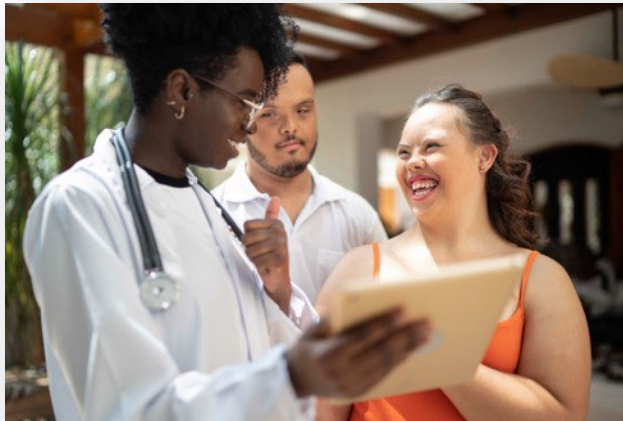


Pre-Application Webinar for the INCLUDE Omics Phenotypes Related to Down Syndrome



January 16 , 2024
1:00 PM ET

PAR-24-081

Omics Phenotypes Related to Down Syndrome for the INCLUDE Project (X01 Clinical Trial Not Allowed)



Notice of Funding Opportunity (NOFO) – PAR-24-081 – Purpose

Purpose

This Notice of Funding Opportunity (NOFO) invites applications to:

- Submit samples to NIH INCLUDE Project supported resources to generate a large volume of integrated **genomic** and **multi-omics** data that will facilitate discovery of the molecular mechanisms of health conditions related to Down syndrome.

Notice of Funding Opportunity (NOFO) – PAR-24-081

Specific Requirements And Expectations

- The resulting data, and associated clinical and phenotypic data, will become part of the INCLUDE Data Hub for sharing with the research community
- No funding will be provided directly to applicants under this NOFO
- Applicants are encouraged to propose omics assays of existing biospecimens collected from individuals with Down syndrome and controls
- Applicants are encouraged to propose cohorts of underrepresented racial and ethnic groups or to increase racial and ethnic representation of existing collections
- Projects selected under this NOFO will be expected to work in a collaborative manner with designated INCLUDE sequencing center(s)
- Data from the studies selected under this NOFO will be submitted to the INCLUDE DCC
- PDs/PIs of selected cohort projects will participate in a collaborative effort to inform the development of the integrated data resource to maximize its impact on the research community

X-01 Key Dates

Application Due Dates	Scientific Merit Review	Advisory Council Review	Earliest Start Date
March 13, 2024	May 2024	October 2024	December 2024
March 13, 2025	May 2025	October 2025	December 2025
March 13, 2026	May 2026	October 2026	December 2026

- Open Date (Earliest Submission Date) – February 13, 2024
- Letter of Intent Due Date(s) – 30 days prior to the application due
- Expiration Date – March 14, 2026

Page Limitations

- All page limitations described in the [How to Apply – Application Guide](#) and the [Table of Page Limits](#) must be followed
- For this specific NOFO, the Research Strategy section is limited to **6 pages**

Letter of Intent

- Not binding
- Information needed
 - Descriptive title of proposed activity
 - Name(s), address(es), and telephone number(s) of the PD(s)/PI(s)
 - Names of other key personnel
 - Participating institution(s)
 - Number and title of this funding opportunity

The letter of intent should be sent to:

Huiqing Li, PhD

National Heart, Lung and Blood Institute
(NHLBI)

Telephone: 301-435-0554

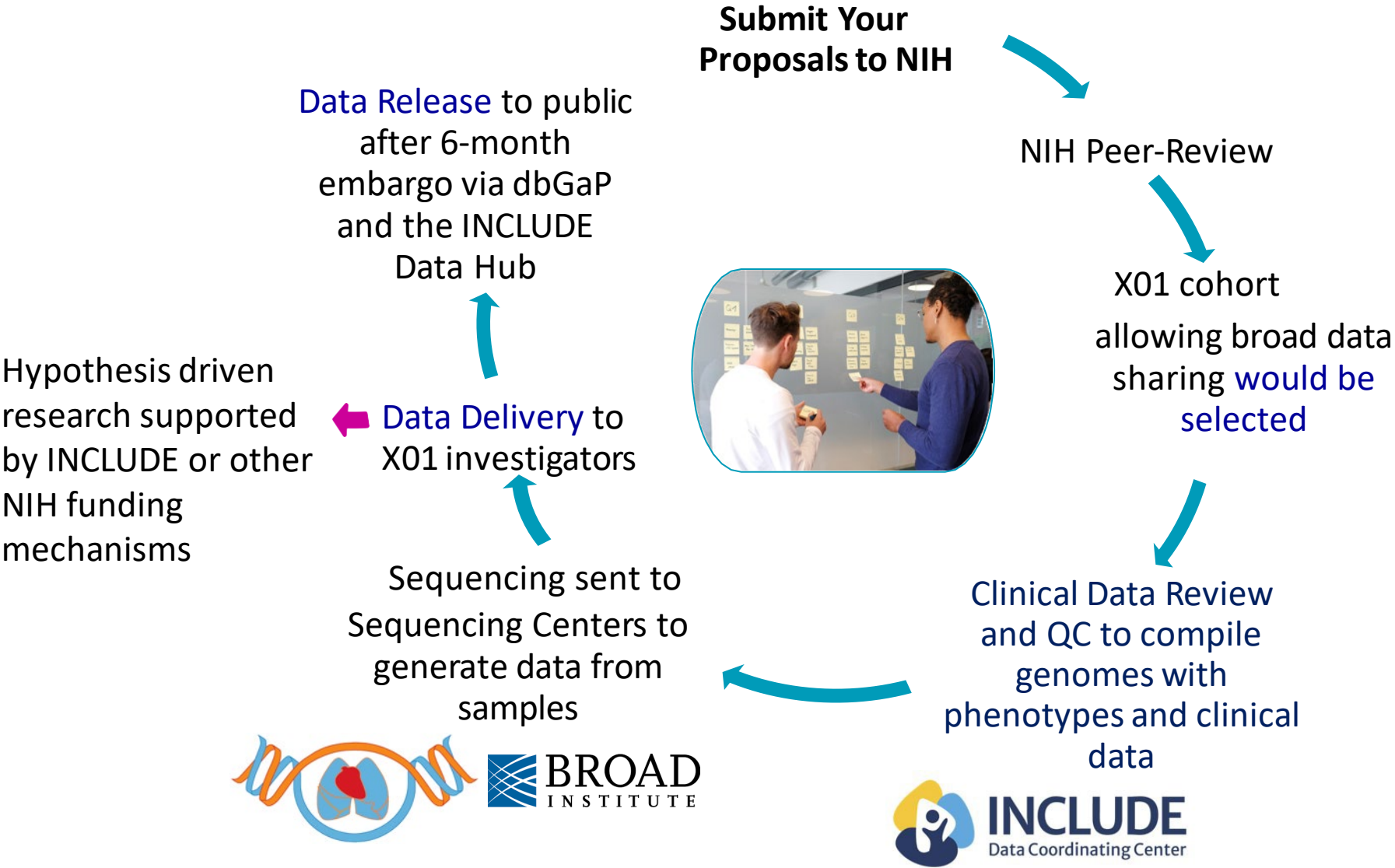
Email: huiqing.li@nih.gov

PAR-24-081 Updates

- ❑ **Data sharing is critical to the INCLUDE Project and the entirety of the X01 application, but there is no requirement for a separate Data sharing plan**
- ❑ **Research plan remains a requirement**
- ❑ **Representation, diversity, and inclusion are encouraged**
- ❑ **Description of **cohort, family tree, phenotypes, and clinical information** remains a requirement**
- ❑ **Description of **genomic data sharing through the INCLUDE Data Hub** remains a requirement**
- ❑ **Other attachments remains a requirement**
 - **Institutional Certification** (or provisional certification with data use limitations designations)
 - **Sample Information**
 - **Clinical/Phenotypic Data**
 - **Family Structure (Optional)**



Workflow after X01 Submission



Sequencing Centers

1. One of the Kids First Sequencing Centers: the Broad Institute

- a. Whole Genome Sequencing
- b. RNA Sequencing
- c. Long Read Sequencing

2. Leveraging [NHLBI TOPMed](#) for other omics:

- a. Methylomics
- b. Proteomics
- c. Metabolomics

NHLBI Trans-Omics for Precision Medicine Omics Centers

Short Name	Center Name	PI	Contact for Sample Submissions	Type of Sequencing
Baylor	Baylor College of Medicine Human Genome Sequencing Center	Richard Gibbs	Ginger Metcalf	WGS
Baylor-UTHealth Metabolomics	Baylor-UTHealth Metabolomics Center	Eric Boerwinkle	Eric Boerwinkle	Metabolomics
Broad Metabolomics	Broad Institute and Beth Israel Metabolomics Platform	Clary Clish	Julian Avila-Pacheco	Metabolomics
Broad BIDMC Proteomics	Broad Institute and Beth Israel Proteomics Platform	Robert Gerszten	Robert Gerszten	Proteomics
Broad Genomics	Broad Institute Genomics Platform	Stacey Gabriel	Namrata Gupta	WGS, RNASeq
Illumina	Illumina	Karine A. Viaud Martinez	Karine A. Viaud Martinez	WGS
Keck MGC	Keck Molecular Genomics Core Facility	David Van Den Berg	David Van Den Berg	Methylomics
MGI	McDonnell Genome Institute	Susan K. Dutcher	Kelley Foyil	WGS
NYGC Genomics	New York Genome Center Genomics	Soren Germer	Soren Germer	WGS
NYGC RNA-seq	New York Genome Center RNA-seq	Nicolas Robine	Nicolas Robine	RNASeq
NWGC	Northwest Genomics Center	Evan Eichler	Colleen Davis	WGS, RNASeq, Methylomics
Psomagen	Psomagen	Ryan Kim	Ryan Kim	WGS

Sample Information: Fillable Table

<u>DNA (and RNA) tissue of origin</u>	<u>Number of Samples</u>	<u>Extraction Method</u>	<u>Concentration</u>	<u>Quality (Metric used:[edit here to specify])</u>	<u>Method of Quantitation</u>	<u>Number of Samples Ready to Ship by August 2019</u>	<u>Number of Samples Ready to Ship by January 2020</u>
Blood							
Saliva or Buccal swab							
[other tissue, edit here to describe]. Note: cell lines will not be accepted							
Tumors or Affected Somatic Tissue							
DNA – Frozen Tissue							
RNA – Frozen Tissue							
DNA – Embedded Tissue							
RNA – Embedded Tissue							
Total							

*For tumor specimens or affected tissue samples, please also describe the fixation methods and the pathology review to which the specimens were subjected, separate from the table. For tumors, describe the percentage of tumor cells within the specimen used for DNA and/or RNA isolation and % necrosis.

Clinical and Phenotypic Data

Goal: Well-phenotyped data empowers analyses and informs how pathways/conditions overlap. The Data Coordinating Center (DCC) will leverage existing community standards to harmonize clinical/phenotypic data which facilitates searching, analysis, and interoperability with other data efforts.

Expectations:

- Basic data elements and deep phenotyping is preferred
- Describe what clinical/phenotypic information is available and how these data will:
 - Support your proposed analysis
 - Enhance research through the INCLUDE Data Hub

Biospecimen and Phenotypic Data Elements

- Sex, race, ethnicity, age at enrollment and/or diagnosis, diagnoses (e.g., type of co-occurring conditions), phenotypes for affected cases and unaffected family members, vital status, age at last known vital status, clinical information, and family medical history (e.g., family history of cancer or birth defects).
- **For templates and additional resources related to information required, please visit DCC's [Data Contributor Guide](#).**

Analysis Plans

Goal: Have investigators demonstrate that the proposed project has an adequate research design for genetic discovery, and that the X01 applicants are prepared to perform these analyses.

Expectations:

- While INCLUDE recognizes that analytical power will increase when the data from each individual study is incorporated with other data that will be part of the Data Hub, it is important to demonstrate that the data will be useable on its own
- Consider partnering with other research teams with relevant expertise to develop the analysis plan
- Researchers are highly encouraged to use the cloud-based infrastructure, including Cavatica, for their analysis

Institutional Certification Required

- 1) Download the current NIH Institutional (or Provisional) Certification template:
<https://osp.od.nih.gov/scientific-sharing/institutional-certifications/>
- 2) Fill out the first page, include all sites contributing samples for sequencing.
- 3) Provide the Institutional Certification to the IRB (or equivalent) along with the participant consent forms for each site and any other pertinent information.
- 4) The IRB reviews the consent form(s) to determine the data use limitations (DULs) and/or DUL modifiers for each consent form.
 - **“General Research Use” with no modifiers is expected for individual-level data, unless specific uses are clearly prohibited in the participant consent**
 - **“Unrestricted” access is expected for genomic summary results unless a justification is provided for designating the dataset as “sensitive”**
- 5) After IRB review, the Institutional Certification is signed by the appropriate officials and submitted to NIH.
[Download a letter](#) that explains these steps.

Data Sharing



Goals:

1. Make data generated by INCLUDE as accessible and usable as possible to the research community.
2. Enable researchers to easily combine/compare datasets for cross-disease analyses.

Expectations:

- Individual-level sequence and relevant phenotypic data are approved by the NIH to be shared through the INCLUDE Data Hub
- Samples that are consented in a way that allows broad access and use, including combining and cross-analyzing datasets (General Research Use, Health/Medical/Biomedical), will be prioritized

Visit [INCLUDE Data Sharing FAQs](#) for more information

Examples of Data Use Limitations and Modifiers Limit Broad Data Sharing

- **Disease Specific Consent Group:** When data use is restricted to a specific disease area, the data cannot be combined with a dataset with a different disease specific data use limitation. Combining and cross-analyzing datasets are a primary goal of Kids First and therefore datasets that are consented for General Research Use and/or Health/Medical/Biomedical purposes will be prioritized over datasets restricted to Disease Specific use.
- **IRB modifier:** With this box checked, the Requester must provide documentation of their local IRB's approval for the proposed research. We find that it is rare for consent language to include such a requirement and that this modifier is often included in error. As a reminder, every requester and their institution must agree to the terms of the Data Use Certification (DUC), which verifies that the requesting PI is accredited within the institution, the institution is aware of the project for which the PI is proposing to use the data, and that the Institution has all appropriate security measures in place to manage and maintain the controlled-access dataset(s) being retrieved. For a sample DUC, see: https://osp.od.nih.gov/wp-content/uploads/Model_DUC.pdf
- **COL modifier:** This box is checked when the consent form states that collaboration with the original/submitting investigator is required in order to use the dataset; therefore, the Requestor must provide a collaboration agreement document in order to be approved for access the dataset. This can limit the number of end-users who are able to use the dataset.

For more detail on DUL definitions:

https://osp.od.nih.gov/wp-content/uploads/NIH_PTC_in_Developing_DUL_Statements.pdf

<u>Site</u>	<u>Data Use Limitation</u> (GRU, HMB, DS)	<u>Data Use Limitation Modifiers</u> (IRB, PUB, COL, NPU, MDS, GSO)

Genomic Data Sharing Contacts

Valerie Cotton

valerie.cotton@nih.gov

Genomic Program Administrator (GPA)

Deputy Director of Data Science and Sharing
NICHD

General NIH Genomic Data Sharing

GDS@mail.nih.gov

dbGaP and the DCC

Data is made accessible through dbGaP and the INCLUDE Data Hub

- All INCLUDE projects are registered, authenticated, and approved through dbGaP.
- All dbGaP Data Access Requests will be processed by NIH Data Access Committees.
- INCLUDE Data Hub is the NIH designated repository for INCLUDE data.



INCLUDE's Cloud- Based Platforms: INCLUDE Data Hub



INCLUDE Data Hub - portal.includedcc.org

EXPLORE datasets and build cohorts of participants

DISCOVER harmonized genomic data files for further research

CONNECT data from multiple INCLUDE studies

CAVATICA - cavatica.sbgenomics.com

COMPUTE large scale workflows on genomic data files

ANALYZE data in the cloud via R Studio and Python Notebooks

SHARE tasks and findings with collaborators around the world



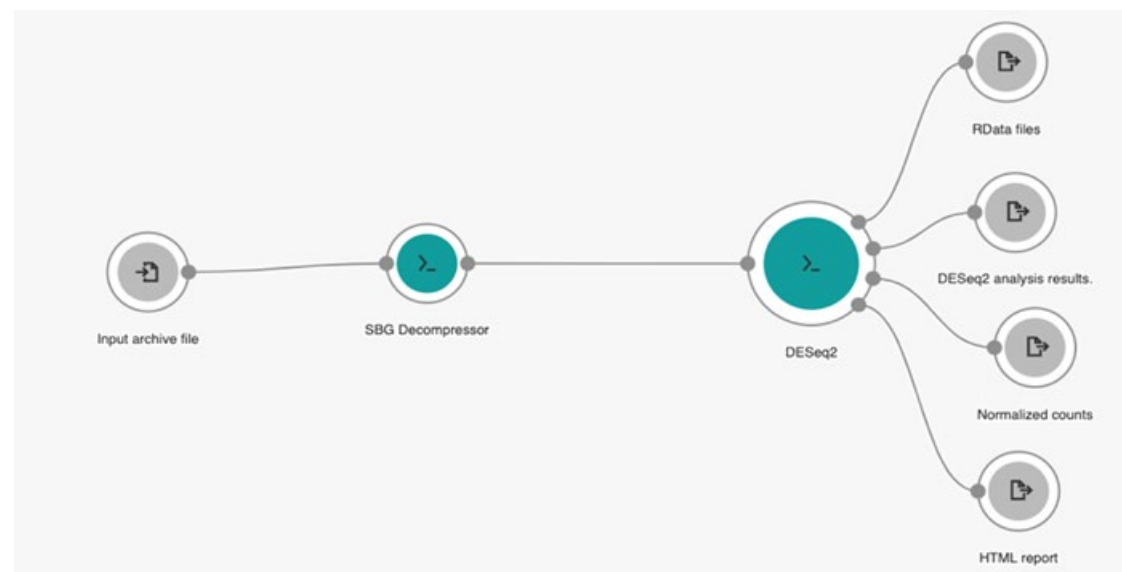
Sitting at the big table!

A mandate for interoperability with other major data resources



INCLUDE Cloud Credits Pilot Program

- [CAVATICA](#) is the INCLUDE DCC's cloud platform, where researchers run bioinformatics workflows and interactive analysis (R, Jupyter)
- INCLUDE is allocating cloud credits for use on CAVATICA:
<https://help.includedcc.org/docs/include-cloud-credits-pilot-program>
 - *Please contact Huiqing Li, huiqing.li@nih.gov*

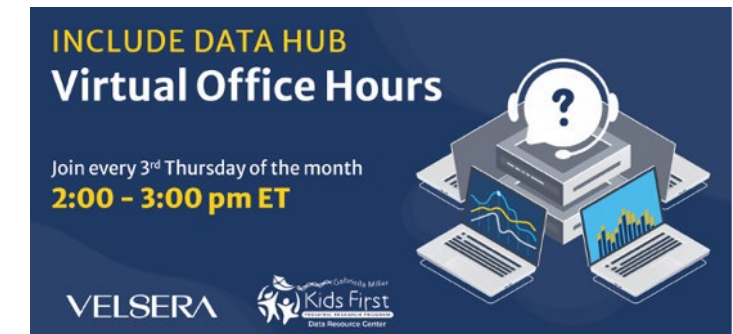


INCLUDE Cloud Credits Pilot Program, cont.

The [INCLUDE Cloud Credits Pilot Program](https://help.includedcc.org/docs/include-cloud-credits-pilot-program) allocates cloud credits for use on the INCLUDE DCC's cloud platforms to research into conditions affecting people with Down syndrome. Learn more at <https://help.includedcc.org/docs/include-cloud-credits-pilot-program>

- \$100 Pilot Credits: All INCLUDE Users
- \$1,000 Limited Credits Billing Group: with a brief description of project
- Up to \$20,000 Extended Credits Billing Group: include a detailed description of their intended use for the credits, administrative review
- Currently, there are ten cloud computing projects on CAVATICA

INCLUDE Data Hub monthly virtual office hours are a great opportunity to get your technical questions answered by our experts and learn more about utilizing the INCLUDE Data Hub including both the Data Hub and CAVATICA platform.



Funding Opportunities

1. **Small Research Grants for Analysis, Curation, and/or Sharing of Down syndrome-related Research Data for the INCLUDE Project (R03 Clinical Trial Not Allowed):**
 - Awards for up to 2 years
 - Budget up to \$100,000/year in direct costs
 - Preliminary data is not required
2. **Other INCLUDE Funding Opportunities:**
<https://www.nih.gov/include-project/funding>

What Is Important to Keep in Mind When Applying to X01



- Read and follow guidance from the program announcement!
 - Omics Phenotypes Related to Down Syndrome for the INCLUDE Project (X01 Clinical Trial Not Allowed)
- X01 gives access to generate genomic and other omics data performed at designated Omics Centers
- X01 does not fund data analysis
- Data with consent allowing broad sharing
 - Data use does not require approval by an Institutional Review Board for secondary analyses
 - Data have no publication embargo
- The program prioritizes proposals for inclusive and diverse cohorts with rich phenotypic and clinical information

Agency Contacts

Application Submission Contacts

1. eRA Service Desk
2. Finding Help Online
 - <https://www.era.nih.gov/need-help>
 - Telephone: 301-402-7469 or 866-504-9552
3. General Grants
 - Information Email: GrantsInfo@nih.gov
 - Telephone: 301-637-3015
4. Grants.gov Customer Support
 - Contact Center Telephone: 800-518-4726
 - Email: support@grants.gov

Peer Review Contact(s)

1. Barbara Thomas, PhD
NHGRI
Telephone: 301-402-8837
Email: barbara.thomas@nih.gov

Financial/Grants Management Contact(s)

1. **Deanna L Ingersoll**
NHGRI
Phone: 301-435-7858
E-mail: deanna.ingersoll@nih.gov
2. **Christopher Myers**
NIDCD
Phone: (301) 435-0713
E-mail: myersc@nih.gov
3. **Margaret Young**
NICHD
Telephone: 301-642-4552
Email: margaret.young@nih.gov
4. **Diana Rutberg, MBA**
NIDCR
Phone: (301) 594-4798
E-mail: dr258t@nih.gov

Scientific/Research Contact(s)

1. **Huiqing Li, PhD (Primary point of contact)**
NHLBI
Telephone: 301-435-0554
Email: huiqing.li@nih.gov
2. **Sujata Bardhan, MS, PhD**
NICHD
Telephone: 301-435-0471
Email: sujata.bardhan@nih.gov
3. **Kelly King, AuD, PhD**
NIDCD
Phone: 301-402-3458
E-mail: kingke@mail.nih.gov
4. **Jyoti Dayal**
NHGRI
Phone: 301.480.2307
E-mail: jyotig@nhgri.nih.gov
5. **Jason Wan, Ph.D.**
NIDCR
Phone: 301-594-9898
E-mail: jasonwan@mail.nih.gov

Appendix

Institutional Certification

Institutional Certification: Page 1

Extramural Institutional Certification*

OMB Control Number: 0925
Expiration Date: July 2019

For studies using data generated from cell lines created or clinical specimens collected after January 25, 2015

Date:

Name of GPA:

Genomic Program Administrator
 NIH, HHS
9000 Rockville Pike
Bethesda, MD 20892-7395

Re: Institutional Certification of (NAME OF INSTITUTION) to Accompany
Submission of the Dataset from (ORIGINAL STUDY NAME) for
 (PROJECT TITLE FOR DATA TO BE SUBMITTED)
to an NIH-designated data repository.

Dear
The submission of data to the NIH-designated data repository is being made with institutional approval from
 along with appropriate institutional approvals from
collaborating sites, as listed here:

(IF APPLICABLE ENTER COLLABORATING SITE NAMES HERE AND CLICK "ADD TO LIST")

LIST OF COLLABORATING SITES

The hereby assures that submission of data from the study entitled
 to an NIH-designated data repository meets
the following expectations, as defined in the [NIH Genomic Data Sharing Policy](#):

- The data submission is consistent, as appropriate, with applicable national, tribal, and state laws and regulations as well as relevant institutional policies.
- Any limitations on the research use of the data, as expressed in the informed consent documents, are delineated in the table on page 3.
- The identities of research participants will not be disclosed to NIH-designated data repositories.
- An Institutional Review Board (IRB), and/or Privacy Board, and/or equivalent body, as applicable, has reviewed the investigator's proposal for data submission and assures that:
 - The protocol for the collection of genomic and phenotypic data is consistent with [45 CFR Part 46](#);²
 - Data submission and subsequent data sharing for research purposes are consistent with the informed consent of study participants from whom the data were obtained;
 - Consideration was given to risks to individual participants and their families associated with data submitted to NIH-designated data repositories and subsequent sharing, including unrestricted access to genomic summary results;
 - To the extent relevant and possible, consideration was given to risks to groups or populations associated with submitting data to NIH-designated data repositories and subsequent sharing, including unrestricted access to genomic summary results; and
 - The investigator's plan for de-identifying datasets is consistent with the standards outlined in the [NIH Genomic Data Sharing Policy](#) (See section IV.C.1).

* Certification must be provided for all sites contributing samples. If more than one site is contributing samples, the primary site may submit one Institutional Certification indicating that they are providing certification on behalf of all collaborating sites. Alternatively, each site providing samples may provide its own Institutional Certification.

GPA: Valerie Cotton, NICHD

List all sites contributing samples

Institutional Certification: Page 2

Availability of Individual-Level Human Data

The individual-level data are to be made available through (check one)

- ☒ Controlled-access²
☐ Unrestricted access³

If **unrestricted access** is marked, the data use limitations table on the following page(s) does not need to be completed.

Is the individual-level, human genomic data to be submitted funded in whole or in part by NIH?

- ☒ YES ☐ NO

IMPORTANT: If your research involves the generation of individual-level, human genomic data and is funded in whole or in part by NIH, your research is automatically deemed to be issued a Certificate of Confidentiality (CoC). For more information, see [the NIH Certificates of Confidentiality webpage](#).

Is the individual-level, human genomic data to be submitted covered by a CoC?

- ☒ YES ☐ NO

Availability of Genomic Summary Results (GSR)

NIH provides genomic summary results⁴ (GSR) from most studies submitted to NIH-designated data repositories through unrestricted access. However, data from data sets considered to have particular “sensitivities” related to individual privacy or potential for group harm (e.g., those with populations from isolated geographic regions, or with rare or potentially stigmatizing traits) may be designated as “sensitive” by _____ and public posting would be prohibited.

In such cases, “controlled access” should be checked below and a brief explanation for the sensitive designation should be provided. GSR from any such study will only be available through controlled access and public posting would be prohibited.

- ☐ Controlled access

If “controlled access” is checked, include a brief explanation for the sensitive designation.

Individual-level sequence data are expected to be “controlled-access,” unless consent allows for unrestricted access

Keep “Controlled access” box unchecked for “unrestricted” access to **genomic summary results** unless the IRB provides a justification for designating the dataset as “sensitive”

Institutional Certification: Page 3

NIH expects the submitting institution(s) to select one of the three standard [Data Use Limitations](#) (DULs) for appropriate secondary use, or, if necessary, create a customized DUL. DULs are developed based on the original informed consent of the participant(s).

Data Use Limitations

General Research Use	GRU	Use of the data is limited only by the terms of the Data Use Certification: these data will be added to the dbGaP Collection .
Health/Medical/Biomedical	HMB	Use of the data is limited to health/medical/biomedical purposes, does not include the study of population origins or ancestry.
Disease-specific [list disease]	DS	Use of the data must be related to the specified disease.
Other		[ENTER CUSTOMIZED TEXT, IF APPLICABLE]

Additional modifiers to the standard DULs (e.g., Not-for-profit Use Only) can be indicated, if appropriate. Use of the modifiers should have a basis in the informed consent from the participants or in special knowledge of the preferences of the original study population.

Data Use Limitation Modifiers (Optional)

IRB Approval Required	IRB	Requestor must provide documentation of local IRB approval.
Publication Required	PUB	Requestor agrees to make results of studies using the data available to the larger scientific community.
Collaboration Required	COL	Requestor must provide a letter of collaboration with the primary study investigator(s).
Not-for-profit Use Only	NPU	Use of the data is limited to not-for-profit organizations.
Methods	MDS	Use of the data includes methods development research (e.g., development and testing of software or algorithms).
Genetic Studies Only	GSO	Use of the data is limited to genetic studies only.

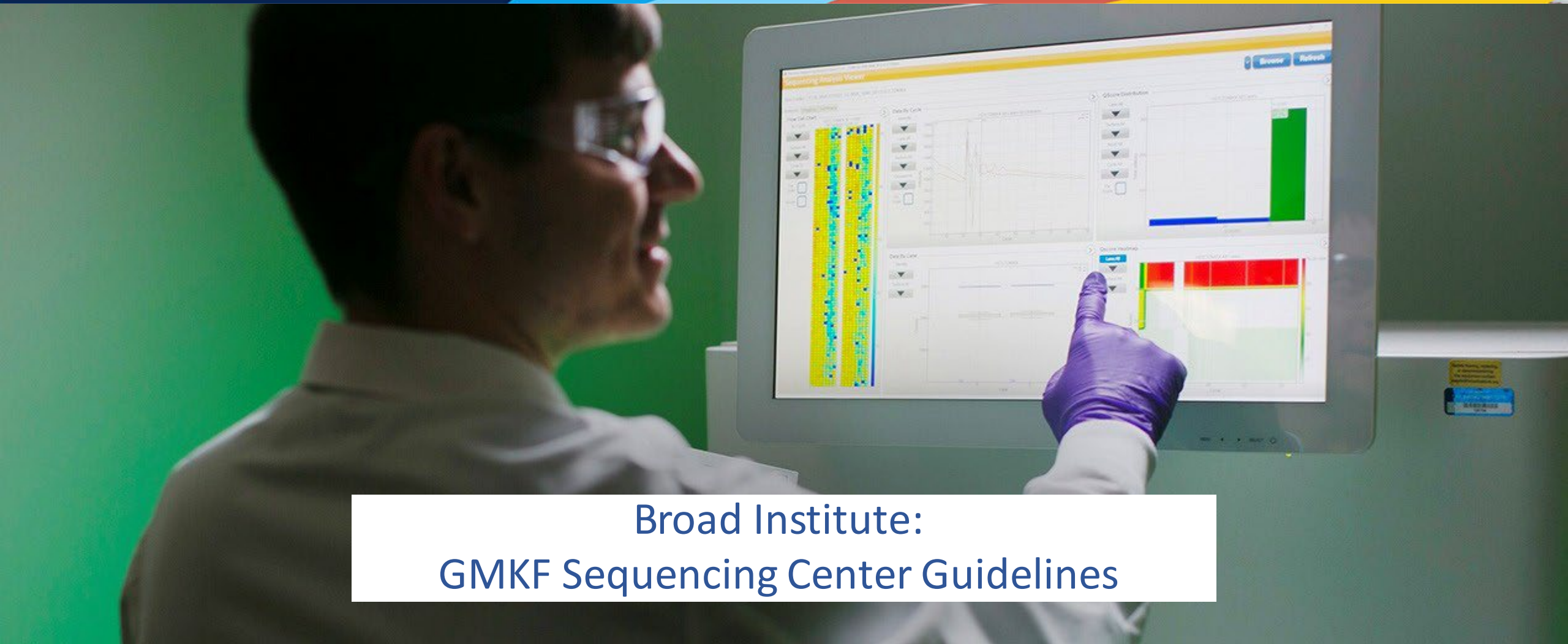
Using the tables above, please indicate in the table below the consent group(s) for each collaborating study site. Use one row per consent group.

Collaborating Site Name	Data Use Limitation	Data Use Limitation Modifiers (optional)
Eg: Cold Cohort Study	Health/Medical/Biomedical	IRB ☐ PUB ☐ COL ☐ NPU ☐ MDS ☐ GSO ☐
Eg: Cold Cohort Study	Disease Specific Research [Lung Cancer]	IRB ☐ PUB ☐ COL ☐ NPU ☒ MDS ☐ GSO ☐
	General Research Use	IRB ☐ PUB ☐ COL ☐ NPU ☐ MDS ☐ GSO ☐

**“General Research Use”
with no modifiers is
expected for individual-
level data, unless specific
uses are clearly
prohibited**

Appendix 2

Kids First Sequencing Center



Broad Institute: GMKF Sequencing Center Guidelines

Broad Team



Broad GMKF X01 Team	Title	Contact
Stacey Gabriel	PI	stacey@broadinstitute.org
Namrata Gupta	Sr. Director / Support for Stacey Gabriel	namrata@broadinstitute.org
Katie Larsson	Sr. Project Manager	katiel@broadinstitute.org
Sheli McDonough	Project Manager II	sheli@broadinstitute.org

Broad Genomics Platform - Our Model

Build on our history



Partner with Broad Investigators



Nick Wagle

*Cancer Genomics
Direct to Patient*



Heidi Rehm

*Clinical Genomes &
Data Aggregation
Rare Disease*



Steve McCarroll

*Single-Cell
Genomics*



Ramnik Xavier

Microbiome

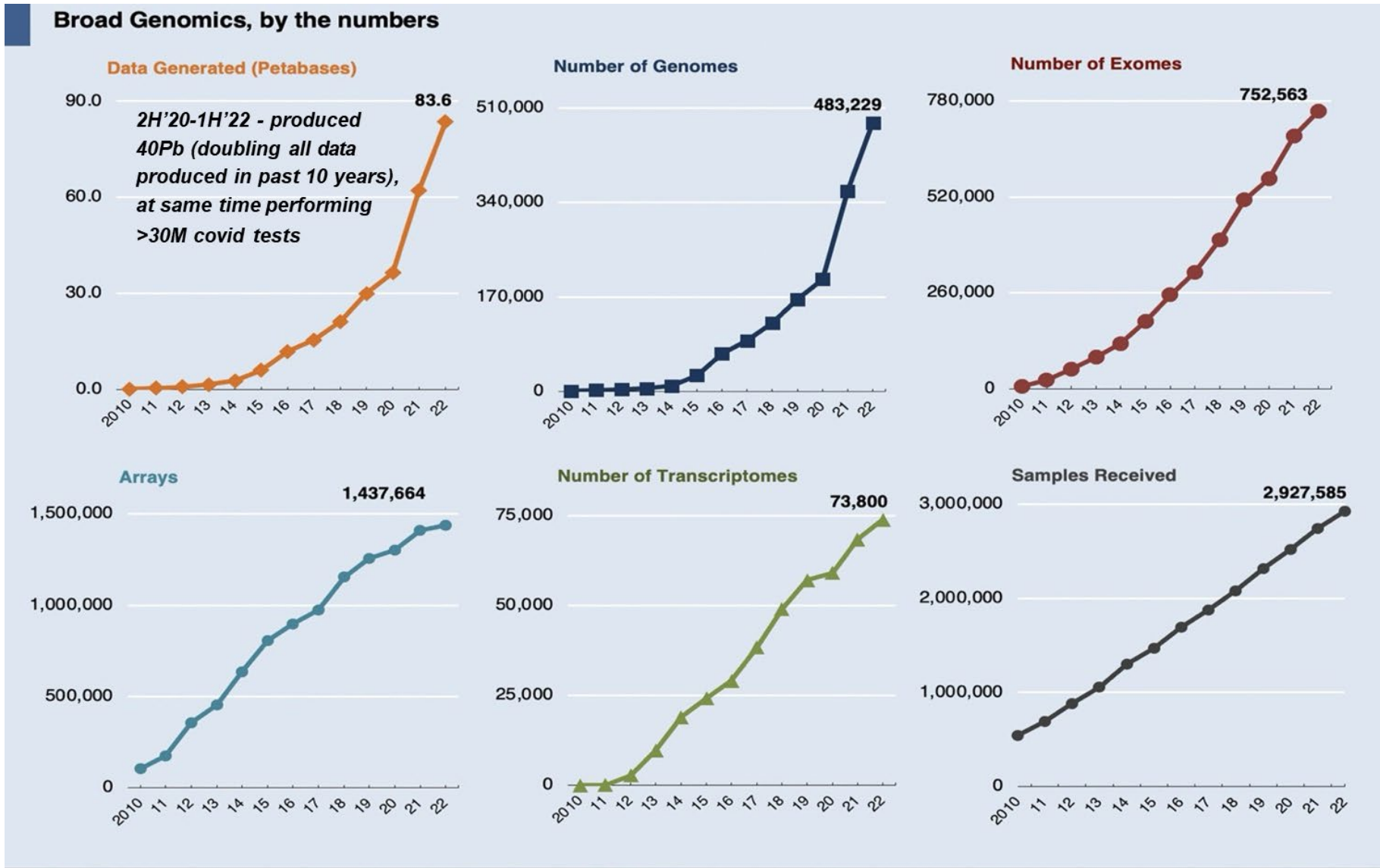
Create Resources for the Community



Enable Service Offerings



Broad Genomics, by the Numbers



Sequencers at the Genomics Platform

Illumina NovaSeq 6000™: **27**

Illumina NovaSeq X™: **1***

Illumina HiSeq X™: **5** Illumina

HiSeq 2500™: **7** Illumina

MiSeq™: **4**

Illumina NextSeq 500™: **3**

Illumina NextSeq 2000™: **2**

Illumina iSeq™: **1**

PacBio Sequel IIe™: **25**

PacBio Revio™: **1**

ONT PromethION™: **1**

ONT GridION™: **1**

Ultima UG 100: **2**

Element Biosciences AVITI™: **3**

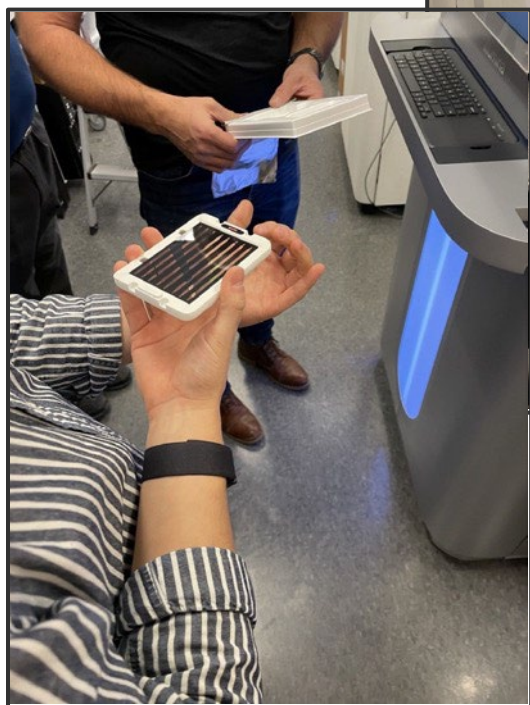


**4 more NVX expected by March 3rd, 2023*

Illumina Nova Seq Xtm (NV X) and PacBio Reviotm



Illumina Nova Seq Xtm (NV X)

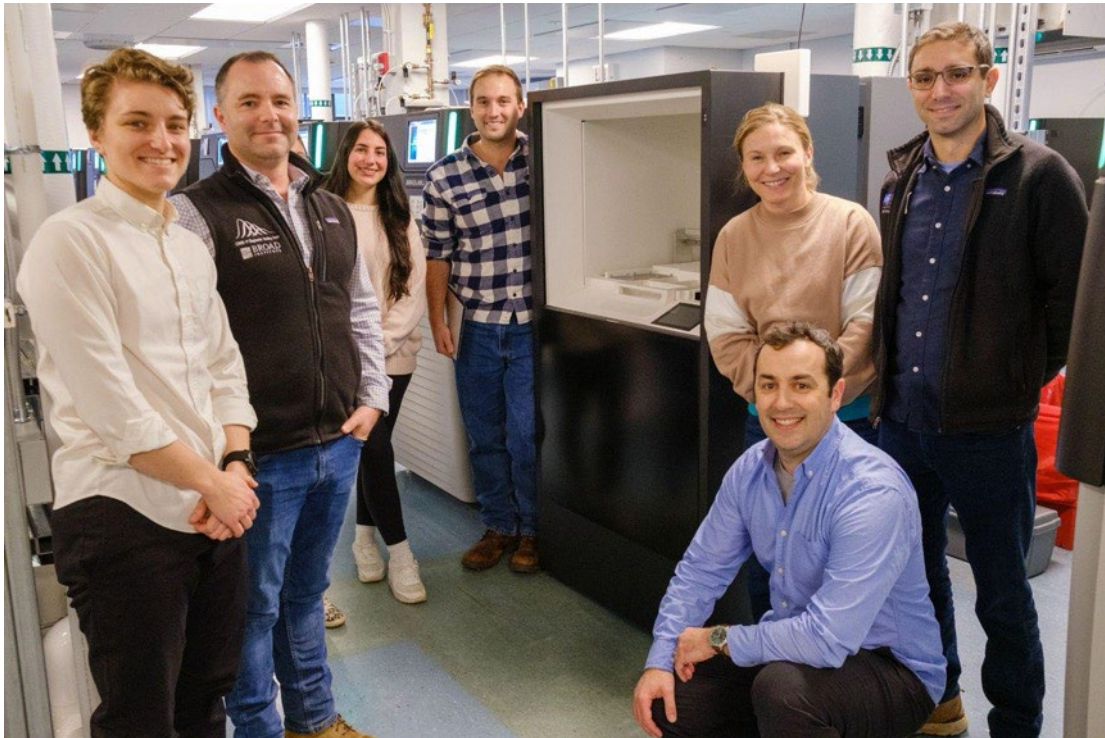


- First machine arrived Feb 2023.
- Current flowcell iteration ('10B') is very similar to NovaSeq 6000 S4 in terms of output (3,000 Gb), but twice as fast.
- Future flowcell ('25B') is 25% improvement over 10B due to 2.5x output (8,000 Gb). Tentatively available Q3 2023.
- Long-term plan includes all analysis up to final file type (CRAM, gvcf, etc.) to be done by onboard Dragen.
- First products to move will be high impact (WalkUpSeq) and high scale (BGE).

Increased Throughput Compared to NovaSeq 6000

Sequencer	Kit	Lanes	Reads per FC	Gb per FC (300c)	Gb per Lane (300c)	Run Length (Days)	Notes
NovaSeq 6000	S4	4	10 Billion	3,000	750	2	Currently Available
NovaSeq X	10B	8	10 Billion	3,000	375	1	Newly validated
NovaSeq X	1.5B	2	1.5 Billion	500	250	1	Available Q3 2023
NovaSeq X	25B	8	25 Billion	8,000	1,000	2	Available Q3 2023

Pac Bio Revio™



- First machine arrived February 2023.
- 360Gb per machine run in 24 hours (4 cells) verses 120Gb per machine run in 138 hours (4 cells).
- 15x increases in HiFi read throughput vs Sequel II due to higher density cells and increased stages within the instrument.
- Updated SMRT Link Software (v11.0 to 12.0)
- Same fundamental output as previous instrument.
- Downstream analysis workflows remain the same.

Sample Plating Requirements

Whole Genome Sequencing

(WGS)* Concentration:

Minimum 15 ng/uL Volume:

Minimum 50uL

Total ng DNA: Minimum 750 ng

RNA Sequencing*

Concentration: Between 7ng/uL -

60ng/uL Volume: Minimum 50uL

Total ng RNA: Minimum

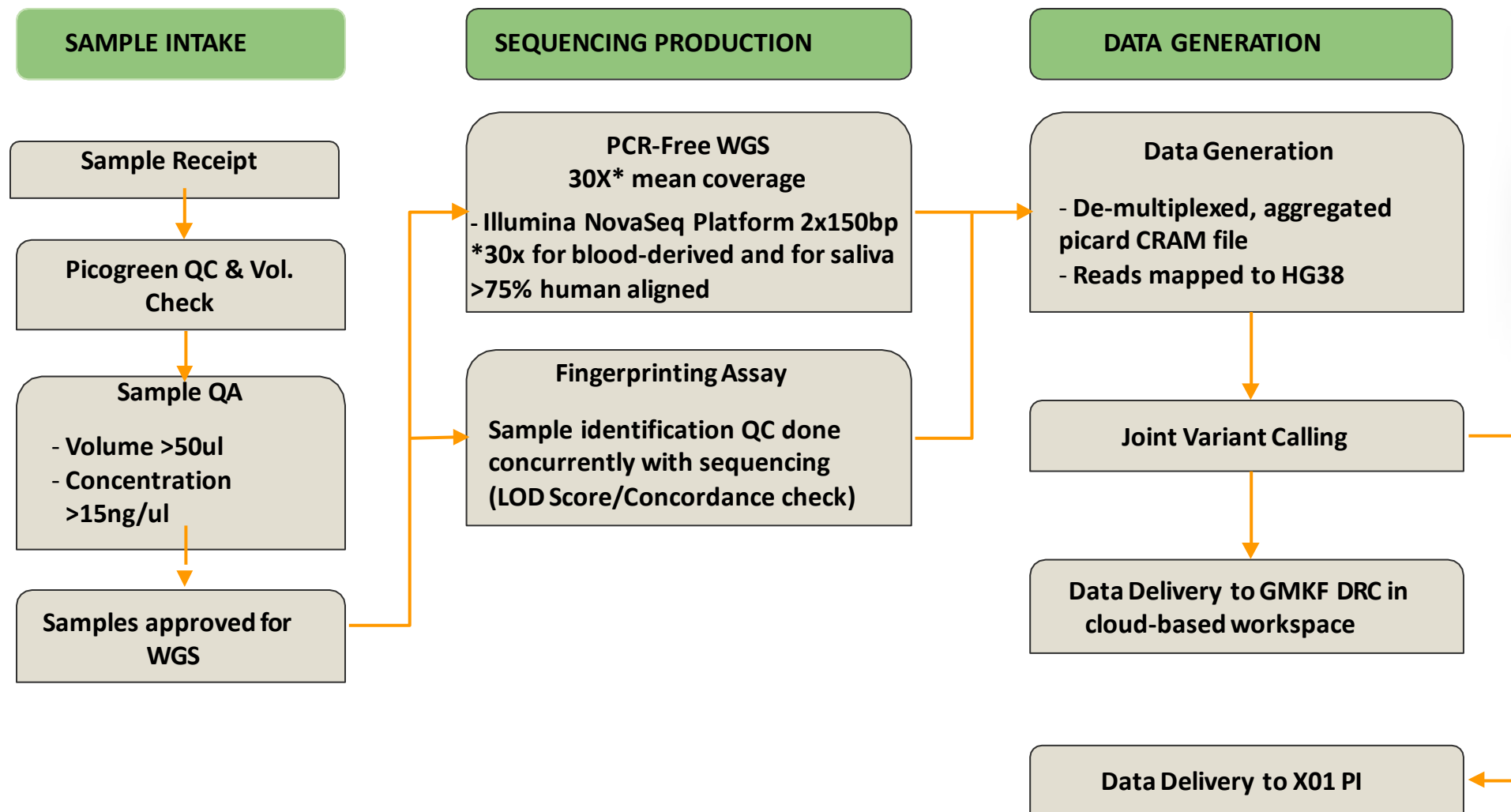
350ng RQS (RNA QC

Score): >5.5

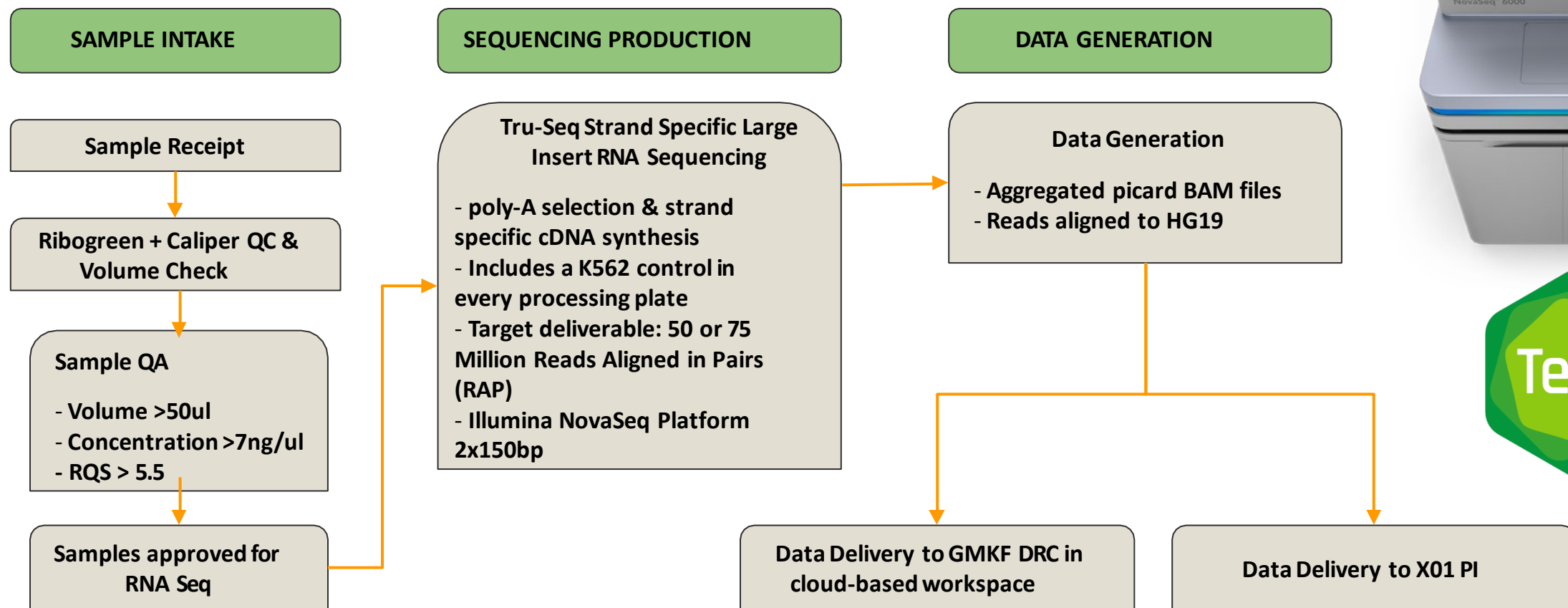


- ThermoFisher 2D Barcoded Matrix Racks (95 samples*)
- Upon sample receipt - barcode on incoming stock tube is scanned and this ID is linked to the sample in our LIMS (Laboratory information management system)
- All workflows are fully automated with barcoded LIMS messaging throughout all E2E process

Sequencing and Data Delivery - 30 x WGS



Sequencing and Data Delivery - RNAS eq



Sequencing and Data Delivery - Long Read Seq

