Hogan Lovells

FDA Cleared First DTC Genetic Tests for Health Risks

17 April 2017

Last week, the U.S. Food and Drug Administration (FDA) granted marketing authorization for 23andMe's Personal Genome Service (PGS) Test for 10 diseases or conditions.[1] This was the first FDA authorization for a direct-to-consumer (DTC) genetic test that provides information on personal risk for certain diseases.

The 23andMe PGS Test uses qualitative genotyping to detect the clinically relevant variants in genomic DNA isolated from human saliva collected from individuals over 18 years for the purpose of reporting and interpreting Genetic Health Risks (GHR). Specifically, the test is intended to detect genetic variants that are associated with an increased risk for developing the following 10 diseases or conditions:

- Parkinson's disease
- Late-onset Alzheimer's disease
- Celiac disease
- Alpha-1 antitrypsin deficiency
- Early-onset primary dystonia
- Factor XI deficiency
- Gaucher disease type 1
- Glucose-6-Phosphate Dehydrogenase deficiency
- Hereditary hemochromatosis
- Hereditary thrombophilia

FDA emphasizes that such GHR tests are intended to provide genetic risk information to consumers, but cannot determine a person's overall risk of developing a disease or condition, and therefore cannot be used for diagnostic purposes. Consistent with this view, the indications for use statement has been carefully crafted for each disease/condition, which specifies the particular variant(s) being tested and includes limiting language on how the results can be interpreted. For example, for hereditary thrombophilia, the following indications for use language is used:

> *"The 23andMe PGS Genetic Health Risk Report for Hereditary Thrombophilia is indicated for reporting of the Factor V Leiden variant in the F5 gene, and the Prothrombin G20210A*

[1] https://www.fda.gov/NewsEvents/Newsroom/PressAnnouncements/ucm551185.htm; See also the reclassification letter: https://www.accessdata.fda.gov/cdrh_docs/pdf16/DEN160026.pdf.

> *variant in the F2 gene. This report describes if a person has variants associated with a higher risk of developing harmful blood clots, but it does not describe a person's overall risk of developing harmful blood clots. This test is most relevant for people of European descent."*

Similar wording is used for the other diseases/conditions. It should be noted, however, that the claim highlights the risks associated with markers in developing a condition, but **not** in developing the disease symptoms arising from each condition, a distinction that may well be lost on patients. In addition, while each indication identifies a population where the results have greatest relevance, no population is excluded.

The authorization was granted under the *de novo* pathway, a regulatory pathway for novel, low-to-moderate-risk devices that are not substantially equivalent to an already marketed device. Devices that are classified through the *de novo* pathway may be used as predicates for future 510(k) submissions. For 23andMe's PGS test and other similar GHR tests, FDA created a new device classification: Genetic Health Risk Assessment System.

> *A genetic health risk assessment system is a qualitative in vitro molecular diagnostic system used for detecting variants in genomic DNA isolated from human specimens that will provide information to users about their genetic risk of developing a disease to inform lifestyle choices and/or conversations with a healthcare professional. This assessment system is for over-the-counter use. This device does not determine the person's overall risk of developing a disease.*

In addition, in the press release, FDA indicated that additional 23andMe GHR tests may be exempt from premarket review, and that GHR tests from other manufacturers may be exempt after submitting their first premarket notification. However, such future exemptions do not apply to diagnostic tests. The tests must also use a sample collection device that is FDA-cleared, approved, or exempted, with an indication for IVD use in OTC DNA testing.

Contacts

Randy J. Prebula
Partner, Washington, D.C.
Tel +1 202 637 6548
randy.prebula@hoganlovells.com

Erkang Ai, Ph.D.
Associate, Philadelphia
Tel +1 267 675 4631
erkang.ai@hoganlovells.com