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 a submission of a new batch of data, all details from original application are unchanged.

→ remainder of application does not need to be filled out

◯ No -- This is a submission of a new batch of data but some project details have changed

→ complete only portions of application that have changed

Protocol Title/Number (if available):

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

◯ Yes ◯ No - explain:

PI:

Precis:

Biesecker Lab Use Only
Project ID:
Batch ID:

Sequencing methodology: ☐ ES ☐ GS

Sequencing facility:

Aligner used to align reads:

Variant caller used to call variants:

Please confirm the following:

- ☐ Data submitted were aligned to GRCh37/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)

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

◯ No ◯ Yes - explain:

Naming convention for individual identifiers:

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

Total number of sequenced samples:

Where does your cohort primarily reside?

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

Estimated number of samples per year:

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

Name: Email: Phone:

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

◯ Yes ◯ No

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

- ☐ High proportion of non-English speaking participants -- language:
- ☐ 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 Kate Driscoll at (301) 594-1754 or Julie Sapp at (301) 435-6689 with any remaining questions.
Please send completed applications along with an electronic version of the draft protocol and consent form to: Kate Driscoll [kate.driscoll@nih.gov] and Julie Sapp [sappj@mail.nih.gov].

Biesecker Lab Use Only
Project ID:
Batch ID: