ACMG STATEMENT | Genetics inMedicine

The use of ACMG secondary findings recommendations for general population screening: a policy statement of the American College of Medical Genetics and Genomics (ACMG)

ACMG Board of Directors[1]

Disclaimer: This statement is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this statement is completely voluntary and does not necessarily assure a successful medical outcome. This statement should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen.

Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this statement. Clinicians also are advised to take notice of the date this statement was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.

Keywords: secondary findings; population screening; ACMG SF v2.0; ACMG 56

Genetics in Medicine (2019) 21:1467–1468; https://doi.org/10.1038/s41436-019-0502-5

The American College of Medical Genetics and Genomics (ACMG) has previously published policy statements on the reporting of secondary findings in clinical exome and genome sequencing (ACMG SF v1.0 and ACMG SF v2.0), also known as the "ACMG 56" and "ACMG 59," respectively.[1,2] These recommendations specifically stated that "reporting some incidental [a.k.a. secondary] findings would likely have medical benefit for the patients and families of patients undergoing ***clinical sequencing***" (ACMG board's emphasis). The ACMG SF v2.0 list of genes was not validated for general population screening. The use of ACMG SF v2.0 for purposes other than reporting incidental findings after clinical sequencing is not endorsed by ACMG. Many of the ACMG SF v2.0 genes have uncertain penetrance when identified in asymptomatic individuals (e.g., *SCN5A* and Brugada syndrome).[3] This policy statement is meant to reduce unproven interventions based solely on genotype information. In the absence of penetrance data that can only be obtained through robust genotype–phenotype correlation, the medical ethical principle of nonmaleficence should dominate.

- 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.
- Further, ACMG SF™, ACMG 59™, ACMG 56™, and related words and designs incorporating ACMG™, are trademarks of the American College of Medical Genetics and Genomics and may not be used without permission.
- The ACMG encourages further ascertainment of genotype–phenotype correlation and research to establish the efficacy of interventions in asymptomatic patients with pathogenic and likely pathogenic variants in known associated genes.

DISCLOSURE

The authors declare no conflicts of interest.

REFERENCES

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The Board of Directors of the American College of Medical Genetics and Genomics approved this statement on 16 October 2018

Submitted 15 March 2019; accepted: 18 March 2019
Published online: 25 April 2019

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