STANFORD UNIVERSITY Research Consent Form

Protocol Director: Roxana Daneshjou

Protocol Title: Clinically Annotating Existing Genomes for Pathogenic Variants Based on the ACMG Recommendations for Reporting of Incidental Findings

Clinically annotating existing genomes for pathogenic variants based on the ACMG Recommendations for Reporting of Incidental Findings

FOR QUESTIONS ABOUT THE STUDY, CONTACT:

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DESCRIPTION: You are invited to participate in a research study on the prevalence of pathogenic or possibly pathogenic variants present in genes listed by the American College of Medical Genetics (ACMG) Recommendations for Reporting of Incidental Findings. This research study is looking for up to 2000 individuals with full genome or exome data.

You will be asked to download a software tool, which will scan your genome for variants present within the ACMG recommended genes and which meet ClinVar criteria for pathogenicity. Please note that ClinVar variants are not universally agreed upon to be pathogenic. For a complete description of the genes that will be tested, please see: http://www.acmg.net/docs/ACMG_Releases_Highly-Anticipated_Recommendations_on_Incidental_Findings_in_Clinical_Exome_and_Genome_Sequencing.pdf

Scanning of your genome is done locally on your computer and at no point will any of your genomic data be transmitted beyond your local environment. At the end of the scan, you will have the option to transmit summary statistics to the research team. For example, if no variants are found, the research team will receive a report that a single genome was scanned and no variants were found. If variants are found, the research team will receive a report that a genome was scanned and variants X and Y were found. These numbers will be automatically added to a running tally of how many genomes have been analyzed with the frequency of the two variant classes. Additionally, you will have the option to submit your ancestry, but this is not required.

RISKS AND BENEFITS: There are no physical or economic risks association with participating in this study. You may, however, experience emotional or psychological distress from finding out that you carry a possibly pathogenic variant that affects your predisposition for a range of diseases such as cancer or significant cardiovascular disease. Moreover, there is a risk of a false sense of security if a variant is not reported; it is important to remember that

not all variants are reported through the use of this tool, and your genome may still carry a variant of unknown clinical significance in one or more of these genes. You should also remember that the genes assessed via this tool are very limited, and your genome may include pathogenic variants or low penetrance SNPs in other genes that may predispose to similar diseases.

The benefits of this study include finding out more about medically actionable genetic variants that you may or may not have. Prior to using our tool, we encourage you to look at the genes and associated conditions this tool will test: http://www.acmg.net/docs/ACMG_Releases_Highly-Anticipated_Recommendations_on_Incidental_Findings_in_Clinical_Exome_and_Genome_Sequencing.pdf. We cannot and do not guarantee or promise that you will receive any benefits from this study.

Your decision whether or not to participate in this study will not affect your employment/medical care/grades in school.

TIME INVOLVEMENT: Your participation in this experiment will take approximately however long it takes for you to download the tool and to scan your data – likely between 10 to 15 minutes

PAYMENTS: You will receive no payment for your participation.

PARTICIPANT'S RIGHTS: If you have read this form and have decided to participate in this project, please understand your participation is voluntary and you have the right to withdraw your consent or discontinue participation at any time prior to submission of summary statistics without penalty or loss of benefits to which you are otherwise entitled. Since we do not label or identify the summary statistics, we cannot withdraw them once they have been submitted. The results of this research study may be presented at scientific or professional meetings or published in scientific journals. However, your identity will not be disclosed**.**
You have the right to refuse to answer particular questions.

CONTACT INFORMATION:

Questions, Concerns, or Complaints: If you have any questions, concerns or complaints about this research study, its procedures, risks and benefits, or alternative courses of treatment, you should ask the Protocol Director, Roxana Daneshjou You may contact him/her now or later at 972-489-2311..

Injury Notification: If you feel you have been hurt by being a part of this study, please contact the Protocol Director, Roxana Daneshjou at(650) 725-3394.

Independent Contact: If you are not satisfied with how this study is being conducted, or if you have any concerns, complaints, or general questions about the research or your rights as a participant, please contact the Stanford Institutional Review Board (IRB) to speak to someone

independent of the research team at (650)-723-5244 or toll free at 1-866-680-2906. You can also write to the Stanford IRB, Stanford University, MC 5579, Palo Alto, CA 94304.

If you have any questions, concerns or complaints about this research study, its procedures, risks and benefits, or alternative courses of treatment, you should ask the Protocol Director, Roxana Daneshjou at (650) 725-3394. You should also contact him/her at any time if you feel you have been hurt by being a part of this study.

Please print a copy of this page for your records.

If you agree to participate in this research, please download the PATH-SCAN tool, analyze your genome, and select Submit to anonymously submit non-identifiable summary statistics about your data.