefit of honesty—we are actually telling the patient what we are testing them for.

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