Webinar Instructions

• Welcome to the Pre-Application Webinar for the Development of the Gabriella Miller Kids First Pediatric Data Resource Center (U2C)

• Every participant is muted upon entry. Please remain muted until the question/discussion period. You can unmute yourself by clicking on the mic symbol to the right of your user name or by selecting *6 on your telephone.

• You can ask technical questions using the chat service to the host throughout the webinar. Please save questions related to the program or the FOA for the question period. Additional program-related questions can be emailed to: KidsFirst@od.nih.gov.

This Webinar will be recorded. We will start at 11:00 am.
Pre-Application Webinar to Discuss the Development of the Gabriella Miller Kids First Pediatric Data Resource Center (U2C)

July 29th, 2016
Webinar Introductions

• Jonathan Kaltman, NHLBI & Kids First contact
• Tracee Foster, NHLBI Grants Management
• Other Kids First program staff online:
  – Lorette Javois, NICHD
  – Jaime Guidry Auvil, NCI
  – Marie Nierras, Common Fund
Gabriella Miller Kids First Pediatric Research Program: Background

- Initiated in response to the **2014 Gabriella Miller Kids First Research Act**
  - Signed into law on April 3, 2014
  - Ended taxpayer contribution to presidential nominating conventions
  - Transferred remaining $126 million into the Pediatric Research Initiative Fund
  - Authorized appropriation of $12.6 million per year for 10 years to the NIH Common Fund for pediatric research; first appropriation was for FY2015
The Gabriella Miller Kids First Pediatric Research Program (Kids First) is a trans-NIH effort supported by the NIH Common Fund. Facilitates involving multiple NIH Institutes in high-impact efforts. Common Fund is run by the Office of Strategic Coordination, Office of the NIH Director. Institutes that chair Kids First:

- NHGRI
- NHLBI
- NCI
- NICHD
- (Several others are part of our Working Group)
Gabriella Miller Kids First Pediatric Research Program: **Overall Goals**

Develop a data resource for the pediatric research community to access well-curated phenotype and sequence data that will help determine the biological basis of childhood cancers and structural birth defects.

1. Cohort identification and enrichment
2. Web-based Data Resource development - integrating genomic and clinical data in a central portal where these data and analysis tools will be readily accessible to promote comprehensive and cross-cutting research and collaboration within and across various types of pediatric diseases
3. Pilot projects using the data resource to mine, aggregate, link, and analyze data
GMKF Major Initiatives

Activities and Timeline

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- Cohort ID (X01) & Sequencing (U24)
- NICHD R03 for Data Analysis
- Pediatric Data Resource (U2C)
- New Insights from Data Mining/Demonstration Projects
Data Sources

• Discovery of the Genetic Basis of Childhood Cancers and of Structural Birth Defects: Gabriella Miller Kids First Pediatric Research Program (X01)
    • 2 childhood cancer (more expected in subsequent years)
    • 5 structural birth defects
  – FY-2016 (PAR-16-150) project cohorts will be determined in October 2016

• Small Research Grants for Analyses of Data for the Gabriella Miller Kids First Data Resource (R03) – PAR-16-348
  – May bring in sequence data from external pediatric cancer or structural birth defect cohorts
Development of the Gabriella Miller Kids First Pediatric Data Resource Center (U2C)

- **RFA-RM-16-010**
- Goal: accelerate discovery of genetic etiology and shared biologic pathways within and across childhood cancers and structural birth defects
- By enabling data
  - Aggregation
  - Access
  - Sharing
  - Analysis

Whole Genome Sequence + Phenotype
## Gabriella Miller Kids First Pediatric Data Resource Center

| **Data Resource Portal** | • Web-based, public facing platform  
• House, organize, index, and display data and analytic tools |
|--------------------------|------------------------------------------------------------------|
| **Data Coordinating Center** | • Facilitate deposition of sequence and phenotype data into relevant repositories  
• Harmonize phenotypes |
| **Administrative and Outreach Core** | • Develop policies and procedures  
• Facilitate meetings and communication  
• Educate and seek feedback from users |
Birth defect or childhood cancer cohorts

Sequencing center

DNA

Birth defect BAM/VCF

dbGaP

Phenotype

Birth defect VCF

NCI GDC

Cancer BAM/VCF

Gabriella Miller
Kids First Data
Resource

Index of datasets
Phenotype
Variant summaries

Users

Birth defect BAM/VCF

Cancer VCF

Cancer BAM/VCF
Example Use-Cases

1. Serves as a comprehensive catalog to inform the science community of what data exists in the field

2. Investigate within and/or across different types of structural birth defects and/or childhood cancers. Example: “Does a set of genes associated with cleft-lip, overlap with genes associated with congenital heart disease and/or childhood cancer?”

3. Explore how a gene variant identified from an animal model may affect a human phenotype. Example: “Does a gene variant identified from an animal model that shows developmental defects occur in people, and what is the phenotype?”

4. Query the resource to investigate gene function in an animal model. Example: “How does a gene, identified as leading to a congenital heart defect in kids, control the developmental pathway in a mouse model?”
Data Resource Center Timeline

- Sept 20 – Letter of Interest Due
- Oct 20 – Applications Due
- Feb – Scientific Merit Review
- June 6 – NHLBI Council Final Level Review of Applications
- Target: July – Award Grant
- Sept – Data Resource “Kickoff” Meeting

Timeline:
- Fall 2016
- Spring 2017
- Summer 2017
- Fall 2017
Questions