Should Secondary Findings in Genetics Be Used for General Population Screening? 
A Statement of the American College of Medical Genetics and Genomics (ACMG)

BETHESDA, MD – April 25, 2019— The American College of Medical Genetics and Genomics (ACMG) has previously published recommendations on the reporting of secondary findings in clinical exome and genome sequencing. Secondary findings are health-related interpretations of a patient’s genetic code that are unrelated to the primary reason for ordering the original testing. In order to promote standardized reporting of medically actionable information from clinical genomic sequencing, the ACMG in 2013, and then again in 2016, published a minimum list of genes to be reported as secondary findings during exome or genome sequencing (ACMG 56™, ACMG SF v2.0™). This list of recommended secondary findings to be reported reflects ACMG’s commitment to giving the genetics community a voice about which genes to include to further the goal of providing people information that can have a huge impact on their health outcomes.

Now, the ACMG has published a follow-up statement regarding whether secondary findings in genetics should be used for general population screening: “The Use of ACMG Secondary Findings Recommendations for General Population Screening – a Statement of the American College of Medical Genetics and Genomics.”

The ACMG statement stresses that the recommended list of genes for inclusion in reporting to patients was not validated or intended for general population screening. ACMG’s new statement clearly states that the use of ACMG SFv2.0 for purposes other than reporting incidental findings after clinical sequencing is not endorsed by ACMG.

"The reporting of secondary findings raises many difficult questions and ACMG has been a leader in addressing these issues. New challenges arose when ACMG SF v2.0 was extrapolated for use in the general population. The ACMG Board of Directors felt it was important to address concerns raised by many members in such situations,” said ACMG President Tony Gregg, MD, MBA, FACOG, FACMG, co-author of the new ACMG statement.

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The document also states:

- The ACMG strongly discourages any reference to the term ACMG SF v2.0 or ACMG 59 outside of the reporting of incidental findings after clinical sequencing.

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- The ACMG encourages further ascertainment of genotype-phenotype correlations and research to establish the efficacy of interventions in asymptomatic patients with pathogenic and likely pathogenic variants in known associated genes.

"The use of ACMG secondary findings recommendations for general population screening – a statement of the American College of Medical Genetics and Genomics" is available at: https://www.nature.com/articles/s41436-019-0502-5

**About the American College of Medical Genetics and Genomics (ACMG) and ACMG Foundation**

Founded in 1991, the American College of Medical Genetics and Genomics (ACMG) is the only nationally recognized medical society dedicated to improving health through the clinical practice of medical genetics and genomics. The ACMG provides education, resources and a voice for more than 2,300 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. The College's mission is to develop and sustain medical genetics-related initiatives in clinical and laboratory practice, education and advocacy.

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Four overarching strategies guide ACMG’s work: 1) to reinforce and expand ACMG’s position as the leader and prominent authority in the field of medical genetics and genomics while educating the medical community on the significant role that genetics and genomics will play in understanding, preventing, treating and curing disease; 2) to secure and expand the professional workforce for medical genetics and genomics; 3) to advocate for the specialty; and 4) to provide best-in-class education to members and nonmembers. *Genetics in Medicine*, published monthly, is the official ACMG peer-reviewed journal. ACMG’s website (http://www.acmg.net/) offers resources including policy statements, practice guidelines, educational programs and a ‘Find a Genetic Service’ tool. The educational and public health programs of the ACMG are dependent upon charitable gifts from corporations, foundations and individuals through the ACMG Foundation for Genetic and Genomic Medicine.

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