Application for a protocol to participate in the Intramural Secondary Genomic Findings Service (SGFS)

Is this a new application to the SGFS?

◯ Yes → Please complete all fields below

◯ No, this is an updated application as my former application has expired

Protocol Title/Number (if available):

IRB-reviewed version of protocol and consent forms are included with this application:

◯ Yes ◯ No - explain:

PI:

Protocol Precis:

Sequencing/Bioinformactics Details

Sequencing methodology: ☐ ES ☐ GS

Sequencing facility:

Aligner used to align reads:

Variant caller used to call variants:

Please confirm all of the following:

- ☐ Data submitted are aligned (or lifted over) to GRCh37 or hg19 reference genome
- ☐ We will only submit variants pertaining to the genomic coordinates provided by SGFS
- ☐ We will only submit one merged VCF file
- ☐ We will only submit "PASS" variants
- ☐ We will transfer the VCF using NIH secure email (NIH investigators) or Globus (non-NIH investigators)

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Project ID:

Application Acceptance Date:

Please note that we will accept a maximum batch size of 200 samples every 6 months for prospective application or a one-time batch of 300 samples per year in the case of requests for retrospective analysis. Please come to office hours or talk with our staff if your protocol exceeds these parameters.

What is the current status of the sequence data you plan to submit to the SGFS?

- ◯ We request retrospective analysis of *(fill in number)* participant samples.
- ◯ We request ongoing analysis of *(fill in number)* prospective participant samples per year.

When do you anticipate you will submit your first batch of data to the SGFS? Jan 2020

What is the TOTAL number of participant samples you are requesting the SGFS to analyze?

Over how many years?

Cohort Details

Is the disorder under investigation related to a gene on the current ACMG list?

◯ No ◯ Yes - explain:

Naming convention for individual identifiers (e.g., "6 digit coded identifier"):

Source of DNA: ◯ peripheral blood ◯ tissue ◯ other:

Where does your cohort primarily reside?

- ◯ Most participants live in the US
- ◯ Most participants live outside of the US - country:

Person(s) to notify if a secondary variant is present:

Name(s):	Email(s):	Phone number(s):

Will SGFS return secondary findings per the protocol outlined in the SGFS Guidance Memo?

◯ Yes ◯ No

If "Yes," please complete the contact information for the person(s) consenting participants to the protocol:

Name(s):	Email(s):	Phone number(s):

Any characteristics of your cohort/population that may have special relevance with respect to the return of secondary findings?

- ☐ High proportion of non-English speaking participants -- Language(s):
- ☐ Many participants unlikely to have access to healthcare
- ☐ Anticipate difficulty re-contacting participants
- ☐ Other:

Questions?

Please visit [http://genome.gov/sgfs] for more information. Contact Emily Modlin (301-435-6689) or Julie Sapp (301-435-2832) with any remaining questions. Please send completed applications along with an electronic version of the protocol and consent form to Julie Sapp [sappj@mail.nih.gov]. A protocol draft is acceptable if not yet approved by the IRB.

We will review this application and notify you of any changes requested or of acceptance to the SGFS. This application is valid for one year after the date of acceptance. If we do not receive any data after a year, we ask that you to fill out a new application to update us on any changes with your protocol before submitting data.

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