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Secondary findings from genomic sequencing are frequent

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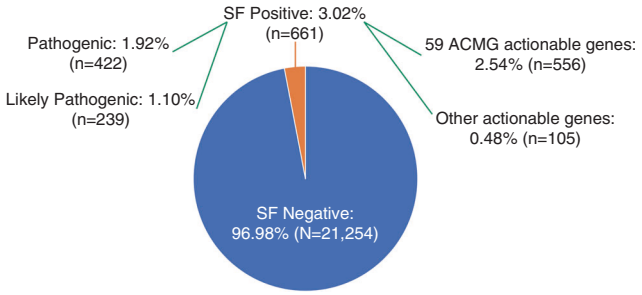

More and more DNA sequencing is finding its way into the clinic and quickly becoming a routine part of clinical care. As a result, secondary findings—sequencing results that are medically actionable but unrelated to the reason for testing—are also becoming more prevalent. Just how prevalent has been a subject of debate. In this issue, Gordon and colleagues report the frequency of secondary findings (SFs) from a cohort of more than 21,000 participants in the National Human Genome Research Institute's Electronic Medical Records and Genomics (eMERGE) Network. The researchers developed a sequencing panel called eMERGEseq that includes 56 genes on the American College of Medical Genetics and Genomics (ACMG)'s recommended list of reportable SFs as well as 11 additional genes and 14 variants in 11 more genes. The panel was deployed at ten clinical sites across the United States in a diverse cohort. The researchers identified 661 medically actionable findings that were unrelated to patients' test indication, yielding an overall SF frequency of 3%. The great majority of SFs (84%) were in variants in genes that the ACMG recommends reporting. Most SFs were in cancer-associated genes, followed by cardiac disease and lipid disorder–associated genes. Most cancer-associated SFs were classified as pathogenic, while cardiac-associated SFs were likely pathogenic. Secondary findings were most prevalent in participants who self-reported as Caucasian/white (3.1%), followed by Asian (2.7%), Black/African American (2.3%), Hispanic/Latinx (2%), and American Indian, Alaska Native, or Native Pacific Islander (1.3%). However, when the researchers excluded results from an allele known to be common only in those with European ancestry, the frequency of SFs between those self-reporting as white versus all other groups combined was no longer significant. The researchers conclude that the findings offer an important resource for patients and research participants as well as facilitating the development of practice standards and guidelines in genomic medicine. —*V. L. Dengler, News Editor*

How Australia integrated genetic testing into oncology practice

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Up to a fifth of high-grade serous ovarian cancers possess *BRCA* variants, yet only 30% of women with epithelial ovarian cancer are referred to genetic counseling. In Australia, genetic testing no longer requires referral to a genetics specialist. Instead, oncology health professionals have taken up the process of testing and delivery of results. O'Shea and colleagues investigated how genetic and oncology experts view making *BRCA* testing a routine part of oncology care for epithelial ovarian and breast cancer patients. The researchers conducted semistructured telephone interviews with genetic counselors, clinical geneticists, nurses, oncologists, and a surgeon, from rural, metropolitan, and statewide hospitals across Australia. The interviews, which used an implementation science framework and consisted of 17 questions, uncovered four themes that affected the implementation of mainstreaming *BRCA* testing. First, most participants found *BRCA* testing to be clinically useful, as it offered patients further treatment options. However, they also pointed out a need for optimizing delivery of results and follow-up. Second, the interviews identified communication networks and role delineation as central components of implementation. In particular, participants cited a need for relationships between genetic counselors and oncologists as well clarity regarding providers' patient responsibilities. The interview results also revealed factors in sustaining routine testing, including having ongoing training in genetics, limited resources and funding, and challenges in integrating public and private systems for sharing genetic information. Finally, the interviews identified intervention characteristics influencing implementation. For example, most participants supported having a genetic counselor embedded in an oncology clinic as a version of mainstreaming; however, some interviewees indicated that funding and state health system structures might preclude this intervention. The researchers conclude that the results will facilitate future research for guiding the integration of routine genetic testing in other cancer types. —*V. L. Dengler, News Editor*

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