Kids First Started with Gabriella Miller’s Pediatric Cancer Advocacy Empowering Research Across Pediatric Conditions

Oct 2013
Gabriella Miller childhood cancer advocate, died at age 10 from an aggressive brain cancer

April 2014
Gabriella Miller Kids First Research Act authorizes $12.6 million/year for 10 years to NIH for pediatric research

Sept 2015
NIH Kids First Program first annual appropriation. The program is funded through FY 2024
The Kids First Working Group is a collaboration to accelerate discoveries in pediatric research. The group is charged with building a cloud-based genomic data resource to share data and accelerate collaborative research leading to better prevention, diagnosis, and treatments for patients and families with pediatric cancer and structural birth defects.

The Kids First Working Group comprises representatives from various National Institutes of Health (NIH) institutes and centers, including:

- NIDCR (National Institute of Dental and Craniofacial Research)
- NINDS (National Institute of Neurological Disorders and Stroke)
- NEI (National Eye Institute)
- NCATS (National Center for Advancing Translational Sciences)
- NIDA (National Institute on Drug Abuse)
- NIDDK (National Institute of Diabetes and Digestive and Kidney Diseases)
- NIEHS (National Institute of Environmental Health Sciences)
- NIAMS (National Institute of Arthritis and Musculoskeletal and Skin Diseases)
- NIAID (National Institute of Allergy and Infectious Diseases)
- ORIP (Office of Research Infrastructure Programs)
- NCIC (National Cancer Institute for Cancer Research)
- CDC (Centers for Disease Control and Prevention)
Association Between Structural Birth Defects and Childhood Cancer

Shared mutations and drug targets across pediatric conditions

Cancer risk increased among children with birth defects:

**BRAF**

**MAPK**

**ALK**
Kids First 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 Empowered by X01 Mechanism, Genome Sequencing Centers, and Data Resource Center. Starts with Patients Cohorts and Investigator Proposals

Data analysis leading to discovery and hypothesis generation and new X01 and/or other NIH research grant proposals

Submit Your Proposals to NIH X01 Mechanism

Data Release to public 6-months later via dbGaP and KF Data Resource Center

NIH Peer-Review

Selected X01 cohort allowing broad data sharing receive access to generate genomic data at KF Sequencing Centers

Collaboration

Data Delivery to X01 investigators

KF Sequencing Centers provide service to generate genomic data from DNA/RNA samples

KF Data Resource Center Performs Clinical Data Review and QC to compile genomes with phenotypes and clinical data

Kids First has supported 63 X01 projects to date

63 X01 projects funded though Kids First program annual appropriation
Kids First’s Cloud-Based Platforms

**Kids First Data Resource Portal** - portal.kidsfirstdrc.org

- **EXPLORE** datasets and build cohorts of participants
- **DISCOVER** harmonized genomic data files for further research
- **CONNECT** data from multiple Kids First 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
Kids First Data Available Today

29 studies at dbGaP

Get started at https://portal.kidsfirstdrc.org

Harmonized data/ 22 birth defect and 15 cancer cohorts/ >28,500 participants
Kids First Data Resource Shares Multiple Data Types Across Pediatric Conditions

• Kids First Data Resource Center allows for multiple data types and cross disease associations
   ▪ better view of the condition’s underlying biology
   ▪ knowledge of common biology across pediatric conditions can speed up the development of diagnostics, treatments, and prevention tools

• Kids First commitment to sharing genomic data associated with clinical data empowers genetic variant discovery
   ▪ within a single disease area or dataset
   ▪ across diagnoses and cohorts
   ▪ allow users to explore similar underlying causes of cancers, structural birth defects, and other rare disorders

• Individual level genomic data are available though dbGaP data access request.
Kids First is Part of a Larger Data Ecosystem

NIH Cloud-Based Platforms Interoperability (NCPI):
Empower end-user analyses across platforms through federation & interoperability
# Kids First Data & the Data Resource Portal

## ABOUT THE DATA

<table>
<thead>
<tr>
<th>22</th>
<th>Birth defect cohorts</th>
</tr>
</thead>
<tbody>
<tr>
<td>15</td>
<td>Cancer cohorts</td>
</tr>
<tr>
<td>32,000+</td>
<td>Study participants</td>
</tr>
<tr>
<td>192,000+</td>
<td>Data files available</td>
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</tbody>
</table>

## ABOUT THE PORTAL

<table>
<thead>
<tr>
<th>50</th>
<th>Unique countries represented by portal users</th>
</tr>
</thead>
<tbody>
<tr>
<td>500+</td>
<td>Approved access requests for secondary data use</td>
</tr>
<tr>
<td>3,389+</td>
<td>Total portal users</td>
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<tr>
<td>6,500+</td>
<td>Total portal logins in 2022</td>
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Acknowledgements

**Kids First Working Group:**
Director co-Chairs: NICHD, NCI, NHLBI, NHGRI
Other Working Group Representation:
NIDCR   NIAAA   NIDDK   NEI   NIAID   ORIP
NIDA    NINDS   NIEHS   NIAMS   NCATS   CDC

**Sequencing Centers:**

[Images of Broad Institute and HudsonAlpha Institute for Biotechnology]