First Look: Use of Public Human Genetic Variant Databases to Support Clinical Validity for Genetic and Genomic-Based In Vitro Diagnostics (FDA Guidance)

Provides guidance on best practices for genetic variant databases as used for next-generation sequencing tests and details information on applying for FDA recognition of a genetic variant database.

Updated last **April 27, 2018**
for the 04/13/2018 final guidance.

WHAT IT DOES

This guidance, issued by the Food and Drug Administration (FDA), describes the usefulness of a genetic variant database in scientifically supporting future regulatory reviews of next-generation sequencing tests. The guidance specifically outlines predicted best practices and characteristics of genetic variant databases that will likely attain regulatory support. For instance, these databases should function in a transparent and publicly accessible manner, providing adequate information about and assurances for the quality of the data. The databases should also provide evidence that the genetic variant is linked to a disease or condition. The FDA will determine if there is validity to the claims that the data supports a clinically significant result. These databases will collect, preserve, and report genetic variant data following regulations to protect patient privacy and data security.

This guidance applies to genetic variant databases regardless of the technology used (e.g., Sanger sequencing, PCR, next generation sequencing). Emphasis is placed on scientific validity, including the suggestion of written standard operating procedures that are performed by at least two qualified individuals. Administrators of publicly-available genetic variant databases meeting data quality standards may apply to the FDA for official recognition of their database. This application process is outlined in the guidance.

RELEVANT SCIENCE

Sequencing is increasingly used to identify differences in individuals' DNA called genetic variants. Some, but not all, genetic variants can predict increased or decreased risk of diseases and conditions. However, a genetic variant associated with a disease or condition does not always mean the individually will develop the disease or condition.

STATUS

The final guidance was issued by the FDA on April 13, 2018. This is a non-legally binding guidance document.

PRIMARY AUTHOR

Jacqueline Robinson-Hamm, PhD Candidate

EDITOR(S)

Andrew Pericak, MEM

RECOMMENDED CITATION

Duke SciPol, "First Look: Use of Public Human Genetic Variant Databases to Support Clinical Validity for Genetic and Genomic-Based *In Vitro* Diagnostics (FDA Guidance)" available at

http://scipol.duke.edu/content/first-look-use-public-human-genetic-variant-databases-support-clinical-validity-genetic-and
(04/27/2018).

LICENSE