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

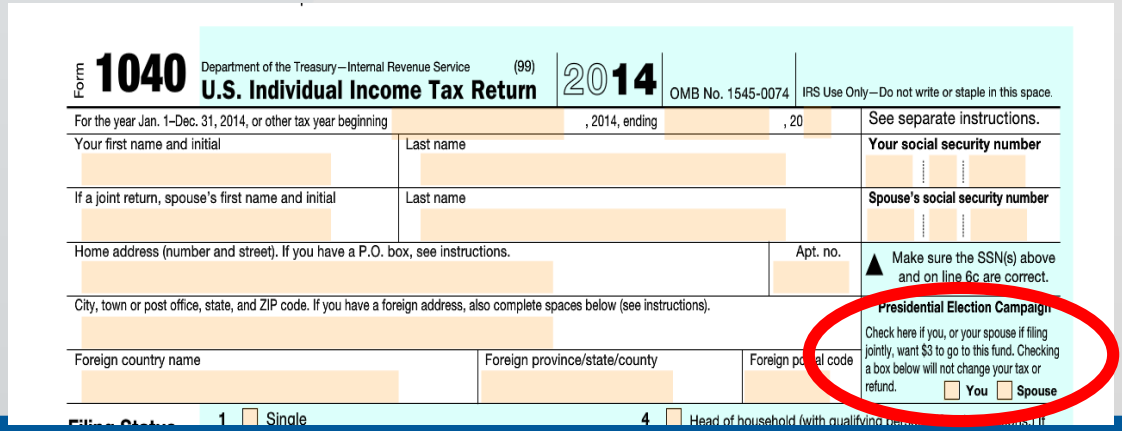
NIH National Institutes of Health
Office of Strategic Coordination - The Common Fund

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


Form **1040** Department of the Treasury—Internal Revenue Service (99) **U.S. Individual Income Tax Return** 2014 OMB No. 1545-0074 IRS Use Only—Do not write or staple in this space.

For the year Jan. 1–Dec. 31, 2014, or other tax year beginning , 2014, ending , 20 See separate instructions.

Your first name and initial | Last name | Your social security number

If a joint return, spouse's first name and initial | Last name | Spouse's social security number

Home address (number and street). If you have a P.O. box, see instructions. Apt. no.

▲ Make sure the SSN(s) above and on line 6c are correct.

City, town or post office, state, and ZIP code. If you have a foreign address, also complete spaces below (see instructions).

Foreign country name | Foreign province/state/county | Foreign postal code

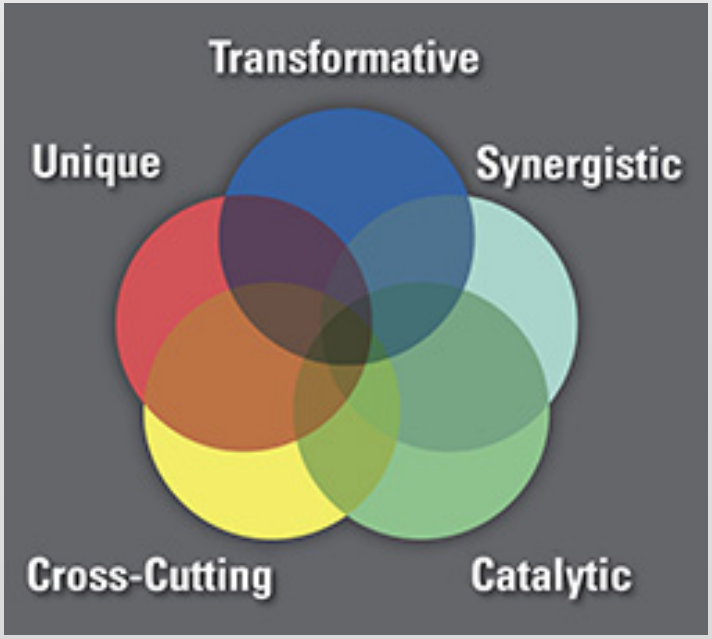
Presidential Election Campaign

Check here if you, or your spouse if filing jointly, want $3 to go to this fund. Checking a box below will not change your tax or refund. ☐ You ☐ Spouse

Filing Status 1 ☐ Single 4 ☐ Head of household (with qualifying

Gabriella Miller Kids First Pediatric Research Program: Background

- 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)



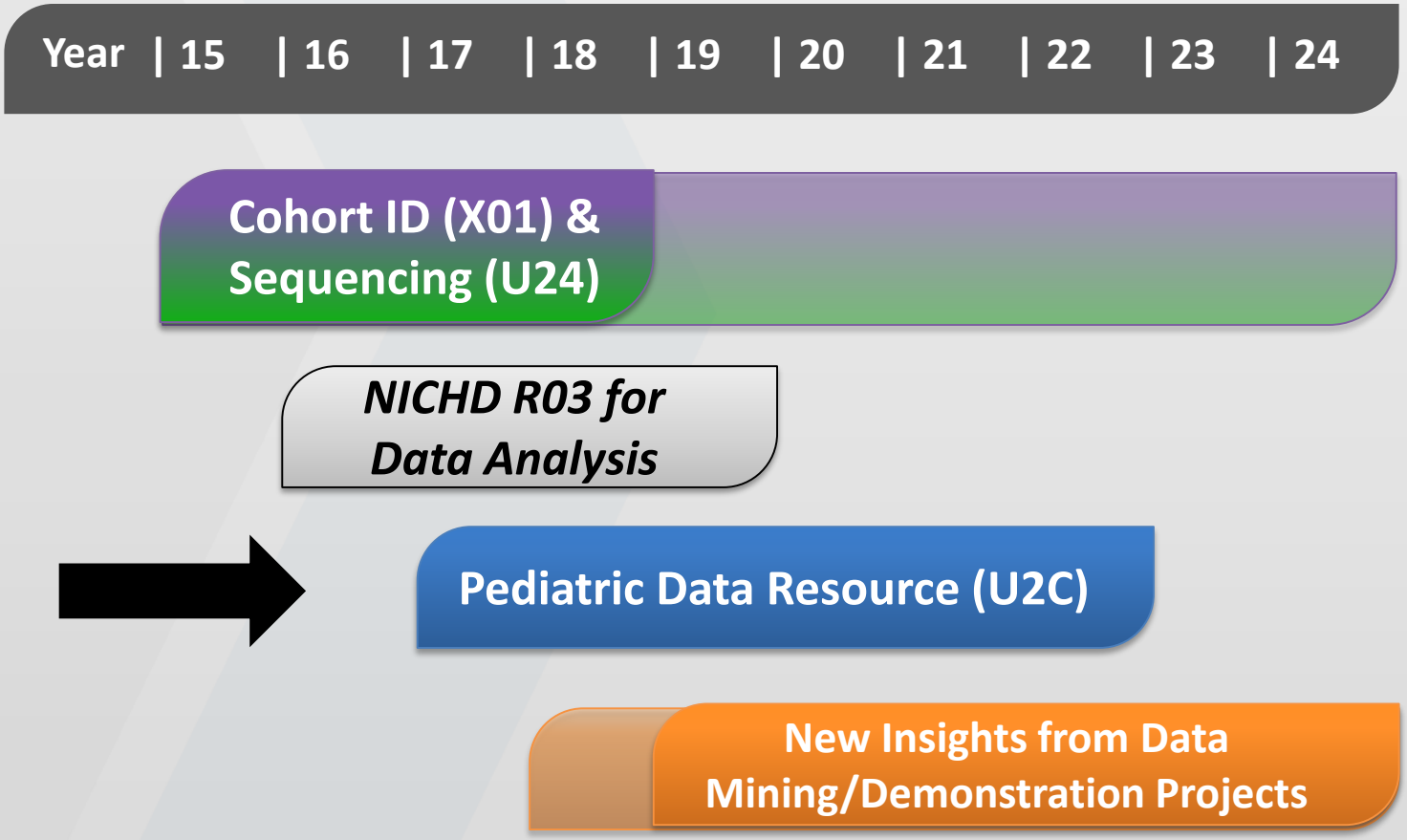
Gabriella Miller Kids First Pediatric Research Program: Overall Goals

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



Data Sources

- Discovery of the Genetic Basis of Childhood Cancers and of Structural Birth Defects: Gabriella Miller Kids First Pediatric Research Program (X01)
  - FY-2015 project cohorts (PAR-15-259):
    - 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 defects cohorts

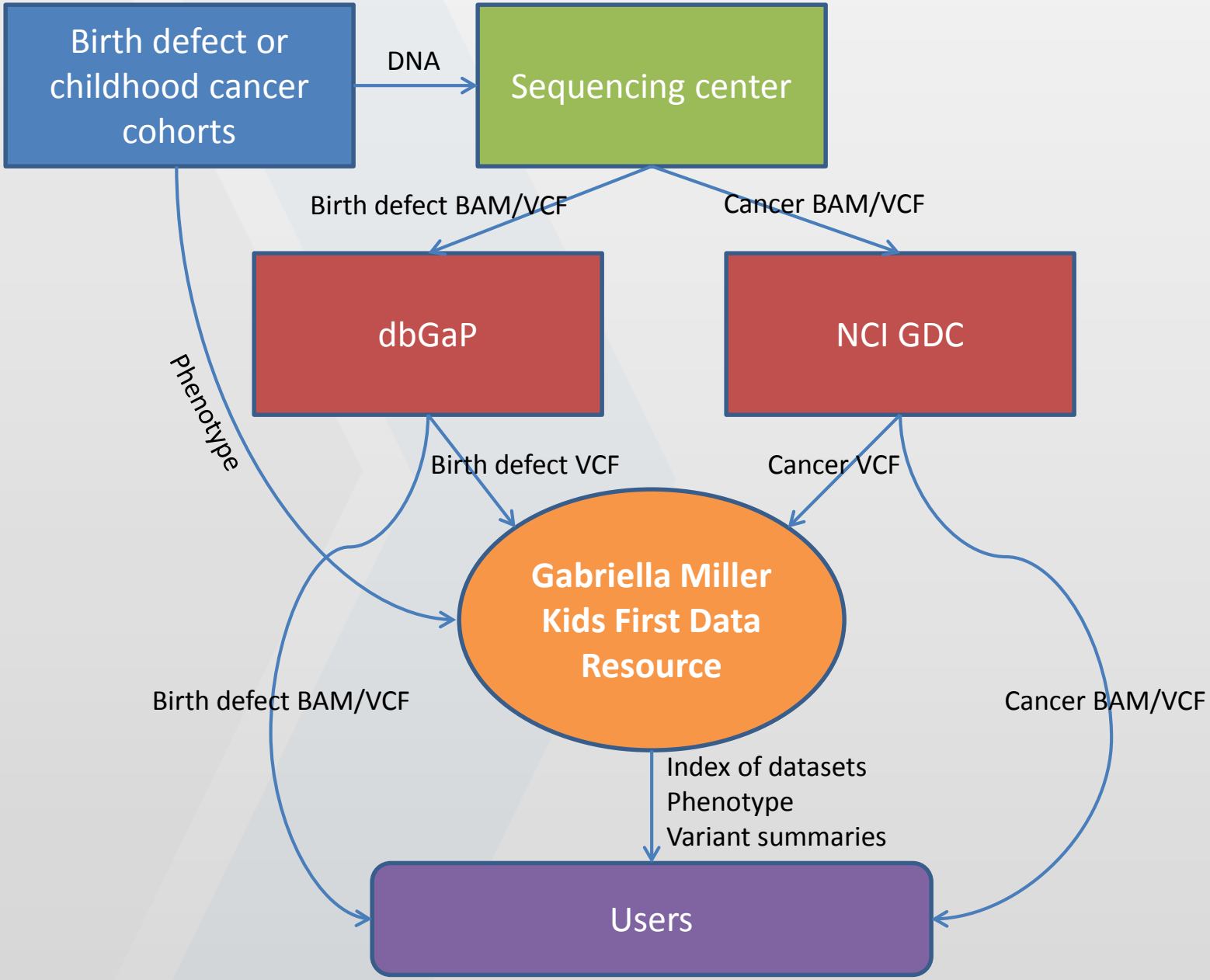
Development of the Gabriella Miller Kids First Pediatric Data Resource Center (U2C)

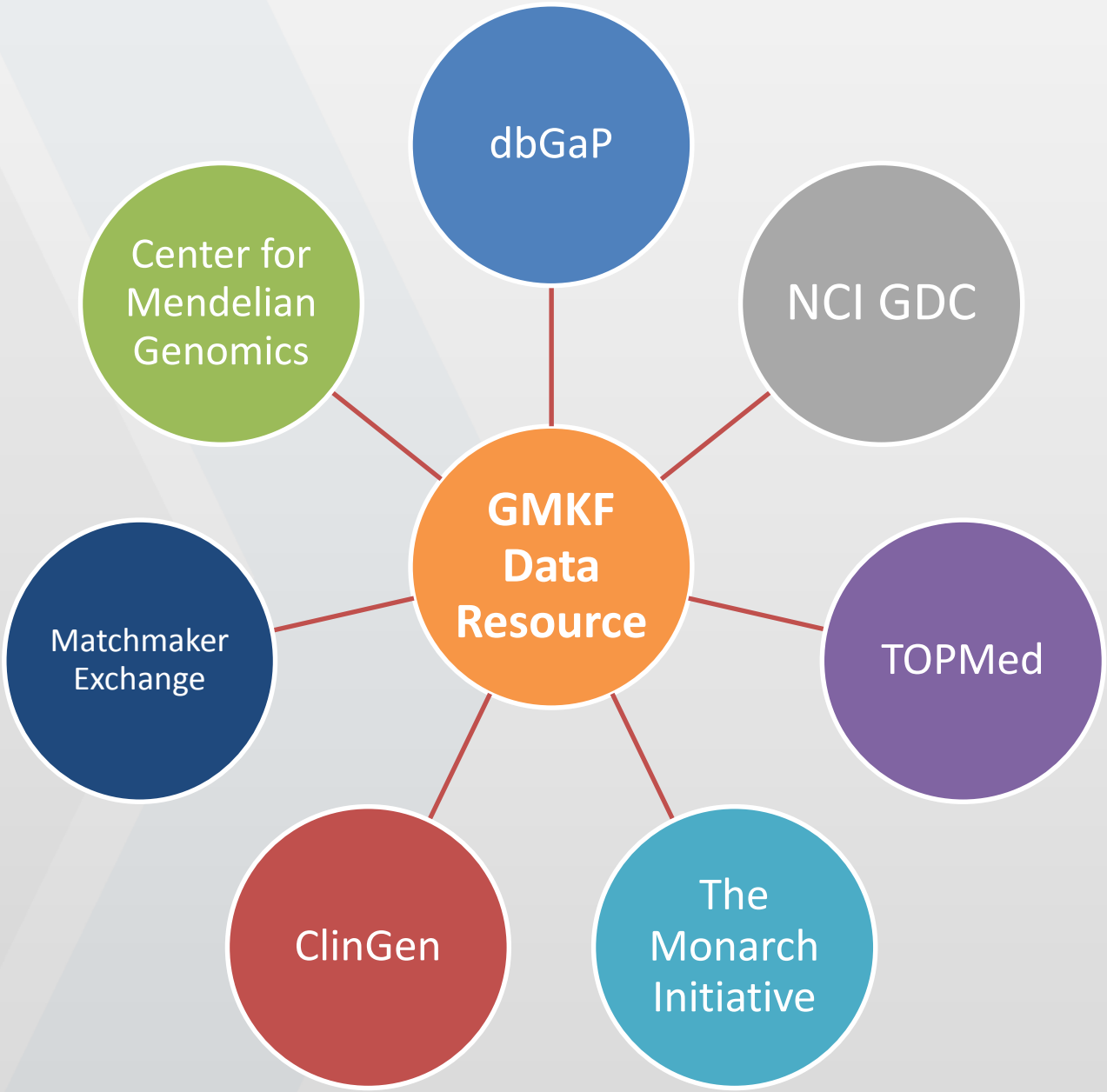
- RFA-RM-16-010
- Goal: accelerate discovery of genetic etiology and shared biologic pathways within and across childhood cancers and structural birth defects
- By enabling data
  - Aggregation
  - Access
  - Sharing
  - Analysis

} 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

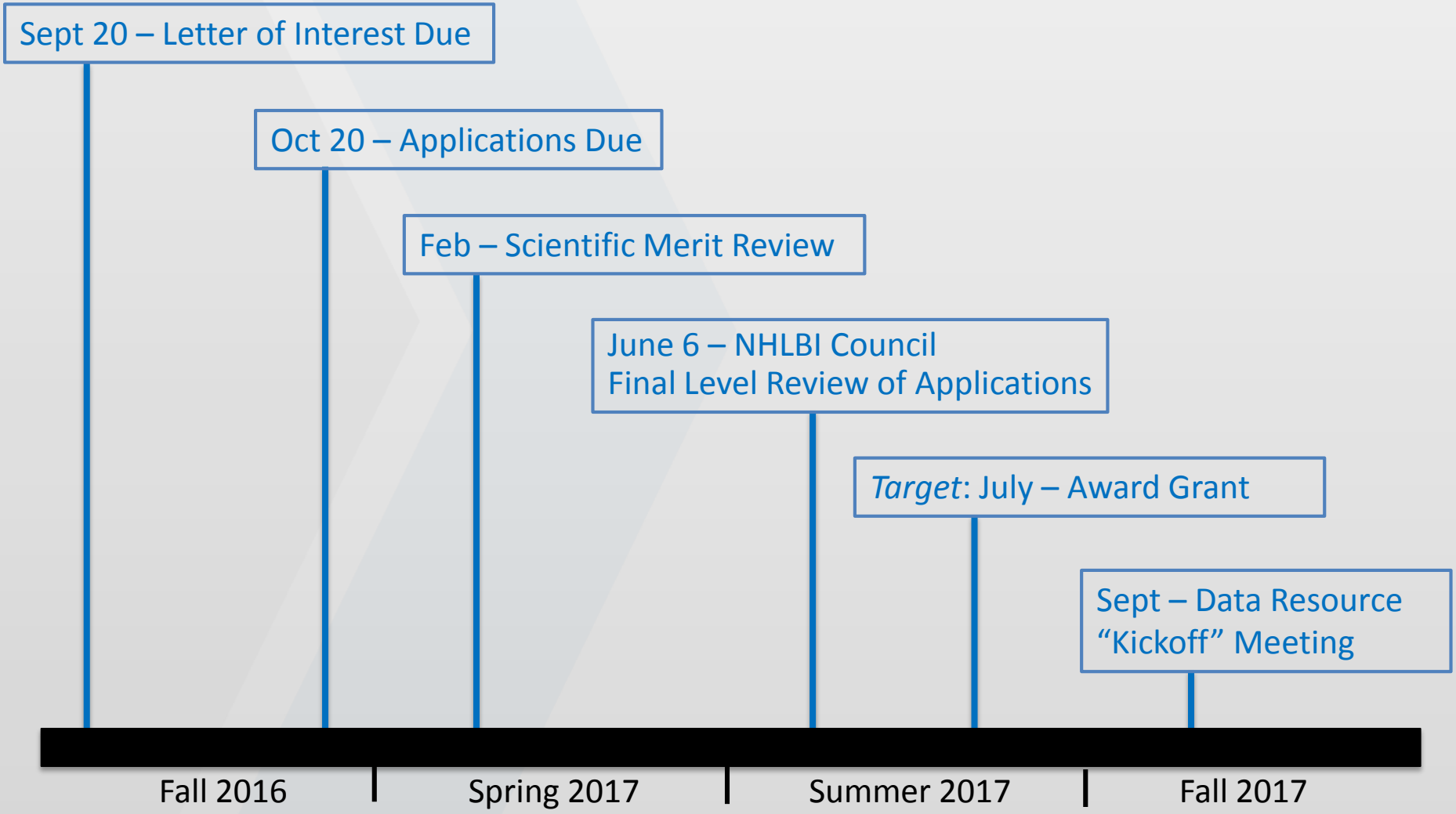

Birth defect or childhood cancer cohorts
DNA
Sequencing center
Birth defect BAM/VCF
Cancer BAM/VCF
dbGaP
NCI GDC
Phenotype
Birth defect VCF
Cancer VCF
Gabriella Miller Kids First Data Resource
Birth defect BAM/VCF
Cancer BAM/VCF
Index of datasets
Phenotype
Variant summaries
Users


dbGaP
Center for Mendelian Genomics
NCI GDC
GMKF Data Resource
Matchmaker Exchange
TOPMed
ClinGen
The Monarch Initiative

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



Questions