EMPIRICA BIOSCIENCES

Empirica Biosciences Terms of Service and Privacy Statement

What is tested

Empirica Biosciences offers two types of tests. In the standard test, >20,000 variants known or likely to be associated with disease in ~1,800 genes are tested. In the premium test, the exons of all ~20,000 genes are tested for variants that indicate an increased risk of developing a disease. Exons are the parts of a gene that contain the information for making proteins. We are able to detect both well-characterized and novel genetic variants in the premium test, whereas the standard test only detects a specific set of well-known genetic variants.

How it is tested

Empirica Biosciences tests for genetic variants by collecting a saliva sample that you provide. This sample is transported to a contracted, accredited gene testing facility, where DNA is extracted and analysed. In the standard test, your DNA sample is tested using a technique known as microarray genotyping. This technique tests a specific set of genetic sites with known variants. The results of this test determine whether you have the variant or the reference ('normal') sequence at these sites. In the premium test, your DNA sample is prepared using a commercially available sequence capture method and the sequence is determined for all exons (protein-coding regions) of all genes, using a technique known as whole-exome sequencing. Using this technique, >99% of all known protein-coding regions are covered. Your DNA sequence information is then compared to an established reference genome sequence to identify genetic variants. Any genetic variants that are identified are then classified according to the American College of Genetics and Genomics (ACMG) criteria and using expert-curated clinical genetics databases, as benign (harmless), likely benign, pathogenic, likely pathogenic or of unknown significance. For both tests, your results are returned to you in the form of a comprehensive report outlining your disease risk. Only those variants that are classified as pathogenic or likely pathogenic are included in your report, as only these variants are likely to increase your disease risk. If no such variants are detected, your report will state "no pathogenic variants detected". If you opt for carrier screening, your results will be compared with your partner's results and screened against a database of recessive disease genes.

Information collected

We collect some basic personal information to allow us to provide the service you request. This includes your name, phone number, e-mail address, delivery address, gender, and date of birth. Knowing your gender and date of birth allows us to customize your report. If you consent to volunteer your results for research, we will email you a questionnaire requesting details of the medical history of you and your family. We will retain your contact information on file so that we can stay in touch with you and keep you updated on new service offerings, changes, or any other information that may be deemed relevant. You may opt out of this at any time, in which case, we will remove you from our mailing list and delete your personal information from our database. We will only share your contact information with other parties when required for the successful delivery of the service you have requested.

Quality control measures

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Empirica Biosciences takes steps along the service delivery pathway to ensure data quality is maintained. Saliva samples are collected in barcoded tubes to allow tracking throughout the testing process. Each sample is assigned at least two unique identifiers to ensure accurate sample tracking. Gene testing is performed in a certified genetic testing facility, in which procedures meet industry standards. Samples must pass DNA sample, DNA sequencing, and bioinformatics quality metrics before your results are analyzed and reported to you. The standard test is performed in a clinically certified laboratory and the premium test is performed in a laboratory accredited by the technology provider.

We regularly review the scientific literature, relevant databases, and medical association guidelines concerning all genes and their associated diseases included in our test to ensure that your report is based on the most up-to-date information available.

Benefits of the test

The information in your test report provides insights into how your genes affect your risk of developing certain diseases. This information can be useful for your healthcare provider so that they can make informed decisions on the most appropriate detection and preventative measures. This information could also be useful to family members, who may also have the same genetic predisposition as you.

Based on your results, you can take actions to mitigate the risk of developing certain diseases or undergo a more rigorous screening plan to detect disease at an earlier and more treatable stage. Research has shown that indiviuals with a genetic risk for heart disease can decrease this risk by 46% with lifestyle changes[1] or by 48% by taking certain medication[2]. Early detection of cancer is key to successful treatment and long-term survival. Knowing that you have a genetic predisposition to a certain type of cancer allows you to undergo specific targeted screening to detect the cancer, if it occurs, at an early stage. If you opt for carrier screening, you can identify your risk of passing on an inheritable disease to your children.

Limitations of the test

This test does not provide a disease diagnosis and does not replace or substitute a medical professional's advice. The presence of a pathogenic gene variant indicates an increased risk of disease. It does not mean that you will definitely develop the disease in your lifetime. Empirica Biosciences recommends that you consult with your healthcare provider or a genetic counsellor to determine how your results affect your ongoing medical care. We advise against making any changes to your current medication or to other aspects of your medical care without first consulting your healthcare provider.

This test focuses on the protein-coding regions of all genes (premium test) or a specific set of known disease-associated genetic variants (standard test). However, there may be other regions of the genome not specifically tested here that affect your disease risk. There may also be non-genetic factors that increase your risk of disease. The information included in your report is based on published research, current at the time of testing. New scientific data regarding genetic risk are continually being published in the scientific literature. This new information may affect the interpretation of your results.

While Empirica Biosciences will take all care to deliver accurate results, as with all tests, there is the risk of a false negative or a false positive result. While unlikely, the results could also be interfered with by poor sample quality, contaminants or technical errors in the laboratory.

Risks of the test

There are no physical risks associated with this test, as we use a non-invasive sample collection procedure that involves donating a saliva sample. This test may reveal sensitive information about hereditary conditions that you may have been previously unaware of and that may also be present in other family members. There may be other genetic causes of disease that are not specifically covered in this test. While you can take actions to lower your disease risk or take steps to help detect a disease early based on the results of this test, these diseases may not currently have curative treatments. Some disease risks identified may not be medically actionable.

Sample/data storage

Unless you have consented to participate in research, your saliva and DNA samples will be destroyed after sequencing results have been obtained. Your results will be kept confidential and will only be used for internal testing and quality control. They will not be shared with third parties without your prior consent. Your genetic and personal data will be stored in a secure, password-protected location and will be retained so that we can contact you as necessary. However, you may also choose to have all your data deleted from our system once you have received your report, by notifying us at info@empiricabiosciences.com.

Research

Empirica Biosciences aims to fill the information gap regarding genetic associations with disease in African populations. When you order a test, you have the opportunity to volunteer your results for research purposes to help us achieve this aim. If you choose to participate, you will be asked to complete a questionnaire regarding you and your family's medical history. By consenting to use your data for research, your sample may be retained and used for additional genetic analyses not included in this test. Additional use of your test data or additional analysis of your DNA sample may involve collaborations with third parties. In such cases, your data will be de-identified, meaning that any details that identify you, such as your name and contact information, will be removed. You can opt out of having your data used for additional research at any time by notifying us at info@empiricabiosciences.com.

Privacy

Your test results will be analyzed and stored using US Health Insurance Portability and Accountability Act[3] (HIPAA)-compliant cloud-based data storage and analysis systems. If you consent to your results being used for research purposes, your genetic test results and/or DNA sample may be analyzed further and these results may be published in scientific journals. In such cases, you will not be identified. Your name and contact information will not be shared with third parties without your prior consent. Your test results and personal information will be stored in a secure location and you may request their deletion at any time by notifying us at info@empiricabiosciences.com.

1. Khera AV, Emdin CA, Drakeet I, et al. Genetic risk, adherence to a healthy lifestyle, and coronary disease. *New England Journal of Medicine*. 2016;375:2349-2358.

2. Mega JL, Stitziel NO, Smith JG, et al. Genetic risk, coronary heart disease events, and the clinical benefit of statin therapy: an analysis of primary and secondary prevention trials. *Lancet*. 2016;385:2264–2271.

3. US Department of Health and Human Services Health Information Privacy. www.hhs.gov/hipaa/

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