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The ACMG Releases 2025 Update to Secondary Findings Gene List; SF v3.3

BETHESDA, MD – July 9, 2025 | The American College of Medical Genetics and Genomics (ACMG) has released its highly anticipated 2025 update to the recommended minimum gene list for the reporting of secondary findings (SF) in clinical exome and genome sequencing: "<u>ACMG SF v3.3 List for Reporting of Secondary Findings in Clinical Exome and Genome Sequencing: A Policy Statement of the American College of Medical Genetics and Genomics.</u>"

As part of its ongoing commitment to annual updates, the ACMG Secondary Findings Working Group (SFWG) has added three new genes, *ABCD1*, *CYP27A1*, and *PLN*, to the SF v3.3 list. Each gene was carefully reviewed and selected based on actionability and potential impact on patient care.

Three genes, *GCK*, *RUNX1*, and *SLC4A1*, were considered for inclusion on the v3.3 list but were not added. One nominated gene, *ADA2*, was also considered for inclusion on the v3.3 list; however, the SFWG deferred a decision on its inclusion until additional information from the nomination submitters or other published data becomes available to inform a vote. A description of the criteria and rationale for inclusion accompanies each gene in the policy statement.

"The v3.3 list includes a small number of changes, consistent with our working group's goal of maintaining a minimum list of actionable results," said David T. Miller, MD, PhD, FACMG, co-chair of the ACMG SFWG and co-author of the policy statement. "Our goal is to ensure that any updates are measured, evidence-based, and continue to prioritize patient benefit. The collaborative, consensus-driven process behind this year's update reflects the high bar we've set for determining what is considered actionable."

The 2025 update also introduces a new publicly available<u>ClinGen webpage</u> to aid clinical labs in determining whether specific variants should be reported.

"As part of the SF v3.3 list, the SFWG has continued our collaboration with the Clinical Genome Resource (ClinGen) to introduce a new webpage that will serve as a resource for the community," said Christa L. Martin, PhD, FACMG, co-chair of the ACMG SFWG and co-author of the policy statement. "It provides guidance for determining whether specific variants or variants in specific gene-disease pairs are reportable, which will make it easier to access up-to-date information."

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The ACMG first established its recommended list of secondary findings in 2013, with the goal of ensuring that laboratories report known pathogenic or likely pathogenic variants in a defined set of medically actionable genes, even when unrelated to the primary indication for testing. These updates help ensure that patients undergoing clinical sequencing continue to benefit from advances in genomic medicine.

The ACMG SFWG includes experts from across the genetics and medical community, including clinical geneticists, molecular and/or cytogenetics laboratory directors, genetic counselors, cardiologists, a bioethicist, and a pediatrician who also serves as a patient advocate.

The ACMG SFWG will continue to review the current list of actionable genes, as well as new nominations throughout the course of the year including nominations submitted through representatives of other professional organizations and individuals via the <u>nomination form on the ACMG website</u>.

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 a prominent authority in the field of medical genetics and genomics and the only nationally recognized medical professional organization solely dedicated to improving health through the practice of medical genetics and genomics. The only medical specialty society in the US that represents the full spectrum of medical genetics disciplines in a single organization, the ACMG provides education, resources and a voice for more than 2,500 clinical and laboratory practice of medical genetics as well as through advocacy, education and clinical research, and to guide the safe and effective integration of genetics and genomics into all of medicine and healthcare, resulting in improved personal and public health. *Genetics in* Medicine and the new Genetics in Medicine Open, a gold open access journal, are the official ACMG journals. ACMG's website, <u>www.acmg.net</u>, offers resources including policy statements, practice guidelines, and educational programs. The ACMG Foundation for Genetic and Genomic Medicine works to advance ACMG educational and public health programs through charitable gifts from corporations, foundations and individuals.

Editor's Note: the versioning nomenclature of the ACMG SF list was designed to differentiate major versus minor revisions. Major revisions include conceptual changes to the categories or genes/variants in the SF list or the removal or addition of a large number of genes in a single update. These major changes are denoted by updating the version number to the next integer, e.g., v4.0 or v5.0. Minor revisions, such as the 2023 release of SF v3.2, reflect the addition or removal of one or a few genes or variants without any significant policy change.

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