Genetic Science Spotlight

ACMG: Recommendations for Reporting of Secondary Findings in Clinical Exome and Genome Sequencing

A revision of terminology from “incidental findings” to “secondary findings” has been made by the ACMG, when a list of genes are now intentionally analyzed for individuals who undergo clinical exome or genome sequencing. In November 2016, an update of Recommendations of Secondary Findings was published by the ACMG, revising the list and bringing it to a current total of 56 genes. Genes included are 1) of high penetrance for associated disorders, 2) having effective confirmatory tests available, and 3) indicative of medical interventions to prevent or reduce morbidity or mortality. In the latest update, the *MYLK* gene associated with familial thoracic aortic aneurysm and dissection was removed due to the lack of an effective confirmatory test, screening modality, or intervention to prevent or reduce morbidity or mortality. In addition to genes associated with high penetrance for actionable phenotypes, ACMG is also developing a list of pharmacogenomic variants focusing on coding variants related to commonly prescribed medications as well as medications associated with serious adverse events. Anyhow, before result reporting, patients are entailed to be sufficiently educated on the medical, legal, social, and economic implications of any secondary findings, and should be provided with the option of declining secondary finding analysis after appropriate counseling.

http://www.nature.com/gim/journal/vaop/ncurrent/pdf/gim2016190a.pdf

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