FDA's New Action to Advance the Development of Reliable Genetic Tests

The agency clarifies how test developers may use clinical evidence in public databases to show that their test is "clinically valid."

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FDA takes new action to advance the development of reliable and beneficial genetic tests that can improve patient care

The U.S. Food and Drug Administration today took a significant step forward in driving the efficient development of novel diagnostic technologies that scan a person's DNA to diagnose genetic diseases and guide medical treatments. For the first time, the agency has formally recognized a public database that contains information about genes, genetic variants and their relationship to disease. The FDA is recognizing the genetic variant information in the Clinical Genome Resource (ClinGen) consortium's ClinGen Expert Curated Human Genetic Data, which is funded by the National Institutes of Health (NIH), as a source of valid scientific evidence that can be used to support clinical validity in premarket submissions. The information contained in this open database has been collected and studied by researchers across the world. This recognition by the FDA will facilitate test developers, including those that use a technology known as next generation sequencing, to rely on the information available in the database to support the validity of their tests, instead of having to generate the information on their own.

"Technological and clinical advances in genetic tests mean that patients and providers have a better understanding of the causes of disease and potential treatment options. The availability of genetic tests is opening up new opportunities to segment illnesses into more treatable subsets and enabling the development of targeted therapeutics aimed at these previously unknown categories of disease. These new medicines increasingly show outsized benefits in small populations of patients with rare, hard-to-treat and sometimes fatal conditions. The ability to use diagnostics to identify these rare subsets is a key element in driving this transformation in medical care and drug development," said FDA Commissioner Scott Gottlieb, MD. "The FDA is committed to supporting the development of these novel technologies that provide critical, potentially life-saving information. With our policies, we've sought to create an efficient regulatory pathway that promotes the more rapid development of important tests that can transform medical care while assuring their accuracy and clinical relevance. Our continued efforts, including today's recognition, will advance this process and help patients gain access to more sophisticated tests that provide important genetic information, allowing for more targeted medical care."

Genetic tests work by looking at a person's DNA to detect genomic variations that may determine whether a person has or is at risk of developing a genetic disease and, in certain cases, may help to inform treatment decisions. Unlike traditional diagnostics that typically detect chemical changes associated with a single disease or condition, DNA-based assays can look at hundreds to millions of DNA changes in a single test to help determine the cause of a person's disease or condition.

Availability of these types of tests plays an important role in advancing how clinicians and researchers learn about diseases, how innovators develop new treatments, and how doctors improve patient care.

Most genetic data are not aggregated and stored in a manner that is accessible to many researchers and clinicians. Recognizing the benefits of public databases, the FDA issued a final guidance in April 2018 to help accelerate the development of reliable and beneficial genetic tests. The agency's policies seek to encourage data sharing and outline an approach clarifying how test developers may rely on clinical evidence provided in FDA-recognized public databases to support clinical claims for their tests and help provide assurance that the test is "clinically valid," which, in the case of genetic tests, is the relationship between a gene variation and a specific disease.

Today's action — which recognizes the ClinGen consortium's ClinGen Expert Curated Human Genetic Data as a source of valid scientific evidence that can support clinical validity — means that developers of genetic tests have assurance of the reliability of the freely available data that they can use in support of their applications for marketing authorizations with the agency, rather than generating the same data on their own. The new recognition means developers will not need to demonstrate to the FDA the reliability of the database, the data and information within which can be used to support the relationship between a gene variation and a specific disease that are within the scope of the recognition. For example, the sponsor of a test that detects variants involved in hereditary cardiomyopathy could point to the cardiomyopathy genetic variant information available in ClinGen as part of a submission to support clinical validity of their test.

"A major current challenge for precision medicine is the need to translate new discoveries and data from the Human Genome Project so that this information can be used by physicians and other health care providers to improve health," said NIH Director Francis S. Collins, MD, PhD. "ClinGen provides a standard curated data reference of genetic variants to facilitate the development and implementation of genetic tests for use by health care professionals, which is critically important for moving science into practice."

The FDA recognized the database using the process detailed in the April 2018 final guidance. The agency reviewed ClinGen's standard operating procedures and policies, including processes and validation studies for variant evaluation, data integrity and security, and transparency of all evidence. The FDA also reviewed the policies for how the consortia qualifies and approves researchers and clinicians to evaluate variants, including conflict of interest and disclosure policies.

ClinGen brings together more than 700 clinical and research experts to develop standard processes for reviewing data and genetic variants and their connections to health and disease. The

experts who are part of the consortium determine how each variant is associated with a specific hereditary disease or condition and make that information available for unrestricted use in the community.

In its recognition of ClinGen, the FDA reviewed variant classifications and the processes that support them for gene changes in reproductive cells (germline variant) in hereditary disease where there is a high likelihood that the disease or condition will materialize if the gene is altered. Genetic tests may use germline variant information to detect for cardiomyopathy, hearing loss, inborn errors of metabolism and other hereditary conditions.

The FDA’s recognition anticipates that ClinGen may add new or modify existing genetic variant information on an ongoing basis, provided they are within the scope of recognition.

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