

## NHGRI-NIHCC-DDIR CCGO (Clinical Center Genomics Opportunity) Application

**Principal Investigator:**

**Institute:**

**Address:**

**Tel:**

**Email:**

**<sup>1</sup>Data Analyst:**

**Address:**

**Tel:**

**Email:**

**Name of Project:**

**Description of Project** (not to exceed 3 single spaced pages with 12 point font. Please carefully read the online document “Considerations in applying for the CCGO 1000exomes program”.)

1. Description of the disorder: what is known about the genetics (inheritance and any prior research efforts to discover cause), scientific importance, and how it fits into the larger program of the PI and the intramural mission.
2. Study subjects: what patients will be brought to the NIHCC and how study will take advantage of NIHCC phenotyping, description of subjects who will be sequenced (unrelated cases & controls, several large pedigrees, mixed approach) with numbers, if linkage will also be used. Provide an estimate of the time window that patients/samples would be available for this program. Please indicate if you are using freshly collected peripheral blood or some other DNA source.
3. Analytic approach: how exome data will be filtered to identify variants, what genetic model will be used (recessive, dominant, de novo, or combination thereof), what approaches are available in the lab to follow up variants with functional studies.
4. Data sharing and dissemination plans: clinical and phenotypic data to be submitted, timeline for dbGaP data deposition.

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<sup>1</sup> This is the scientist in the lab of the PI (or the PI her/himself) who will learn how to use NISC-supported software and tools to analyze the exome data for the primary variant(s).

5. Consent specifications (or plans for re-consent): permission for genomic sequencing, dbGaP data deposition, permission for follow-up visits, iterative phenotyping, and return of results, both primary and secondary.

**Name of Scientific Director:**

**Signature of SD to acknowledge support of project:** \_\_\_\_\_

**Name of Clinical Director:**

**Signature of CD to acknowledge support of project:** \_\_\_\_\_

**Signature of PI to agree to terms of project including IRB review, return of results policy, progress reports, and data submission policy, as described in “Considerations in applying for the CCGO 1000exomes program”:**

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