



December 4, 2018

Julianne O'Daniel
ClinGen Database Coordinator
The University of North Carolina at Chapel Hill
Department of Genetics, CB#7264
Chapel Hill, NC 27599

Re: Q181150
ClinGen Expert Curated Human Variant Data
Dated: June 22, 2018
Received: June 25, 2018

Dear Ms. O'Daniel:

The Center for Devices and Radiological Health (CDRH) of the Food and Drug Administration (FDA) has completed its review of your application for Recognition of ClinGen Expert Curated Human Variant Data. We are pleased to inform you that ClinGen Expert Curated Human Variant Data is recognized with the following scope:

Germline variants for hereditary disease where there is a high likelihood that the disease or condition will materialize given a deleterious variant (i.e., high penetrance).

This recognition does not constitute marketing clearance or approval of this product as a medical device, and does not affect a previous clearance or approval of a device.

Data from FDA-recognized genetic variant databases would generally constitute valid scientific evidence that can be used to support the clinical validity of the genotype-phenotype relationships embodied in the assertions from such databases provided in a premarket submission. Under this policy, FDA expects that test developers will be able to use FDA-recognized genetic variant databases to establish, at least in part, the clinical validity of their test, provided that the evidence supports the intended use of the test. For premarket submissions that rely upon genetic variant databases recognized by FDA, the Agency may determine that submission of additional valid scientific evidence for certain variant assertions found in these genetic variant databases is necessary, depending on the sufficiency of the evidence for these assertions to support the intended use.

CDRH will notify the public of its decision to recognize ClinGen Expert Curated Human Variant Data. You have provided consent for FDA to make public certain information regarding this recognition and further you have committed to make all recommended documents publicly accessible on the genetic variant database's website at the point of recognition.

You may request that CDRH incrementally expand or otherwise modify the recognized portion of your genetic variant database by submitting a new recognition package. If you do, please include a reference to the submission number shown at the top of this letter. CDRH also intends to reconsider recognition decisions as appropriate. For example, If the genetic variant database is not maintained according to the specifications under which it was recognized, FDA may reconsider or withdraw recognition.

If you have any questions concerning this recognition decision letter, please contact Brittany Schuck at 301-796-5199 or Brittany.Schuck@fda.hhs.gov.

Sincerely yours,

Courtney H. Lias, Ph.D.
Director
Division of Chemistry and Toxicology Devices
Office of In Vitro Diagnostics and Radiological Health
Center for Devices and Radiological Health