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ACMG NEWS

For Immediate Release

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ACMG Releases Member Survey Results on Secondary Findings and Reinforces Earlier Recommendations in New Policy Statement on Secondary Findings

Nov. 13, 2014: Bethesda, MD - The American College of Medical Genetics and Genomics (ACMG) has just released two documents "online ahead of print" in its peer-reviewed academic journal, *Genetics in Medicine:*

1. "American College of Medical Genetics and Genomics (ACMG) Policy Statement: Updated Recommendations Regarding Analysis and Reporting of Secondary Findings in Clinical Genome-Scale Sequencing"

2. "Reporting Genomic Secondary Findings: ACMG Members Weigh In."

These two documents include the results of an ACMG member survey about ACMG's initial recommendations on Secondary Findings and also articulate the current recommendations of the ACMG with regard to the analysis and return of secondary findings when clinical genome-scale analysis is pursued.

These two new documents are available at:
http://www.nature.com/gim/journal/vaop/ncurrent/index.html - 13112014

Key points of the **ACMG Policy Statement** regarding its updated recommendations on the analysis and reporting of secondary findings in clinical genome-scale sequencing:

- When clinical genome-scale (e.g. WES, WGS) sequencing is performed, written informed consent should be obtained by a qualified genetics healthcare professional, describing the nature of the test and addressing such points as interpretive uncertainty, privacy, possible impact on other family members and the inevitable generation of data not immediately relevant to the clinical indication for sequencing. At the time of testing, the patient should be made aware that, regardless of the specific indication for testing, laboratories will routinely analyze the sequence of a set of genes deemed to be highly medically actionable so as to detect pathogenic variants that may predispose to a severe but preventable outcome.

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- Patients should be informed during the consent process that, if desired, they may "opt out" of such analysis. However, they also should be made aware at that time of the ramifications of doing so.
- In accordance with the recent recommendations of the Presidential Commission for the Study of Bioethical Issues, as well as a lack of clear consensus in the ACMG membership survey administered in January, 2014, the board recommends that the same policy should be adhered to in children as in adults; *i.e.*, routine analysis of a set of selected genes to identify pathogenic variants associated with severe but preventable disease should be routinely performed. Parents should have the option during the consent process to opt out of such analysis in their children.
- At this time, given the practical concerns and inherent difficulty of counseling patients about the features of each disorder and gene on an ever-changing list, it is not feasible for patients to be offered the option of choosing only a subset of medically actionable genes for analysis. Thus, the decision regarding routine analysis should apply to the entire set of genes deemed actionable by the ACMG.

The **ACMG Policy Statement** concludes, "The ACMG recognizes the complex nature of policies surrounding genome-scale testing and that positions will continue to evolve and change in response to new knowledge, new technologies, and ongoing input and discussion with our membership and the broader medical community. The ACMG will continue to explore these issues in the best interest of patients. A multidisciplinary working group has been formed to develop a process for updating and maintaining the list of genes to be routinely analyzed for secondary findings."

The **ACMG Survey Results article** concluded that, "according to its membership, the ACMG should continue to update a minimal list of medically, actionable genes to be assessed when clinical exome or genome sequencing is performed, but informed consent is necessary and reporting of secondary findings should be optional. More research is needed to understand the multi-level factors that may influence optimal implementation of reporting secondary findings."

Maren T. Scheuner, MD, MPH, FACMG, lead author of "Reporting Genomics Secondary Findings: ACMG Members Weigh In," said about the ACMG survey results, "The survey of ACMG members corroborates many of the original ACMG recommendations about secondary findings. Most agreed that seeking and reporting of secondary findings in the ACMG list of genes is consistent with medical standards, has sufficient evidence, and for adults, the benefits generally outweigh potential harms. However, there was lack of agreement regarding benefits versus harms for children, and lack of agreement about the potential impact on healthcare resources. Contrary to the original 2013 ACMG recommendations, the majority of respondents agreed that patient preferences regarding seeking and reporting of seconding findings should be considered when clinical exome or genome sequencing is pursued, including the ability of patients to opt out of receiving a sequencing report with such findings." (Earlier in 2014, ACMG did, in fact, update its initial recommendations to include an Opt Out option.)

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Editorial Note: ACMG is adopting the use of the term *Secondary Findings* instead of *Incidental Findings* based on the Presidential Commission for the Study of Bioethical Issues report, "Anticipate and Communicate: Ethical Management of Incidental and Secondary Findings in the Clinical, Research and Direct-to-Consumer Contexts" and the subsequent "Clinician Primer: Incidental and Secondary Findings."

According to the Presidential Commission, "*a secondary finding* is not the primary target of the test of procedure; rather, it is an additional result actively sought by the practitioner. Secondary findings might be deliberately sought when doing so is recommended by an expert body or by a consensus of practitioners." (Source: Clinician Primer: Incidental and Secondary Findings 2014.) The Clinician Primer goes on to refer to the ACMG Recommendations on Incidental Findings to be a specific example of *Secondary* Findings.

ACMG intends to use the term Secondary Findings in the future while not retroactively changing earlier documents and recommendations, which had used the term "Incidental Findings."

About the ACMG and ACMG Foundation

Founded in 1991, the American College of Medical Genetics and Genomics (www.acmg.net) advances the practice of medical genetics and genomics by providing education, resources and a voice for more than 1750 biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other healthcare professionals, nearly 80% of whom are board certified in the medical genetics specialties. ACMG is the only nationally recognized medical organization dedicated to improving health through the practice of medical genetics and genomics. The College's mission includes the following goals: 1) to define and promote excellence in the practice of medical genetics and genomics and to facilitate the integration of new research discoveries into medical practice; 2) to provide medical genetics and genomics education to fellow professionals, other healthcare providers, and the public; 3) to improve access to medical genetics and genomics services and to promote their integration into all of medicine; and 4) to serve as advocates for providers of medical genetics and genomics services and their patients. *Genetics in Medicine*, published monthly, is the official ACMG peer-reviewed journal. ACMG's website (www.acmg.net) offers a variety of resources including Policy Statements, Practice Guidelines, Educational Resources, and a Find a Geneticist tool. The educational and public health programs of the American College of Medical Genetics are dependent upon charitable gifts from corporations, foundations, and individuals through the ACMG Foundation for Genetic and Genomic Medicine (www.acmgfoundation.org.)

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