


Webinar Instructions

*Welcome to the Gabriella Miller Kids First
RFA-RM-21-011 Pre-Application Webinar!*

- Every participant is muted upon entry.
- To ask public questions, use the **Q&A** bar (right side of your screen). We encourage you to save these for the question periods.
- You can also use the “chat”  service to send private messages to the host or presenters throughout the webinar.
- After the webinar, additional questions can be emailed to:
valerie.cotton@nih.gov

***This webinar will be recorded.
We will start at 12pm (EDT)***



Gabriella Miller Kids First Pediatric Research Program
Expert-Driven Small Projects to Strengthen
Gabriella Miller Kids First Discovery, RFA-RM-21-011

March 29, 2021

12:00 pm EDT



The Common
Fund



Public Webinar: April 13th!

Register here:

<https://nih.webex.com/nih/onstage/g.php?MTID=e7dcd35de768586aa8c80352b1f544601>

4:00PM **Introduction; NIH Kids First Staff**

4:05PM ***Detection of novel genetic bases for congenital cranial dysinnervation disorders (CCDDs) by whole genome sequencing:***

Elizabeth Engle, MD

4:35PM: **Kids First Data Resource Center**

- **New Portal Updates**

- **Getting Started with Kids First**

5:05PM: **Proteogenomic Analysis: *Breakthroughs for pediatric cancer through cross-platform collaboration***; Adam Resnick, PhD, Pei Wang, PhD, Francesca Petralia, PhD

- **5:25PM: Questions & Answers**

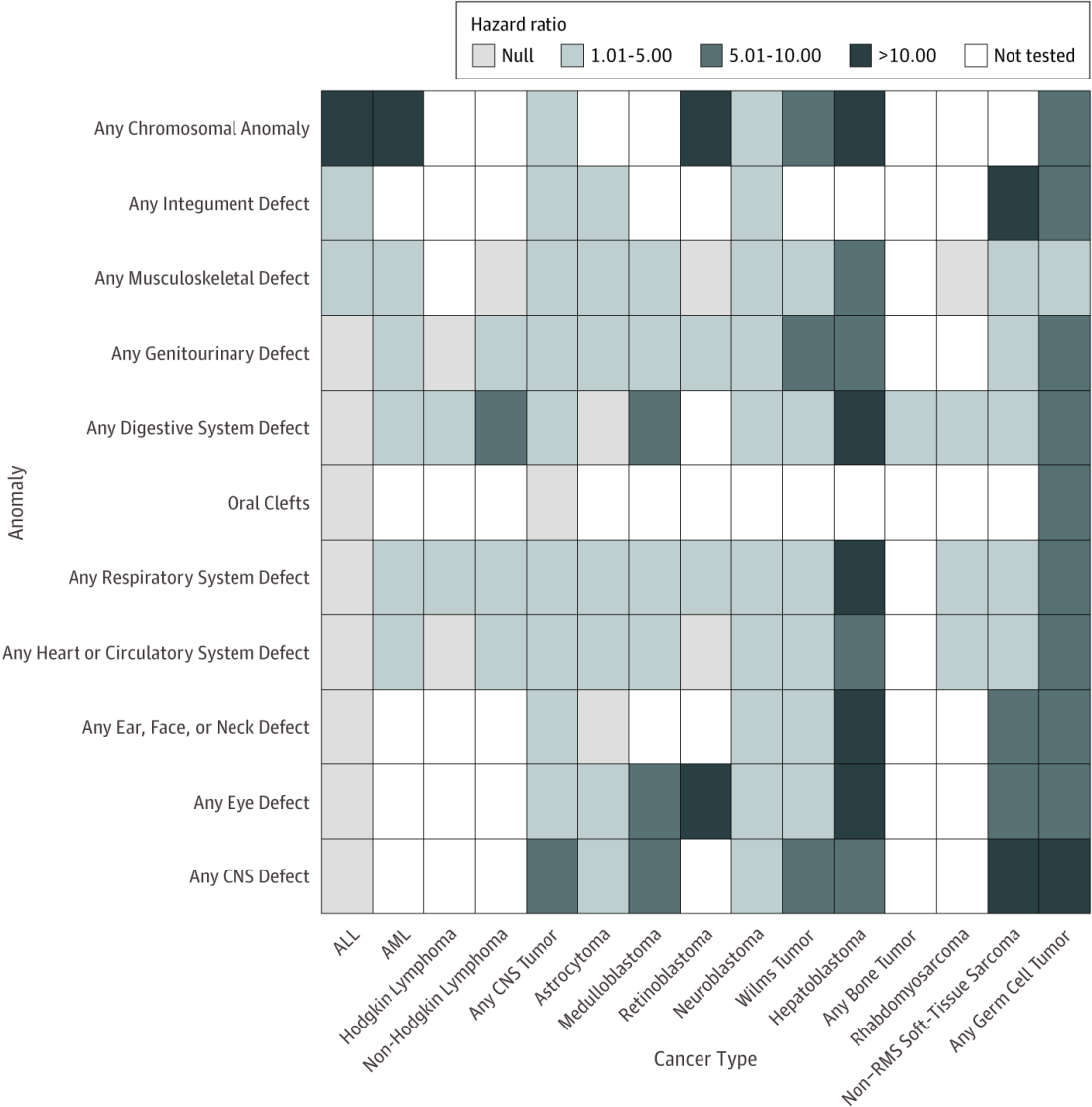
5:35PM: **NIH Program Updates**

5:50PM: **Questions & Answers**



Gabriella Miller Kids First Pediatric Research Program: *childhood cancer & structural birth defects*

Birth defects are associated with increased risk of cancer among children... suggesting shared genetic pathways



From: **Association Between Birth Defects and Cancer Risk Among Children and Adolescents in a Population-Based Assessment of 10 Million Live Births**

Lupo et al, JAMA Oncol. 2019;5(8):1150-1158. doi:10.1001/jamaoncol.2019.1215

Vision



Alleviate suffering from childhood cancer and structural birth defects by fostering **collaborative research** to uncover the etiology of these diseases and supporting **data sharing** within the pediatric research community.



Kids First: Phase 1



Kids First Major Initiatives

1. Identify & sequence cohorts of children with **childhood cancer and/or structural birth defects**.
2. Build the **Gabriella Miller Kids First Data Resource** to empower discovery



Kids First Sequencing Cohorts 2015-2020

40 projects | 40,000 genomes | 16,000 cases | 19 released datasets



BROAD
INSTITUTE



HUDSONALPHA
INSTITUTE FOR BIOTECHNOLOGY



- *Disorders of Sex Development*
- *Congenital Diaphragmatic Hernia*
- *Ewing Sarcoma*
- *Structural Heart & Other Defects*
- *Syndromic Cranial Dysinnervation Disorders*
- *Cancer Susceptibility*
- *Adolescent Idiopathic Scoliosis*
- *Neuroblastomas*
- *Enchondromatoses*
- *Orofacial Clefts in Caucasian, Latin American, Asian & African, Filipino populations*
- *Osteosarcoma*
- *Familial Leukemia*
- *Craniofacial Microsomia*
- *Intersection of childhood cancer & birth defects*
- *Microtia*
- *Esophageal Atresia and Tracheoesophageal Fistulas*
- *Kidney and Urinary Tract Defects*
- Nonsyndromic Craniosynostosis
- Hemangiomas, Vascular Anomalies & Overgrowth
- Bladder Exstrophy
- Hearing Loss
- Cornelia de Lange Syndrome
- Intracranial & Extracranial Germ Cell Tumors
- Fetal Alcohol Spectrum Disorders
- Myeloid Malignancies + overlap with Down syndrome
- Congenital Heart Defects & Acute Lymphoblastic Leukemia in Children with Down Syndrome
- Structural Brain Defects
- Structural Defects of the Neural Tube (Spina Bifida: Myelomeningocele)
- CHARGE Syndrome
- Laterality Birth Defects
- T-cell Acute Lymphoblastic Leukemia
- Pediatric Rhabdomyosarcoma
- Valvar Pulmonary Stenosis



The Kids First Data Resource for Collaborative Discovery

Data Resource Portal

Entry point. Query, search, discover, build & visualize synthetic cohorts

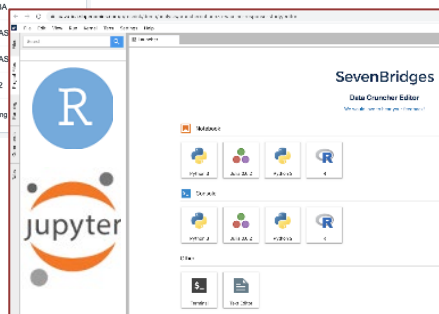


Cavatica

Pull data from multiple sources into one workspace. Use notebooks, bring-your-own or use available workflows.

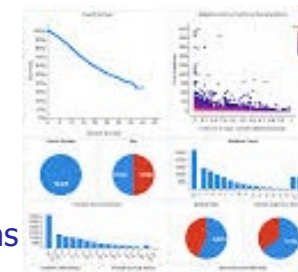
The screenshot shows the Cavatica 'Integrated Kids First/TARGET Analysis' workspace. It features a table with columns for 'Name', 'Case ID', and 'Sample ID'. The table lists various data entries, including sample IDs like '00e47068-3d3c-4b3d-ba10-8371a1d33c3c.bam' and '01102766-626f-4396-850f-3c70d89a8dbb.cram', and associated case and sample IDs. A search bar and filters are visible at the top of the table.

Name	Case ID	Sample ID
TARGET 00e47068-3d3c-4b3d-ba10-8371a1d33c3c.bam	TARGET-30-PAPVEB	TARGET-30-PAPVEB-04A
KIDS-FIRST 01102766-626f-4396-850f-3c70d89a8dbb.cram	PT_T4BK7XD1	BS_7TWWV1Y9
KIDS-FIRST 01859ccc-0993-493b-8bae-08751b04e4f6.cram	PT_J6K6ETOG	BS_DBF88SM2
KIDS-FIRST 018edf5c-1a7e-475b-a451-9f3b5bd11c3.cram	PT_PSYYSZ4	BS_CCGFJW3A
TARGET 01c87d09-4a5d-4cdd-844d-749394dada96.bam	TARGET-30-PASWJ	TARGET-30-PASWJ
TARGET 02a86e96-346e-4f8a-bada-23b182341b6b.bam	TARGET-30-PASWYR	TARGET-30-PASWYR
KIDS-FIRST 02d2d6f8-a5db-4c7e-a196-58d302d14d95.cram	PT_8U43FRQ	BS_8VJNFAF2



Knowledge Base Integrations (PedcBioPortal)

Integrations with existing curated/published data visualizations



Data Services

Model clinical data in FHIR-based data services and API for sharing layers of harmonized data



aws
STRIDES

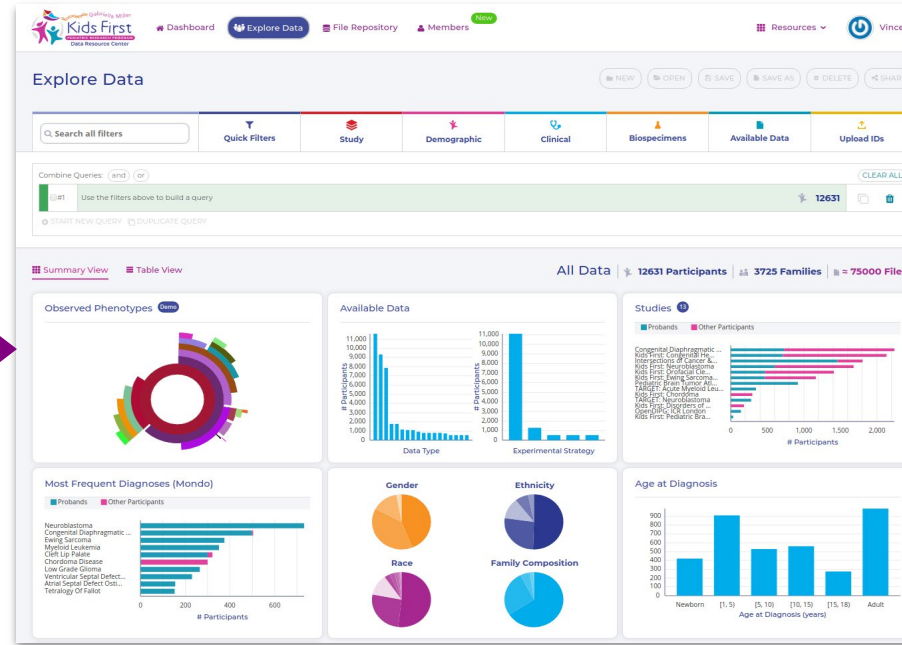
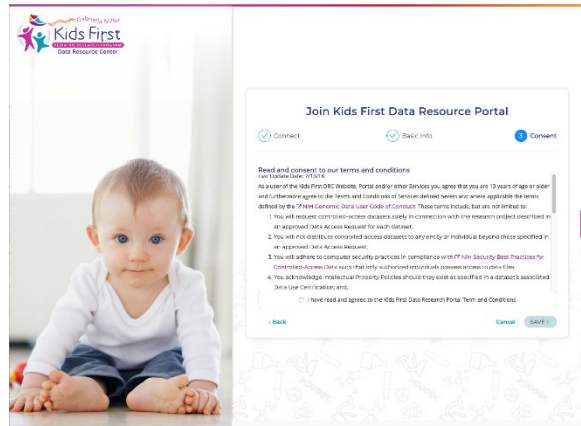
GEN3
DATA COMMONS

Framework Services

Index and point to files in the cloud (for approved users)

Use Case: Compare genetic variants of congenital heart defects & neuroblastoma

Anyone can [register & login](#) to the portal (via ORCID, Google). User agrees to [terms](#)



In *Explore Data*, user searches the terms “[heart](#)” and “[neuroblastoma](#)”. Discovers data from children with congenital [heart](#) disease (KF) & [neuroblastoma](#) (KF & NCI TARGET)



User builds a synthetic cohort based on these criteria and can view summary & deidentified individual-level clinical, demographic, and phenotypic information.

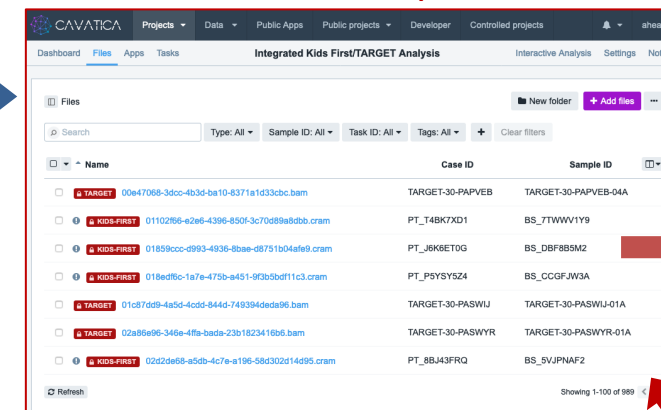
Synthetic cohort is ported to the *File Repository* where user selects which **genomic** and **histology image** files they want to analyze.

File ID	Participants ID	Study Name	Proband	Family ID	Data Type	File Format	File Size
GF_H0230GIP	PT_0240P7	Congenital Diaphra...	No	RM_0085M8	Aligned Reads	cram	15.53 GB
GF_H030CTIV	PT_0315M8P	Congenital Diaphra...	No	RM_0408M5S	gVCF	gVCF	4.13 GB
GF_H040P22	PT_2P18ZVW	Congenital Diaphra...	No	RM_0204V4P	gVCF	gVCF	5.94 GB
GF_H040A2C5	PT_0408M4	Congenital Diaphra...	No	RM_0810G0F	gVCF	gVCF	4.91 GB
GF_H040Q21	PT_0404M7	Congenital Diaphra...	Yes	RM_0404M7	Aligned Reads	bam	69.23 GB
GF_H040Q21	PT_0404M7	Congenital Diaphra...	Yes	RM_0404M7	gVCF	gVCF	5.37 GB
GF_H040Q21	PT_0404M7	Congenital Diaphra...	Yes	RM_0404M7	Aligned Reads	cram	16.87 GB
GF_H040Q21	PT_0404M7	Congenital Diaphra...	Yes	RM_0404M7	Aligned Reads	bam	15.74 GB
GF_H040Q21	PT_0404M7	Congenital Diaphra...	No	RM_0404M7	Aligned Reads	cram	20.77 GB
GF_H040Q21	PT_0404M7	Congenital Diaphra...	Yes	RM_0404M7	Aligned Reads	bam	62.31 GB
GF_H040Q21	PT_0404M7	Congenital Diaphra...	No	RM_0404M7	Aligned Reads	cram	20.62 GB
GF_H040Q21	PT_0404M7	Congenital Diaphra...	No	RM_0404M7	Aligned Reads	bam	64.63 GB
GF_H040Q21	PT_0404M7	Congenital Diaphra...	Yes	RM_0404M7	Aligned Reads	cram	20.26 GB

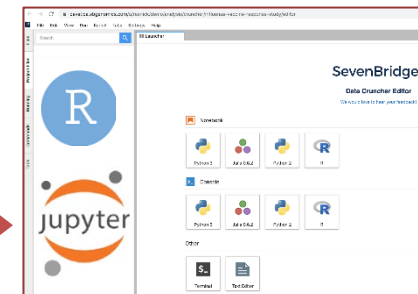
User has or applies for dbGaP access for genomic data



User pushes genomic, clinical, and image data into Cavatica for analysis & visualization



User runs statistical analyses in notebooks



User iterates through genomic workflows

Kids First: Phase 2



Phase 2 Strategic Planning: 7 Consensus Recommendation Themes

1. Innovation: Resource, infrastructure, or tool development.

Activities: Data Visualization tools; other tools for clinical/phenotypic data



2. Clinical/phenotypic data extraction, harmonization, & curation.

Activities: Collect, extract, organize, curate, harmonize, and submit deep clinical and phenotypic data; annotate variants with pathogenicity, ClinGen scores.



3. Collaborative validation and discovery.

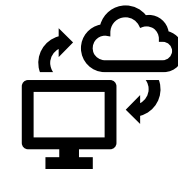
Activities: Building synthetic cohorts; identify structural variants; test pipelines.

Engage trainees in data analysis projectsBring users to the platform**



4. Integration and interoperability of external pediatric datasets.

Activities: Using DRC workflow and best practices to harmonize external pediatric datasets; Building tools that can operate across multiple spaces



5. Consent and data sharing.

Activities: Re-consenting cohorts in line with our data sharing expectations



6. Validation with model organisms.

Activities: validating KF findings/variants, deep phenotyping of animal models



7. Continue WGS & data generation, invest in long-read, consider other –

omics. Reissues of: <https://grants.nih.gov/grants/guide/pa-files/PAR-19-104.html>



NIH Council of Councils

September 11, 2020

Common Fund Concept Clearance: Gabriella Miller Kids First Pediatric Research Program: Plans for FY22-24 (Phase 2)

James Coulombe, Ph.D.

Chief, Developmental Biology and Structural Variation Branch

Eunice Kennedy Shriver National Institute of Child Health and Human Development

Goal of Phase 2 Initiatives: Enhance the value and impact of the Kids First Data Resource to accelerate pediatric research to improve preventative measures, diagnostics, and therapeutic interventions.

The archived videocast of the Council of Councils meeting is publicly available and can be viewed [here](#) (Kids First discussion begins at 4:48:00).

The presentation materials are available [here](#).



Phase 2 Initiatives Approved

\$12.6M/year (FY22-24)

1) Additional generation of childhood cancer and structural birth defects-related -omics data

- Add epigenomic and proteomic assays



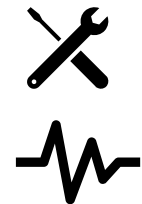
2) Continue development & improvement of the Data Resource

- Plan for sustaining the Data Resource beyond FY24



3) Expert-driven activities to increase the value of Kids First data

- Engage Kids First & community experts in activities such as integration, curation, and/or harmonization of rich clinical and phenotypic data



RFA-RM-21-011

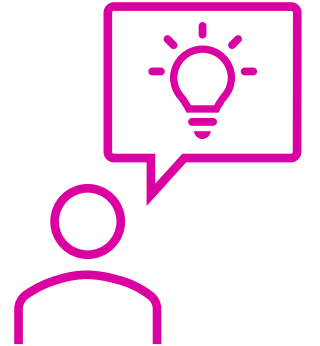
**Expert-Driven Small Projects to
Strengthen Gabriella Miller Kids First
Discovery (R03 Clinical Trial Not Allowed)**



Purpose

Engage **experts** in a variety of activities that will **enhance the utility of childhood cancer and/or structural birth defects datasets generated by the Kids First program and associated resources.**

*These activities should strengthen future analyses with the **ultimate goal of improving diagnostic capabilities and therapies for children and their families affected by these conditions.***



Goals

Build on and improve Kids First:

- These projects must **use Kids First data, standards, infrastructure, and resources**, as much as possible ...

Address challenges or gaps

Collaborate & coordinate:

- Incorporate a diversity of complementary expertise
- Coordinate with X01s, DRC, etc, to build on, but **not** duplicate, existing efforts
- Create a profile in the portal

Share data & resources



Collaborate & Coordinate

Kids First X01 investigators:

- <https://kidsfirstdrc.org/partners/partners-investigators/>
- <https://commonfund.nih.gov/kidsfirst/x01projects>

Kids First DRC members:

- <https://kidsfirstdrc.org/about/>

Kids First Sequencing Centers:

- <https://kidsfirstdrc.org/partners/portal-sequence-data/>



Sharon Plon

Baylor College of Medicine

Research Focus: Identifying novel cancer susceptibility mutations

[VIEW PROFILE >](#)



Jonathan Rios

UT Southwestern Medical Center

Research Focus: Genomics of orthopaedic disease program

[VIEW PROFILE >](#)



Jun Shen

Brigham and Women's Hospital

Research Focus: Hearing loss

[VIEW PROFILE >](#)



Azeez Butali

University of Iowa

Research Focus: Craniofacial genetics

[VIEW PROFILE >](#)

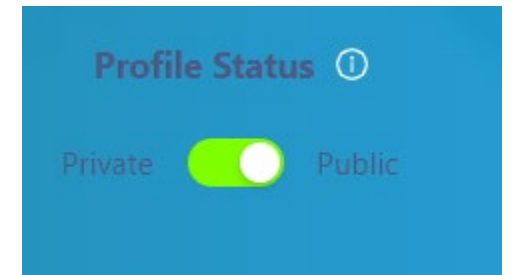
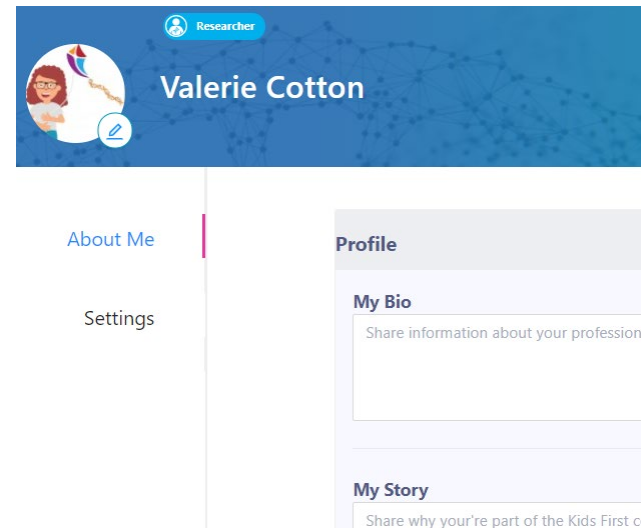
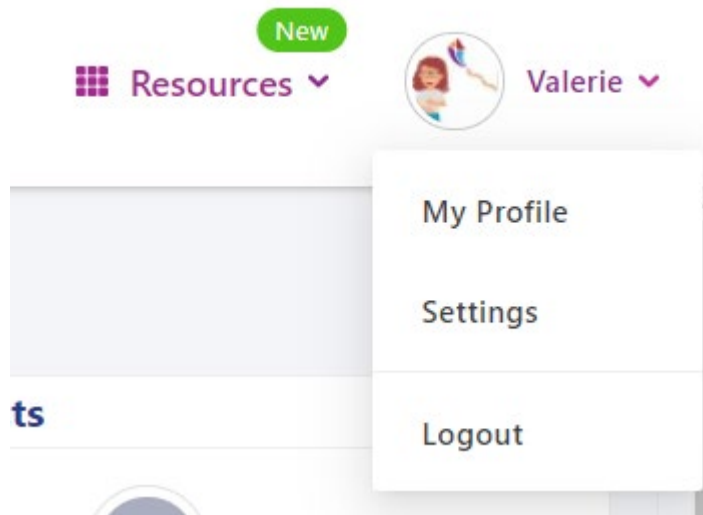


Collaborate & Coordinate

Create & Share a Portal Profile

New to the Kids First Data Resource Portal? [JOIN NOW >](#)

[PORTAL LOGIN](#)



Search other members

<https://portal.kidsfirstdrc.org/memberPage>

Data & Resource Sharing Expectations

- *It is expected that any data (including resultant raw, derived, aggregated, and summary data), tools, workflows, and/or pipelines created or used with support from this FOA will be provided to the Kids First Data Resource Center **to be shared with the wider scientific community, if not already part of the Data Resource, in a timely manner that would enable other researchers to use and build on for future research efforts.***
 - Example: If you create a new survey instrument to collect deeper data, or a tool to extract data from EHRs, the questions, code, and other documentation must be shared.
 - Example data sharing plans:
<https://commonfund.nih.gov/kidsfirst/FAQ>



Examples: Improve Discovery by...

1. Collecting, extracting, submitting deeper data or new data types associated with Kids First datasets
2. Harmonizing or processing data to promote cross-disease or cross-species (or cross-dataset analysis)
3. Portal analysis workflows to deploy withing the Kids First Data Resource (e.g., CAVATICA)
4. Creating or integrating, a new or separate tool to federate with the Kids First Data Resource
5. Consenting for broader data sharing



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FHIR for Electronic Health Record Extractions

HL7 [FHIR](#)® (Fast Healthcare Interoperability Resources)

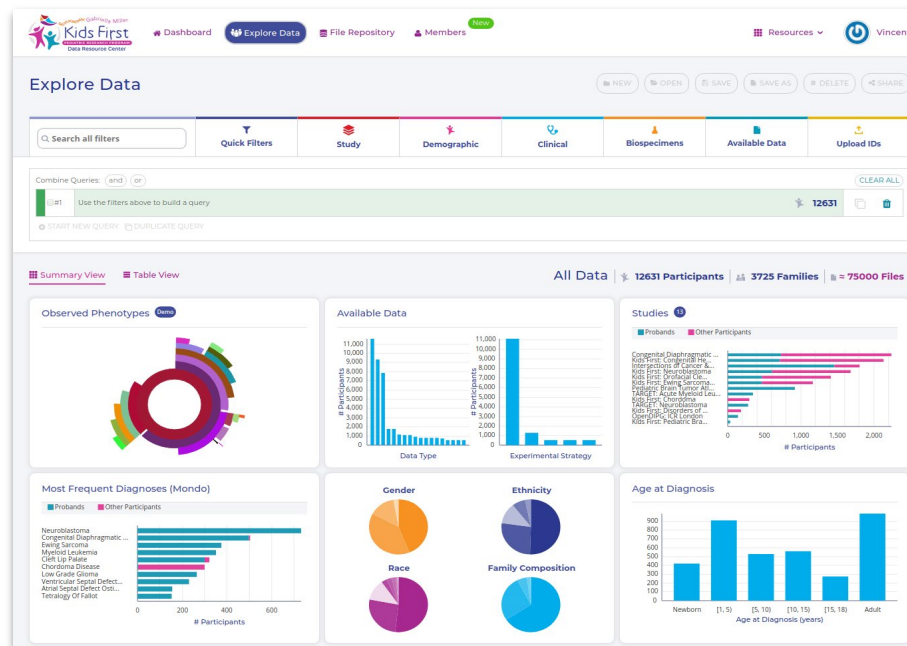
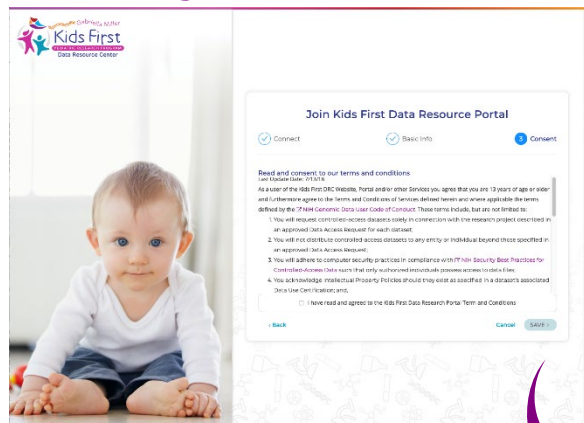
Why FHIR?

- FHIR is the bridge from hospital-based data to research
- Enough **structure** for computational approaches/tools
 - Immediate structure for EHR-based data
 - Transformations to existing data models (e.g., OMOP)
 - Facilitate ingest >> populate into tools & search portals
 - Facilitate analysis across multiple/different datasets
- But enough **flexibility** to capture rich/complex data elements that may get lost in datasets “harmonized” to Common Data Models
 - Ability to expand data models beyond EHR and/or Common Data Models (e.g., research surveys, case report forms)



Got Images?

Anyone can register & login to the portal (via ORCHID, Google). User agrees to [terms](#)

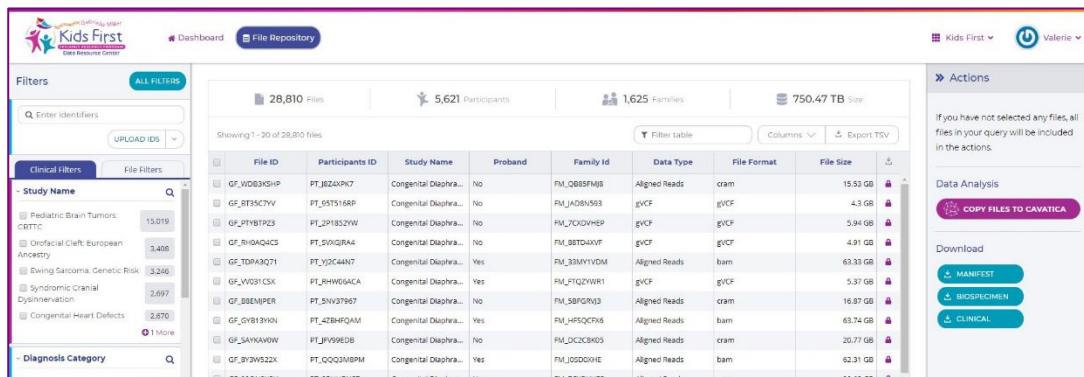


In *Explore Data*, user searches the term “kidney” and discovers data from children with congenital kidney defects (KF data) & COVID-related kidney complications (MIS-C).



User builds a synthetic cohort based on these criteria and can view summary & deidentified individual-level clinical, demographic, and phenotypic information.

Synthetic cohort is ported to the *File Repository* where user selects which **imaging** files they want to analyze (e.g. DICOM).

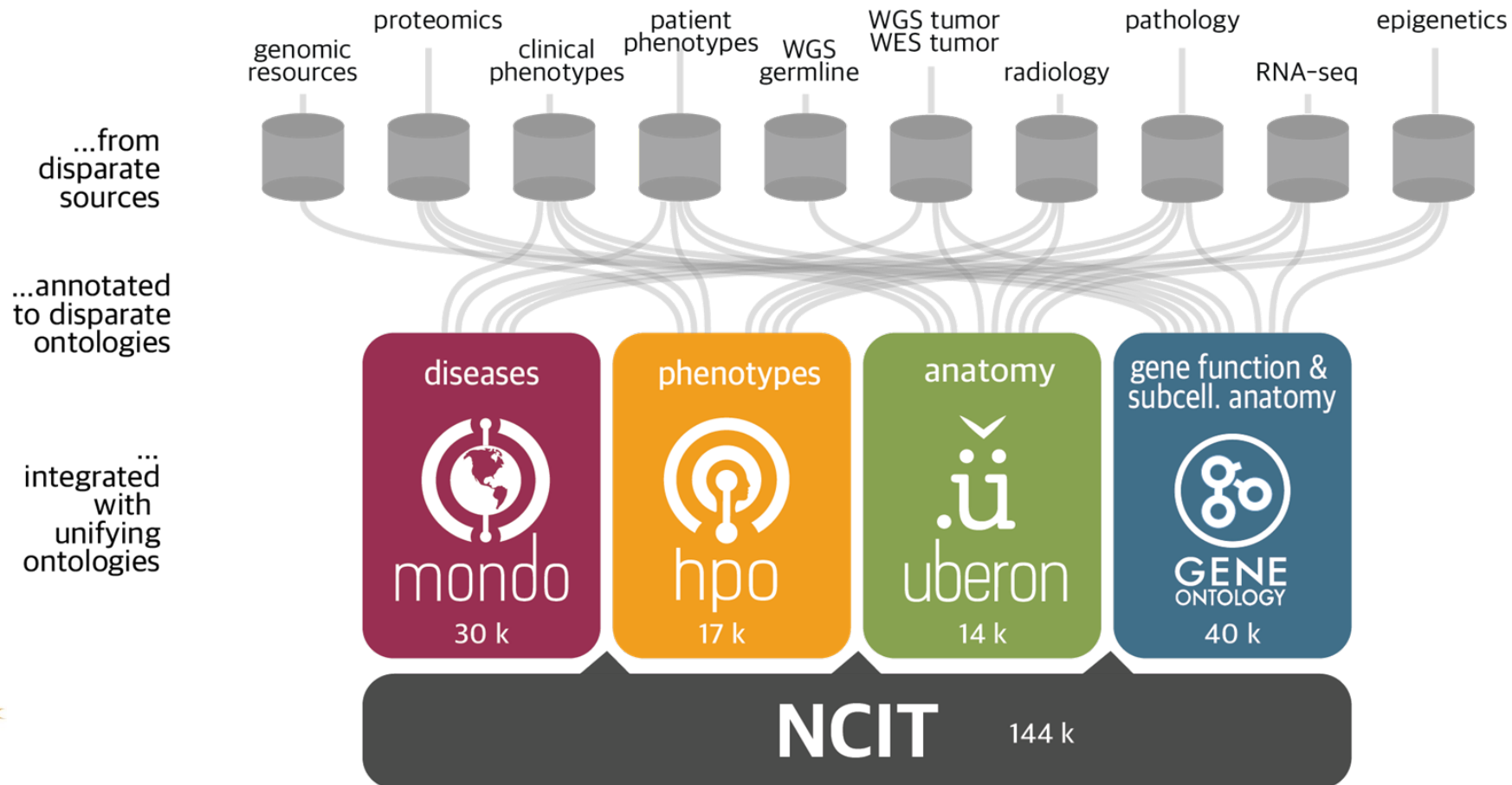


User pushes imaging files into FlyWheel for analysis

Innovation across the Phenotypic Translational Divide Webinar Series

Information: <https://monarch-initiative.github.io/phenomics/pages/clin-phen-webinar.html>

Curation with ontologies that support heterogenous data types in Kids First






Innovation across the Phenotypic Translational Divide

Webinar Series

Part 1

Pediatric Cardiac Genomics Consortium (PCGC) - Betsy Goldmuntz	Down Syndrome -Congenital Heart Disease - Joaquin Espinosa	Neuropsychological Data Harmonization - Stephanie Sherman
Down Syndrome-ALL & Rhabdomyosarcoma - Phillip Lupo		Enchondromatosis and Related Malignant Tumors - Nara Sobreira
Orofacial Clefts - Mary Marazita	Adolescent Idiopathic Scoliosis - Carole Wise	Cornelia de Lange Syndrome - Sarah Raible

Part 2

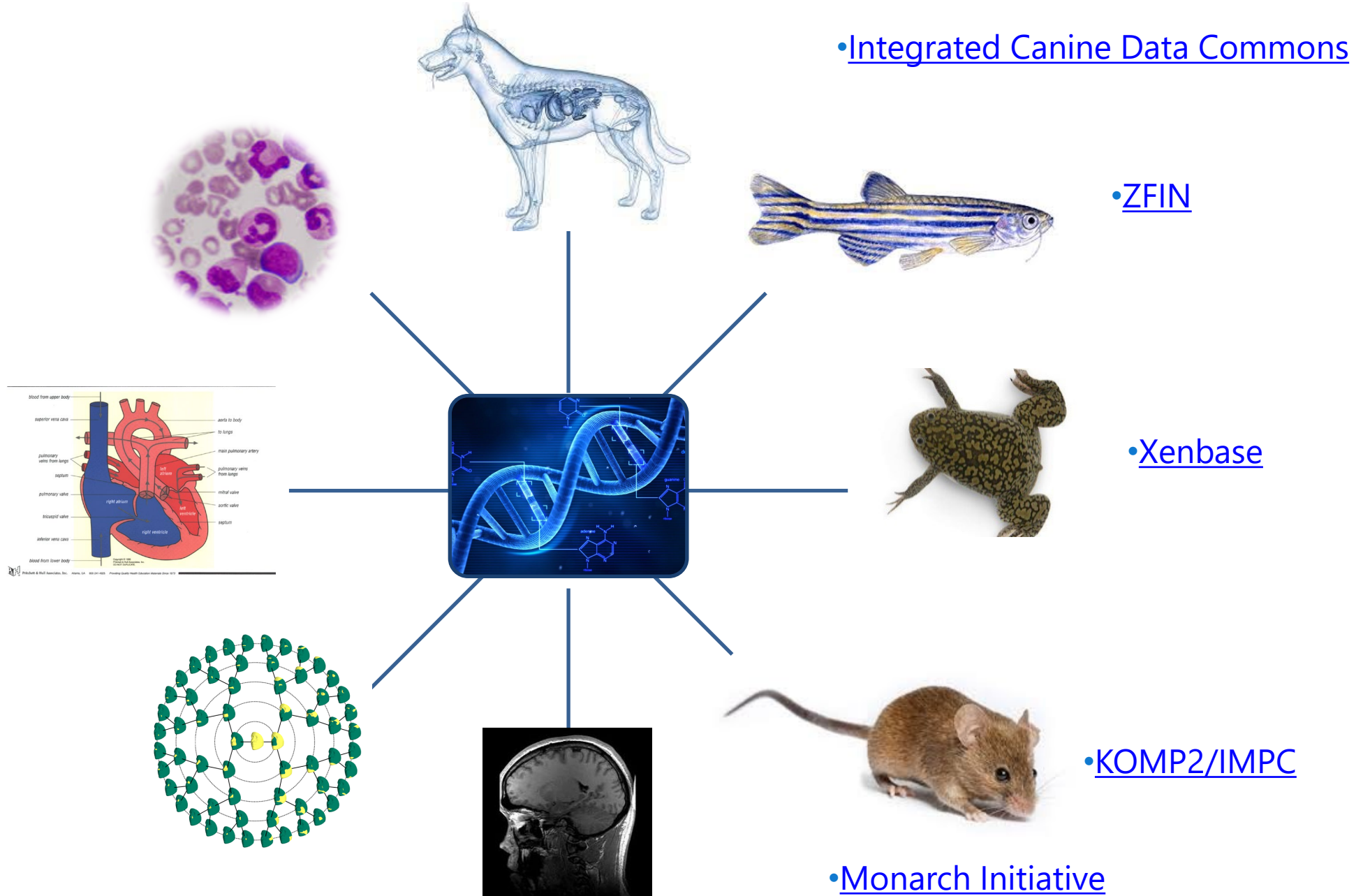
	Kidney and Urinary Tract Defects Ali Gharavi, Columbia University
	Structural birth defects and childhood cancer Xiang Wang, Children's Hospital of Philadelphia
	Neuroblastoma Sharon Diskin, Children's Hospital of Philadelphia
	CHARGE syndrome Donna Martin, Michigan Medicine
	Laterality Birth Defects Stephanie Ware, Indiana University
	Structural Brain Defects, Neural Tube Defects Joe Gleeson, University of California, San Diego

Webinar Information:

<https://monarch-initiative.github.io/phenomics/pages/clin-phen-webinar.html>

Part 3: Cross-Species Genotype-Phenotype Analysis

Part 3 Blog



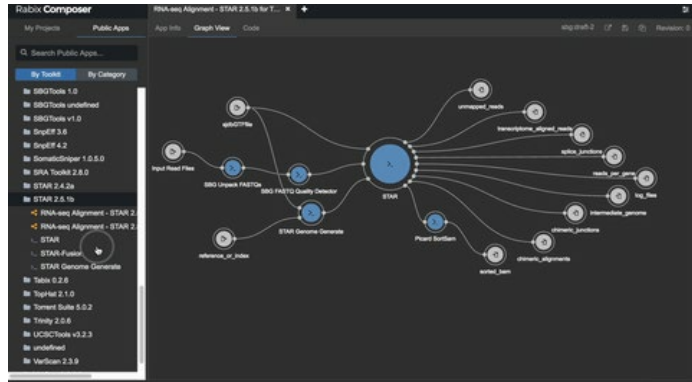
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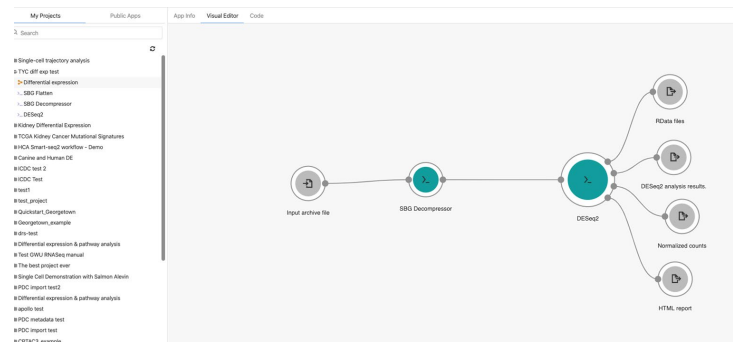


Seven Bridges CAVATICA: Use or Create Workflows

Build, use, optimize, share new or existing workflows



Tailor a workflow with a Workflow Editor



SevenBridges: <https://docs.sevenbridges.com/>
CWL: <https://www.commonwl.org/>

Researchers bring multiple datasets together and run workflows over the data in secure private workspaces

The screenshot shows the CAVATICA web interface. At the top, there's a navigation bar with 'CAVATICA' and various tabs like 'Projects', 'Data', 'Public Apps', etc. Below this, there's a section titled 'Integrated Kids First/TARGET Analysis'. The main part of the interface is a table with columns for 'Files', 'Name', 'Case ID', and 'Sample ID'. The table lists several datasets, each with a unique identifier and a link to the file. The table is paginated, showing 1-100 of 989 items.

Files	Name	Case ID	Sample ID
<input type="checkbox"/>	TARGET 00e47068-3d0c-4b3d-ba10-8371a1d33bc.bam	TARGET-30-PAPVEB	TARGET-30-PAPVEB-04A
<input type="checkbox"/>	KIDS-FIRST 01102f6e-e2e6-4396-850f-3c70d89a8dbb.cram	PT_T48K7XD1	BS_ZTWWV1Y9
<input type="checkbox"/>	KIDS-FIRST 01859ccc-d993-4936-8bae-d8751b04afe9.cram	PT_JK6KETOG	BS_DBF8B5M2
<input type="checkbox"/>	KIDS-FIRST 018edf6c-1a7e-475b-a451-9f3b5bdf11c3.cram	PT_PSYSYSZ4	BS_CCGFJW3A
<input type="checkbox"/>	TARGET 01c87dd9-4a5d-4cdd-844d-749394da9e96.bam	TARGET-30-PASWUJ	TARGET-30-PASWUJ-01A
<input type="checkbox"/>	TARGET 02a86e96-346e-4ffe-bada-23b1823416b6.bam	TARGET-30-PASWYR	TARGET-30-PASWYR-01A
<input type="checkbox"/>	KIDS-FIRST 02d2d6f8-a5db-4c7e-a196-58d302d14d95.cram	PT_8BJ43FRQ	BS_SVJPNF2

Federate a new (or existing) tool with the Data Resource!

Data Resource Portal

Entry point. Query, search, discover, build & visualize synthetic cohorts

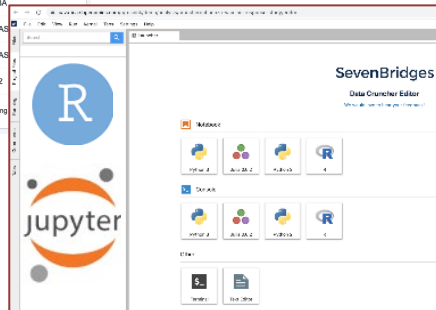


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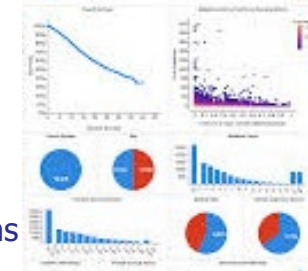
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Name	Case ID	Sample ID
TARGET_00e47068-3d3c-4b3d-ba10-8371a1d33c3c.bam	TARGET-30-PAPVEB	TARGET-30-PAPVEB-04A
KIDS-FIRST_01102766-626f-4396-850f-3c70d89a5b3b.cram	PT_T4BK7XD1	BS_7TWWY1Y9
KIDS-FIRST_01859ccc-0993-493b-8bae-08751b04e4f9.cram	PT_J6K6ETOG	BS_DBF88M2
KIDS-FIRST_018edf5c-1a7e-475b-a451-9f3b5bd11c3.cram	PT_PSYYSZ4	BS_CCGFJW3A
TARGET_01c87d09-4a5d-4cdd-844d-749394dada96.bam	TARGET-30-PASWJ	TARGET-30-PASWJ
TARGET_02a86e96-346e-4f8a-bada-23b182341b66.bam	TARGET-30-PASWYR	TARGET-30-PASWYR
KIDS-FIRST_02d2d6f8-a5db-4c7e-a196-58d302d14d95.cram	PT_8U43FRQ	BS_5VJPNF2



Knowledge Base Integrations (PedcBioPortal)

Integrations with existing curated/published data visualizations



Data Services

Model clinical data in FHIR-based data services and API for sharing layers of harmonized data



aws
STRIDES

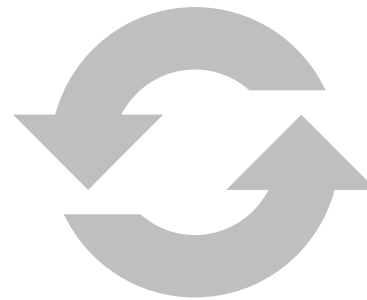
GEN3
DATA COMMONS

Framework Services

Index and point to files in the cloud (for approved users)

NIH Cloud Based Platforms Interoperability (NCPI)

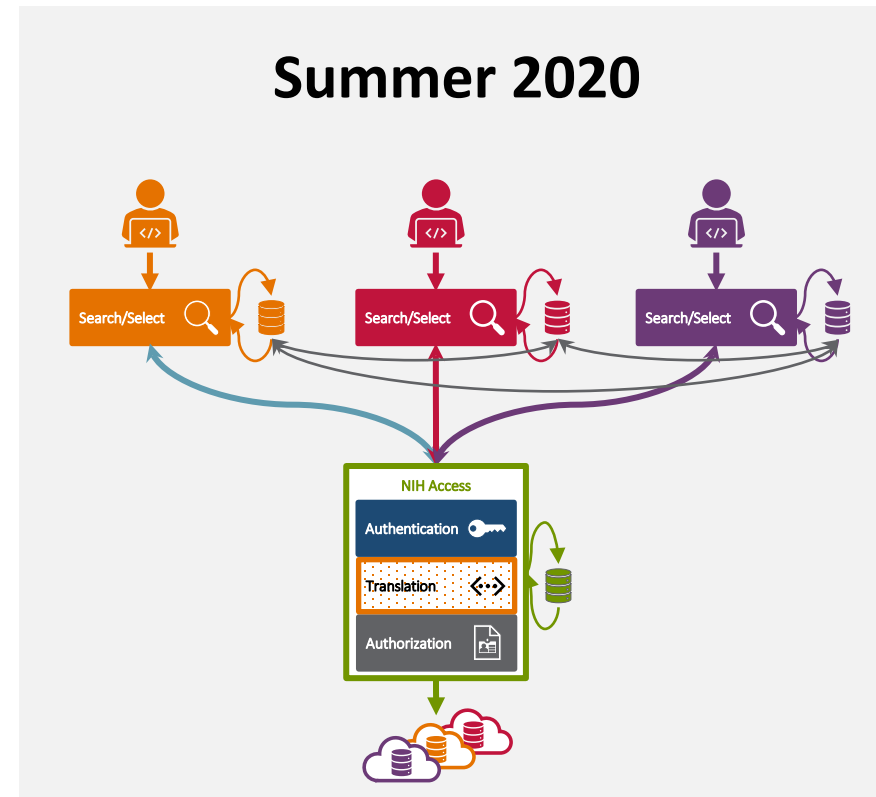
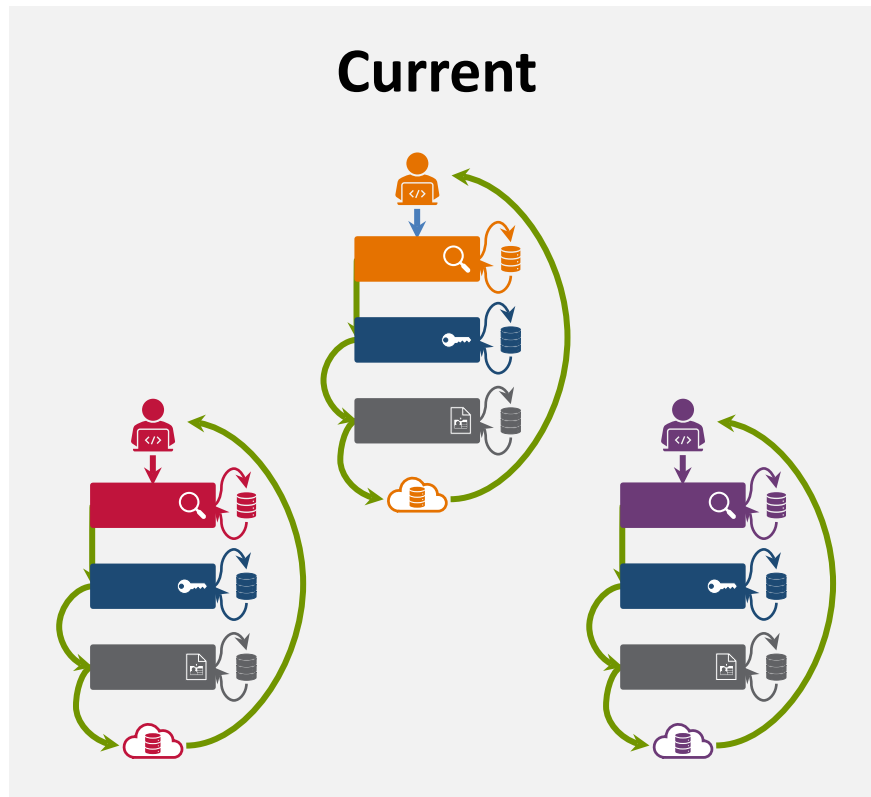
*Goal: Empower end-user analyses across platforms
through federation & interoperability*



NIH Researcher Auth Services (RAS)

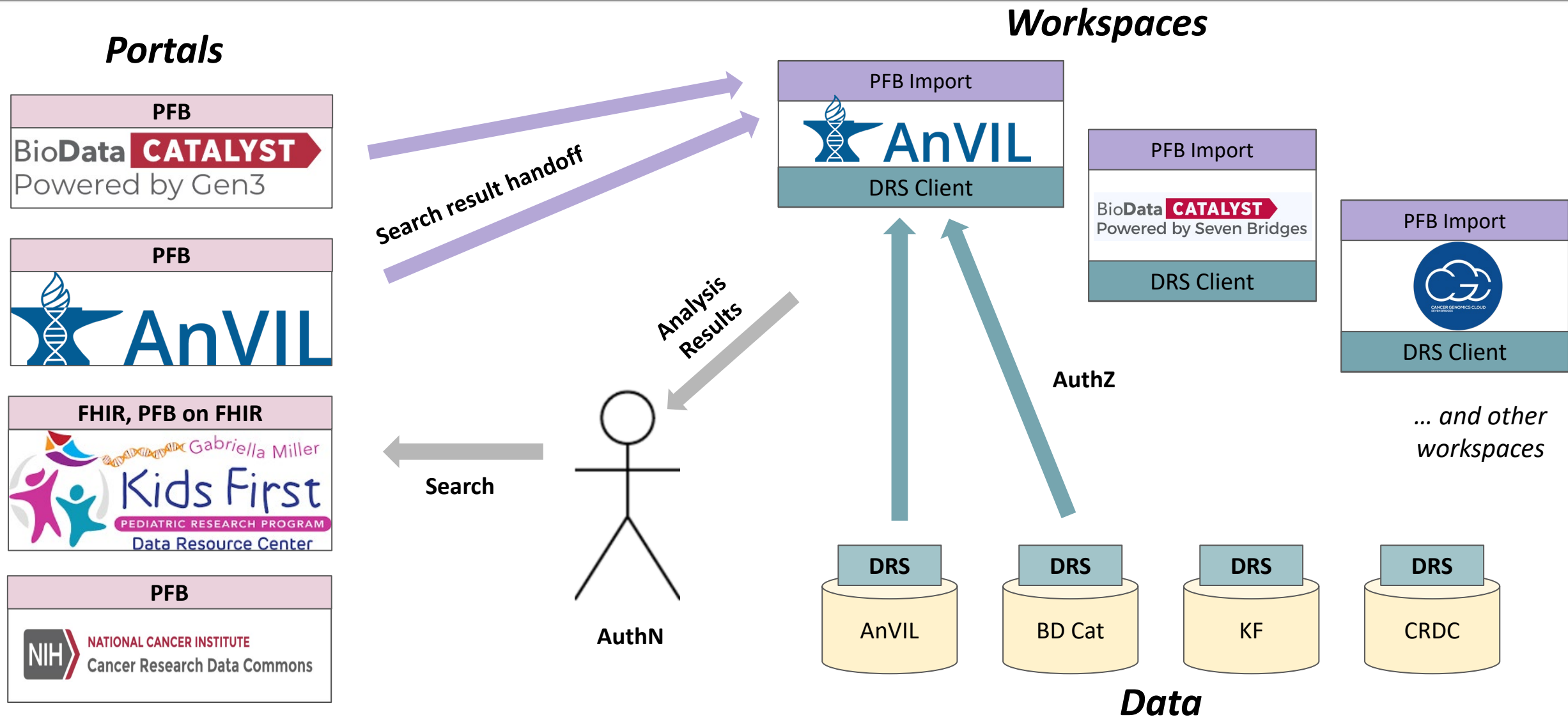
Simplify researcher access to NIH data through federated **authentication** (linking user identity account; “passport”) and **authorization** (claim to access specific studies/datasets; “visa”)

<https://datascience.nih.gov/data-infrastructure/researcher-auth-service>



Adapted from Susan Gregurick, ODSS

Systems Interoperation WG - Technical 1st Year Vision



Key NCPI Standards to Consider

NCPI: <https://datascience.nih.gov/nih-cloud-platform-interoperability>

- Fast Healthcare Interoperability Resources (FHIR; [NOT-OD-19-122](#))
 - Tools that exchange or handle Kids First clinical, phenotypic, or meta-data should consider implementing FHIR standards to interact with the Kids First Data Resource's FHIR server and API
- NIH Researcher Auth Service (RAS; <https://auth.nih.gov/docs/RAS/>)
 - Tools accessing controlled-access data should consider implementing the ability to “consume” RAS for authentication and authorization
- Data Repository Service (DRS; <https://github.com/ga4gh/data-repository-service-schemas>)
 - Tools handling Kids First data (which is stored in the cloud) should consider implementing the ability to “consume” DRS for referencing data objects

Examples: Improve Discovery by...

1. Collecting, extracting, submitting deeper data or new data types associated with Kids First datasets
2. Harmonizing or processing data to promote cross-disease or cross-species (or cross-dataset analysis)
3. Portal analysis workflows to deploy within the Kids First Data Resource (e.g., CAVATICA)
4. Creating or integrating, a new or separate tool to federate with the Kids First Data Resource
5. Consenting for broader data sharing

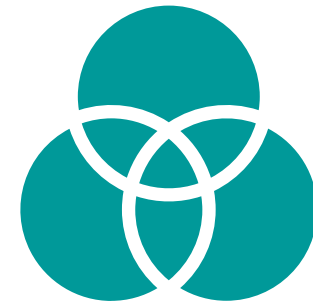
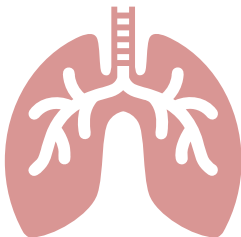


Standard Data Use Limitations (DULs)

The [NIH Genomic Data Sharing Policy](#) “expects investigators generating genomic data to seek consent from participants for future research uses and the **broadest possible sharing.**”

- General Research Use → Broadest
- Health/Medical/Biomedical
- Disease Specific:

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 is a primary goal of Kids First!

Institutional Certification: controlled-access data

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) basis in the informed consent from the participants or in special knowledge

Data Use Limitation Modifiers (Optional)

IRB Approval Required	IRB	Requestor must provide documentation
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.

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

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 <input type="checkbox"/> PUB <input type="checkbox"/> COL <input type="checkbox"/> NPU <input type="checkbox"/> MDS <input type="checkbox"/> GSO <input type="checkbox"/>
Eg: Cold Cohort Study	Disease Specific Research [Lung Cancer]	IRB <input type="checkbox"/> PUB <input type="checkbox"/> COL <input type="checkbox"/> NPU <input checked="" type="checkbox"/> MDS <input type="checkbox"/> GSO <input type="checkbox"/>
<div></div>	General Research Use	IRB <input type="checkbox"/> PUB <input type="checkbox"/> COL <input type="checkbox"/> NPU <input type="checkbox"/> MDS <input type="checkbox"/> GSO <input type="checkbox"/>
<div></div>	Select consent group title	IRB <input type="checkbox"/> PUB <input type="checkbox"/> COL <input type="checkbox"/> NPU <input type="checkbox"/> MDS <input type="checkbox"/> GSO <input type="checkbox"/>
<div></div>	Select consent group title	IRB <input type="checkbox"/> PUB <input type="checkbox"/> COL <input type="checkbox"/> NPU <input type="checkbox"/> MDS <input type="checkbox"/> GSO <input type="checkbox"/>



Consent Considerations

- The [NIH Genomic Data Sharing Policy](#) “expects investigators generating genomic data to seek consent from participants for future research uses and the **broadest possible sharing**.”
- Example from [NHGRI Informed Consent Resource \(https://www.genome.gov/27565449/the-informed-consent-resource/\)](https://www.genome.gov/27565449/the-informed-consent-resource/):
 - *“Your samples, genomic data and health information will be stored and shared with other researchers. The samples and information will be available for any research question, such as research to understand what causes certain diseases (for example heart disease, cancer, or psychiatric disorders), development of new scientific methods, or the study of where different groups of people may have come from.”*
- DUL definitions and considerations:
 - Points to Consider for Institutions and Institutional Review Boards in Submission and Secondary Use of Human Genomic Data under the National Institutes of Health Genomic Data Sharing Policy: https://osp.od.nih.gov/wp-content/uploads/GDS_Points_to_Consider_for_Institutions_and_IRBs.pdf



Other R03s and Analysis Opportunities...

These activities should strengthen Kids First X01 datasets and/or enhance the functionality of the Kids First Data Resource to improve or facilitate future analyses by the broader research community, but applications do not have to address specific analyses themselves.

For applicants seeking to analyze Kids First data, see [PAR-19-375](#). For applicants seeking to analyze other Common Fund datasets see [RFA-RM-21-007](#).



R03-PAR (IC funds)

Small Research Grants for Analyses of Gabriella Miller Kids First Pediatric Research Data

- Support analyses of Kids First X01 datasets
- R03
- IC funds (NICHD, NCI, NHLBI, NIAAA, and NIDCR)
- PAR (no set asides)
- Deadline: Standard Receipt Dates, 3 per year
- Combined direct cost budget for the two-year project period may not exceed \$200,000
- Number of awards depends on IC

R03-RFA (KF funds)

Expert-Driven Small Projects to Strengthen Gabriella Miller Kids First Discovery

- Support activities that will enhance the utility of Kids First data
- R03
- Kids First funds
- RFA (set aside)
- Deadline: June 18, 2021
- Combined direct cost budget for the two-year project period may not exceed \$200,000
- 9-14 awards are anticipated from this solicitation

FOAs for Data Analyses

- “Kids First R03-PAR”: <https://grants.nih.gov/grants/guide/pa-files/PAR-19-375.html>
- Common Fund R03: <https://grants.nih.gov/grants/guide/rfa-files/RFA-RM-21-007.html>
- NIH “Parent” R03: <https://grants.nih.gov/grants/guide/pa-files/PA-20-200.html>
- NIH “Parent” R01: <https://grants.nih.gov/grants/guide/pa-files/PA-20-185.html>
- NCI: *Secondary Analysis and Integration of Existing Data to Elucidate the Genetic Architecture of Cancer Risk and Related Outcomes* (Contact: rotunnom@mail.nih.gov)
 - R01: <https://grants.nih.gov/grants/guide/pa-files/PAR-20-276.html>
 - R21: <https://grants.nih.gov/grants/guide/pa-files/PAR-20-277.html>
- NIDCR: *Notice of Special Interest (NOSI) of NIDCR in Supporting Discovery, Characterization, and Mechanistic Study of Genetic Variants Underlying Dental, Oral, and Craniofacial Diseases and Conditions*
<https://grants.nih.gov/grants/guide/notice-files/NOT-DE-19-016.html>
- NIDCR Research Grants for Analyses of Existing Genomics Data (R01) <https://grants.nih.gov/grants/guide/pa-files/PAR-20-045.html>
- NIDCR Small Research Grants for Analyses of Existing Genomics Data (R03) <https://grants.nih.gov/grants/guide/pa-files/PAR-20-046.html>



FOAs for Variant Validation

- **ORIP: *Development of Animal Models and Related Biological Materials for Research (R21 Clinical Trial Not Allowed)*** <https://grants.nih.gov/grants/guide/pa-files/PAR-19-369.html>
- **ORIP: *Resource-Related Research Projects for Development of Animal Models and Related Materials (R24 Clinical Trials Not-Allowed)*** <https://grants.nih.gov/grants/guide/rfa-files/RFA-OD-19-027.html>
- **NIDCR: *Mechanistic Studies of Gene-Environment Interplay in Dental, Oral, Craniofacial, and Other Diseases and Conditions (R01 Clinical Trial Not Allowed)***. <https://grants.nih.gov/grants/guide/pa-files/PAR-19-292.html>
- **NIDCR: *Development of Novel and Robust Systems for Mechanistic Studies of Gene-Environment Interplay in Dental, Oral, Craniofacial, and Other Diseases and Conditions (R21 Clinical Trial Not Allowed)***. <https://grants.nih.gov/grants/guide/pa-files/PAR-19-293.html>
- **NHGRI: *Novel Approaches for Relating Genetic Variation to Function and Disease (R01 Clinical Trial Not Allowed)*** <https://grants.nih.gov/grants/guide/pa-files/pa-18-868.html>
- To pursue collaborations with the [Knockout Mouse Phenotyping Program \(KOMP2\)](#), contact: KidsFirstKOMP@nih.gov



FAQ #1

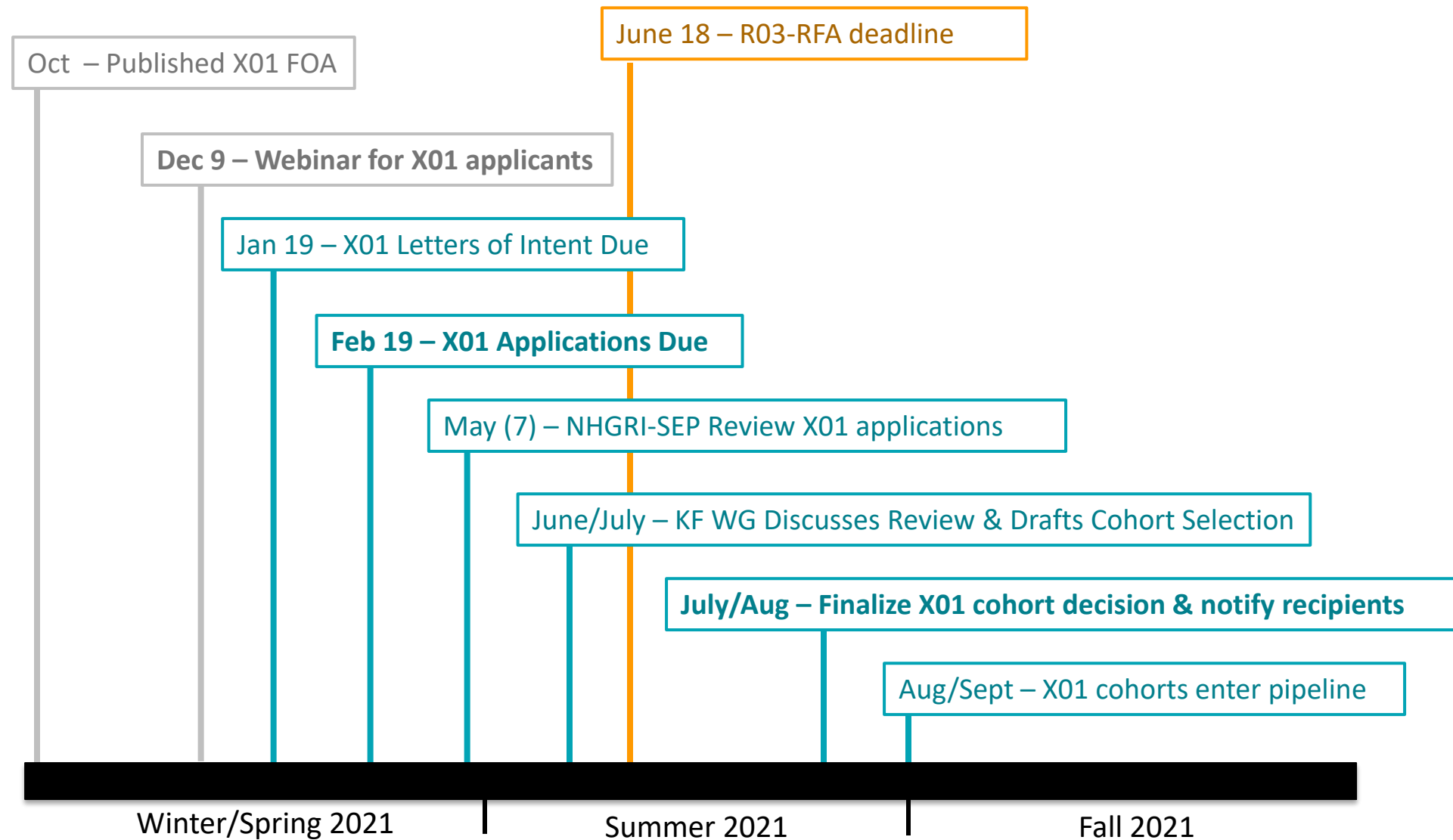
Question: I applied for the FY21 X01 sequencing opportunity, but I would like to apply for the expert-driven R03-RFA to work with colleagues to improve the associated dataset. When will X01 decisions be announced, and should I apply to RFA-RM-21-011?



Answer: Can you make a compelling argument that the activity improve Kids First regardless of the X01? Then yes, apply.



2021 X01 Timeline



FAQ #2

Question: I am planning to propose building and optimizing a workflow that can be deployed in CAVATICA. Are cloud credits available for testing/running the tool?



Answer: Cloud credits are currently only available to X01s via a separate review process. However, there is no guarantee cloud credits will be approved for this purpose, so you should incorporate these cloud costs into your R03 budget.

→ Cost estimate resources: [DRC FAQs](#)



FAQ #3

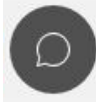
Question: What standards have been adopted by Kids First? What other standards might be relevant?



Answer: See attached list (will be posted on our FAQ page soon!)



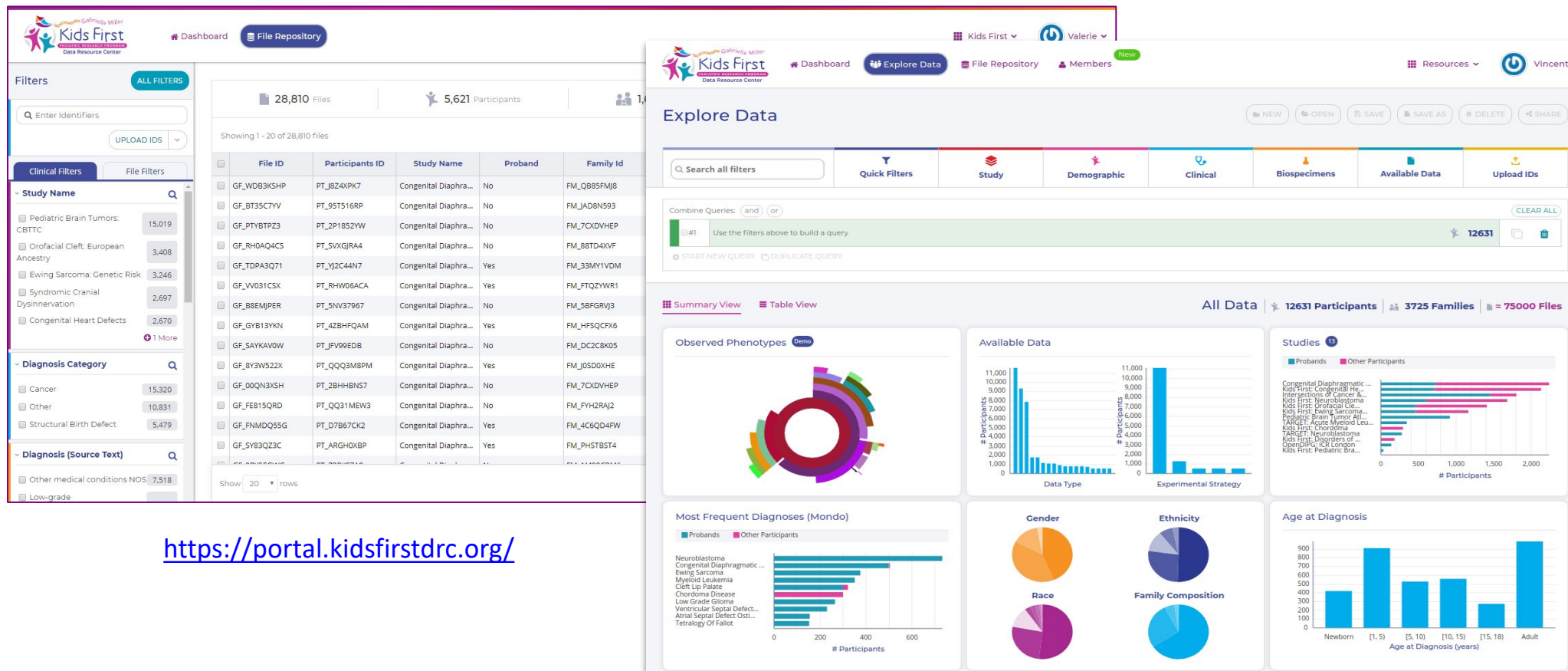
Q & A

- Use the Q&A bar (lower right of your screen) to send your questions to “**All Panelists**”. We will read your questions out loud and answer them.
- You can also use the “chat”  service to send private messages to the host or presenters.



Portal demo

Anyone can
register & login
to the portal to
filter, search,
visualize datasets
(build synthetic
cohorts)



<https://portal.kidsfirstdrc.org/>

Submit [dbGaP Data Access Requests \(DARs\)](#)
for controlled-access data

