Genetics inMedicine | CORRESPONDENCE

Response to Biesecker

From the laboratory standpoint, Dr. Biesecker's[1] logic is sound: possession of sequence data from a gene is the same regardless of the scope of the primary test, and secondary findings should be reported. The challenge to the implementation of Dr. Biesecker's proposal arises in the clinic, where genetic testing is ordered and results reported. A large fraction of nonexome genetic tests are requested by health providers without specific training in genetics. Thus, by reporting all secondary findings on panels, the proportion of genetic tests with the potential for secondary findings would increase dramatically, with no parallel increase in the quality or quantity of pretest counseling or posttest expertise. The American College of Medical Genetics and Genomics (ACMG) probably intended for secondary findings to be released whenever they are available, as Dr. Biesecker observes, but we imagine the ACMG also expected quality counseling and clinical support before and after testing. There is already a large "counseling gap" and Dr. Biesecker's proposal would only increase it further.

Dr. Biesecker's article belies a more fundamental challenge with the concept of the ACMG 59, an artifact of the odd transitional time where the field of genetics finds itself at this moment in history. 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. We are thus left with an "opportunistic" screening model, lacking the organization of a true public health endeavor.

If Dr. Biesecker's proposal is to become reality, we propose that subexome tests that would newly include the possibility of secondary findings require a separate nominal fee and consent form to force recognition by the patient and ordering provider that the scope of the test extends beyond the primary indication, and to prompt discussions of informed consent, testing goals, and access to genetics expertise. Geneticists and genetic counselors must also collaborate with the larger health system to provide more organized and accessible options for preventive genetic screening, which we propose would offer a superior care model compared with examining random subsets of the ACMG 59 when patients undergo unrelated genetic testing with nongeneticists.

DISCLOSURE

Bryce Mendelsohn is on the advisory board of Clear Genetics. He has received speaking fees from Medscape. He has received consulting fees from Genome Medical. Marta Sabbadini has received speaking fees from Medscape.

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