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23andMe authorized by FDA to market first direct-to-consumer genetic test

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Feb23andMe today announced that it has been granted authority by the U.S. Food and Drug Administration (FDA) to market the first direct-to-consumer genetic test under a regulatory classification for novel devices.

23andMe's Personal Genome Service 510(k) submission for Bloom Syndrome Carrier Status test report was evaluated through the de novo regulatory pathway. 23andMe submitted an application for review under the standard 510(k) requirements. Because of the first-of-its kind nature of a direct-to-consumer test, the FDA determined that 23andMe's submission did not have an applicable predicate device, and converted it to a de novo request. The agency took the further step to downclassify carrier status tests for autosomal recessive disorders.

The Food and Drug Administration Modernization Act of 1997 (FDAMA) added the de novo classification option, which provides an alternate path to classify novel devices of low to moderate risk that are not substantially equivalent to an already legally marketed device. Devices that are classified through the de novo process may be marketed and used as predicates for future 510(k) submissions.

In addition to the authorization to market the Bloom Syndrome Carrier Status test report, the FDA is also classifying autosomal recessive carrier screening tests as class II with the intention to exempt such carrier tests from FDA premarket review. According to the FDA's press release, the "agency plans to issue a notice that announces the intent to exempt these tests and that provides a 30-day period for public comment. This action

creates the least burdensome regulatory path for autosomal recessive carrier screening tests with similar uses to enter the market."

23andMe will not immediately begin returning Bloom syndrome Carrier Status test results or other health results to customers until it completes the regulatory process for additional test reports and can offer a more comprehensive product offering.

"This is a major milestone for our company and for consumers who want direct access to genetic testing," said Anne Wojcicki, 23andMe CEO and co-founder. "We have more work to do, but we remain committed to pursuing a regulatory path for additional tests and bringing the health reports back to the US market. This important first step would not have been possible without the hard work and guidance of the FDA."

23andMe met the FDA's premarket requirements to demonstrate the following: accuracy, validity and user comprehension. Specifically this process validated 23andMe's spit test kit and chip array platform for determining whether or not an individual is a carrier for the genetic markers for Bloom syndrome. In addition, 23andMe conducted extensive comprehension studies with consumers from different backgrounds, education levels and incomes.

An accuracy study was performed at two lab sites with seventy samples. The samples included sixty-five saliva samples and five human cell line samples with known BLMAsh variant status. Results of the PGS test for Bloom Syndrome were compared with sequencing results. The results of the study gave an agreement of 70 out of 70 samples.

For the validation study, a total of 2,880 sample replicates were run under standard 23andMe laboratory procedures. An additional study was performed using 105 saliva samples without the BLMAsh variant. The samples were tested by comparing results between the two 23andMe

laboratory sites.

A user comprehension study was performed to assess how well people understand the PGS Bloom syndrome carrier status test reports. A diverse group of people representative of US demography reviewed and answered questions about the test report in a moderator controlled setting. In all the studies, more than 90 percent of participants indicated they understood the test results.

"This regulatory process helped establish the parameters for consumer genetics. We are pleased with the Agency's decision and its affirmation that consumers can understand and benefit from direct access to genetic information," said Kathy Hibbs, 23andMe's chief regulatory and legal officer.

Bloom syndrome is a rare disorder that is more common in people of Central and Eastern European, or Ashkenazi, Jewish background. One in 107 people of Ashkenazi Jewish descent are carriers for Bloom syndrome[1]. The disorder is characterized by short stature, sun-sensitive skin changes and an increased risk of cancer. According to the National Institute of Health, this condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations.

The 23andMe Personal Genome Service (PGS) Carrier Screening Test for Bloom Syndrome is indicated for the detection of the BLMAsh variant in the BLM gene from saliva collected using an FDA cleared collection device (Oragene DX model OGD-500.001). This test can be used to determine carrier status for Bloom syndrome in adults of reproductive age, but cannot determine if a person has two copies of the BLMAsh variant. The test is most relevant for people of Ashkenazi Jewish descent.

For its unique DNA collection kits, 23andMe works exclusively with DNA Genotek, a subsidiary of OraSure Technologies, Inc (NASDAQ:OSUR), which has manufactured the Oragene Dx saliva collection kits for 23andMe since the company launched its Personal Genome Service in 2007.

More information: 1. Gross, S.J., Pletcher, B.A., Monaghan, K.G. (2008). ACMG Practice Guidelines:Carrier screening in individuals of Ashkenazi Jewish descent. *Genet Med*. 10(1):54–56.

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