Gabriella Miller Kids First
PEDIATRIC RESEARCH PROGRAM
Data Resource Center

Data Harmonization and Interoperability of Existing Data Repositories:

The Kids First Data Resource Experience

Aug 2, 2022

Symposium on Pediatric Disaster Science

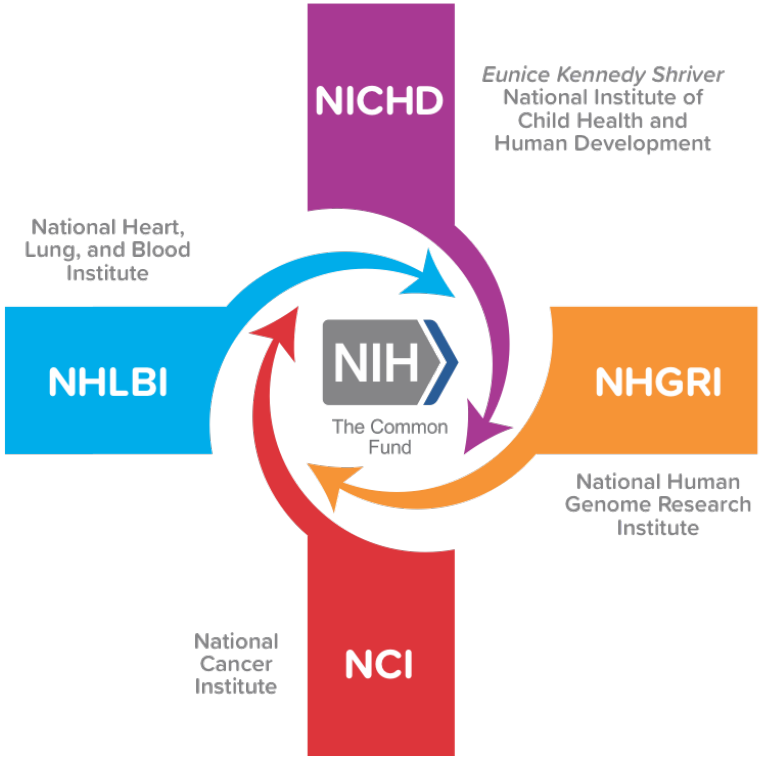
Adam Resnick / d3b.center

Children's Hospital of Philadelphia
Center for Data Driven Discovery in Biomedicine

Follow **@kidsfirstdrc**

Visit **kidsfirstdrc.org**

Kids First Program Vision



Additional Working Group Representation

NIDCR	NIDA
NIAAA	NINDS
NIDDK	NIEHS
NEI	NIAMS
NIAID	NCATS
ORIP	CDC

The Kids First Program was launched via congressional action in 2015

Kids First Program Goal

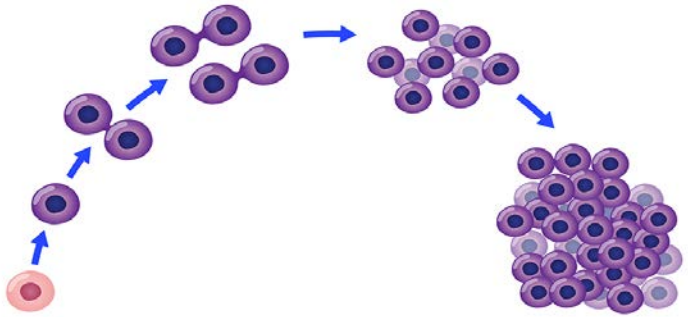
The goal of the Gabriella Miller Kids First Pediatric Research Program (Kids First) is to help researchers **uncover new insights into the biology of childhood cancer and structural birth defects**, including the discovery of shared genetic pathways between these disorders.

Kids First Program 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.**

Can cross-cancer and cross-disease research empower/accelerate discovery?

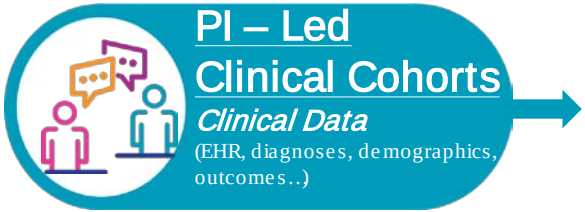
Pediatric Cancer

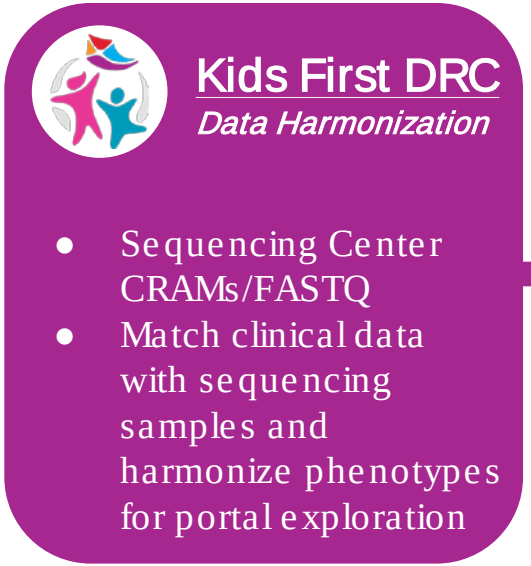


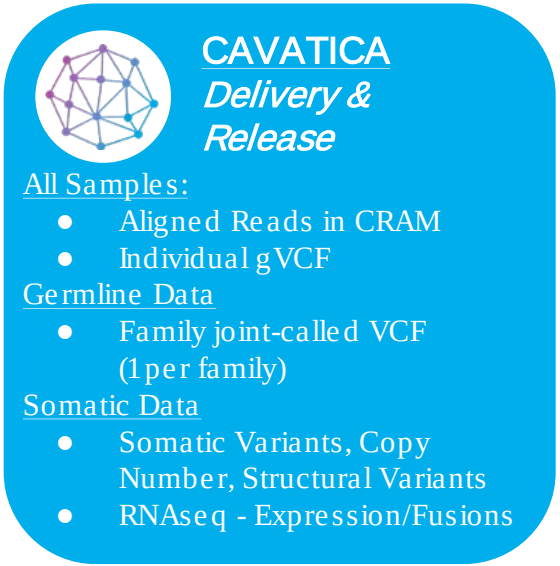
+

Development

Kids First: By the Numbers







Gabriella Miller Kids First Data Resource Center

28,285 Participants

20,222 Families

148,361 Files

1.52 PB of Data

30 Studies Released on the Portal

- Kids First: Orofacial Cleft: African and Asian Ancestry
- Kids First: Neuroblastoma
- Kids First: Myeloid Malignancies
- Kids First: Esophageal Atresia & Tracheoesophageal Fistulas
- Kids First: Disorders of Sex Development
- Kids First: Leukemia & Heart Defects in Down Syndrome
- Kids First: Novel Cancer Susceptibility in Families (BASIC3)
- Kids First: Congenital Diaphragmatic Hernia
- Kids First: Enchondromatosis
- Kids First: Orofacial Cleft – European Ancestry
- Kids First: T Cell ALL
- Kids First: Microtia – Hispanic
- Kids First: Intersections of Cancer & SBD
- Kids First: Orofacial Cleft – Latin American
- Kids First: Familial Leukemia
- Kids First: Craniofacial Microsomia
- Kids First: Syndromic Cranial Dysinnervation
- Kids First: Kidney and Urinary Tract Defects
- Kids First: Adolescent Idiopathic Scoliosis
- Kids First: Hemangiomas (PHACE)
- Kids First: Ewing Sarcoma
- Kids First: Nonsyndromic Craniosynostosis
- Kids First: Congenital Heart Defects
- Kids First: Osteosarcoma

The Gabriella Miller Kids First Program

Big Data + Cloud Platforms + Communities = Ecosystems

Outside this room in the lobby . . .

Realities and metaphors

MOTHERLY

Search...

PREGNANCY PARENTING LIFE SHOP CLASSES VIDEO PODCASTS

Home / Life / Motherly Stories

I never thought I'd be a 'cancer mom'— here's what I want other parents to know

Cancer comes in like a tornado and tears your life apart, and very few people understand that. They tell you to be strong or worse—to stay positive. Some friends show up in the most beautiful way and others stop showing up altogether. The losses hit hard.

By Danielle Moss • July 14, 2022

Realities and metaphors

MOTHERLY

Search...

PREGNANCY PARENTING LIFE SHOP CLASSES VIDEO PODCASTS

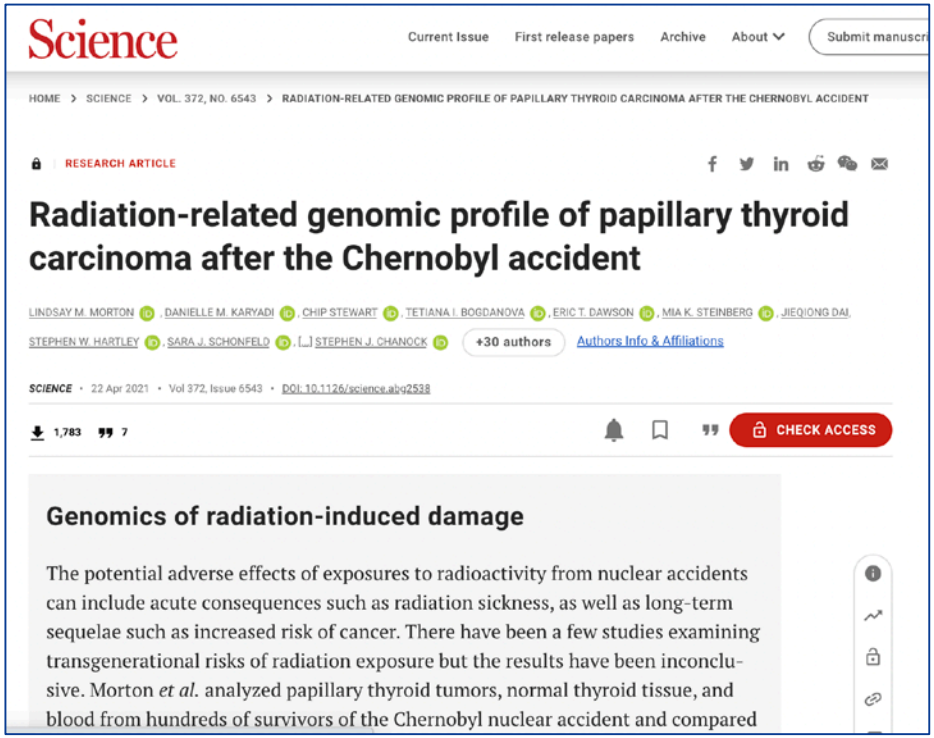
Home / Life / Motherly Stories

I never thought I'd be a 'cancer mom'— here's what I want other parents to know

Cancer comes in like a tornado and tears your life apart, and very few people understand that. They tell you to be strong or worse—to stay positive. Some friends show up in the most beautiful way and others stop showing up altogether. The losses hit hard.

By Danielle Moss • July 14, 2022

Realities and metaphors



Science

Current Issue First release papers Archive About Submit manuscri

HOME > SCIENCE > VOL. 372, NO. 6543 > RADIATION-RELATED GENOMIC PROFILE OF PAPILLARY THYROID CARCINOMA AFTER THE CHERNOBYL ACCIDENT

RESEARCH ARTICLE

Radiation-related genomic profile of papillary thyroid carcinoma after the Chernobyl accident

LINDSAY M. MORTON, DANIELLE M. KARYADI, CHIP STEWART, TETIANA I. BOGDANOVA, ERIC T. DAWSON, MIA K. STEINBERG, JIEQIONG DAI, STEPHEN W. HARTLEY, SARA J. SCHONFELD, [...] STEPHEN J. CHANOCK +30 authors Authors Info & Affiliations

SCIENCE • 22 Apr 2021 • Vol 372, Issue 6543 • DOI: 10.1126/science.abg2538

1,783 7 CHECK ACCESS

Genomics of radiation-induced damage

The potential adverse effects of exposures to radioactivity from nuclear accidents can include acute consequences such as radiation sickness, as well as long-term sequelae such as increased risk of cancer. There have been a few studies examining transgenerational risks of radiation exposure but the results have been inconclusive. Morton *et al.* analyzed papillary thyroid tumors, normal thyroid tissue, and blood from hundreds of survivors of the Chernobyl nuclear accident and compared

Realities and metaphors

Science

Current Issue | First release papers | Archive | About | Submit manuscr

HOME > SCIENCE > VOL. 372, NO. 6543 > RADIATION-RELATED GENOMIC PROFILE OF PAPILLARY THYROID CARCINOMA AFTER THE CHERNOBYL ACCIDENT

RESEARCH ARTICLE

Radiation-related gen
carcinoma after the C

LINDSAY M. MORTON, DANIELLE M. KARYADI, CHIP STEW...
STEPHEN W. HARTLEY, SARA J. SCHONFELD, [...] STEPHEN...

SCIENCE · 22 Apr 2021 · Vol 372, Issue 6543 · DOI: 10.1126/sc...

1,783 7

Genomics of radiation-ind

The potential adverse effects of expo
can include acute consequences such
sequelae such as increased risk of ca
transgenerational risks of radiation e
sive. Morton *et al.* analyzed papillary
blood from hundreds of survivors of

RARE DISEASES : TYPES

Children of Chernobyl: Birth Defects, Deformities, Ailments

Understanding short and long-term health issues caused by the nuclear meltdown

By Mary Kugler, RN | Updated on July 15, 2022

Print

Fact checked by James Lacy

Table of Contents

- 1986 Nuclear Accident
- Immediate Health Consequences
- Long-term Health Consequences
- What the Future Holds

In 1986, a meltdown at the Chernobyl nuclear power plant in what was then the Soviet Union showered radioactive particles all over Ukraine and neighboring countries. It resulted in the largest uncontrolled release of radiation (iodine-131, cesium-134 and cesium-137) in history.[1]

More than 6,000 cases of thyroid cancer linked to radiation exposure in people who were under age 18 at the time were reported between 1991 and 2015.[2] They came to be known as the Children of Chernobyl, and both researchers and the public have remained interested in their challenges.

This article discusses them and their health impacts in adulthood. It looks at Children of Chernobyl birth defects and deformities, as well as genetic mutations—and some of the more positive outcomes despite the early dire predictions.

Realities and metaphors

Science

Current Issue | First release papers | Archive | About | Submit manuscr

HOME > SCIENCE > VOL. 372, NO. 6543 > RADIATION-RELATED GENOMIC PROFILE OF PAPILLARY THYROID CARCINOMA AFTER THE CHERNOBYL ACCIDENT

RESEARCH ARTICLE

Radiation-related gen
carcinoma after the C

LINDSAY M. MORTON, DANIELLE M. KARYADI, CHIP STEW
STEPHEN W. HARTLEY, SARA J. SCHONFELD, [...] STEPHEN

SCIENCE • 22 Apr 2021 • Vol 372, Issue 6543 • DOI: 10.1126/sc

1,783 7

Genomics of radiation-ind

The potential adverse effects of expo
can include acute consequences such
sequelae such as increased risk of ca
transgenerational risks of radiation e
sive. Morton *et al.* analyzed papillary
blood from hundreds of survivors of

RARE DISEASES : TYPES

Children of Chernobyl: Birth Defects, Deformities, Ailments

Understanding short and long-term health issues caused by the nuclear meltdown

By Mary Kugler, RN | Updated on July 15, 2022
Fact checked by James Lacy

Table of Contents

1986 Nuclear Accident
Immediate Health Consequences
Long-term Health Consequences
What the Future Holds

In 198
then t
neigh
radiat

More
people
2015.
resear

This a
at Chi
mutat
predic

JAMA Network

JAMA Pediatrics | Search All | Enter Search Term

Review

FREE

March 2017

Characterizing the Pattern of Anomalies in Congenital Zika Syndrome for Pediatric Clinicians

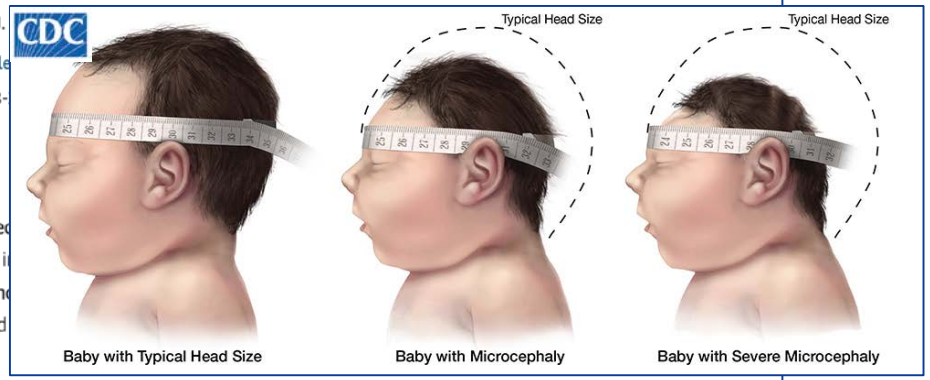
Cynthia A. Moore, MD, PhD[1]; J.

Author Affiliations | Article

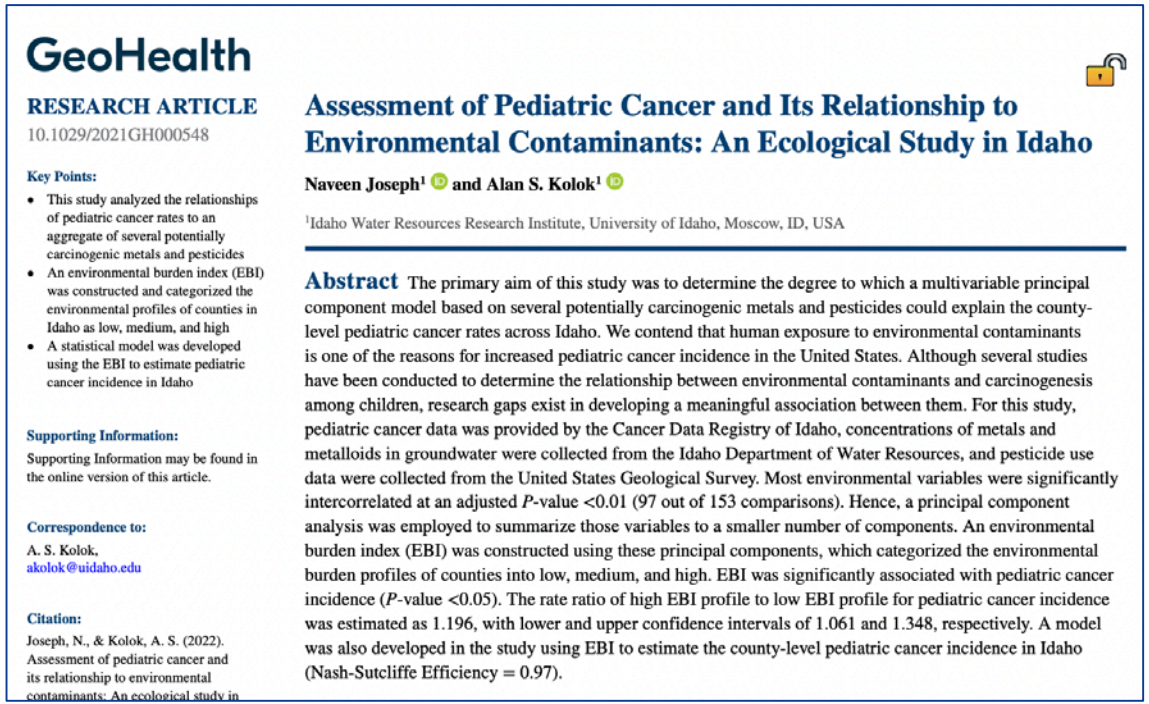
JAMA Pediatr. 2017;171(3):288-

Abstract

Importance Zika virus infec
evidence to conclude that i
but the full spectrum of an
evaluate and treat affected
genital Zika syndrome.



Realities and metaphors



GeoHealth

RESEARCH ARTICLE
10.1029/2021GH000548

Key Points:

- This study analyzed the relationships of pediatric cancer rates to an aggregate of several potentially carcinogenic metals and pesticides
- An environmental burden index (EBI) was constructed and categorized the environmental profiles of counties in Idaho as low, medium, and high
- A statistical model was developed using the EBI to estimate pediatric cancer incidence in Idaho

Supporting Information:

Supporting Information may be found in the online version of this article.

Correspondence to:

A. S. Kolok,
akolok@uidaho.edu

Citation:

Joseph, N., & Kolok, A. S. (2022). Assessment of pediatric cancer and its relationship to environmental contaminants: An ecological study in

Assessment of Pediatric Cancer and Its Relationship to Environmental Contaminants: An Ecological Study in Idaho

Naveen Joseph[1] and **Alan S. Kolok**[1]

[1]Idaho Water Resources Research Institute, University of Idaho, Moscow, ID, USA

Abstract The primary aim of this study was to determine the degree to which a multivariable principal component model based on several potentially carcinogenic metals and pesticides could explain the county-level pediatric cancer rates across Idaho. We contend that human exposure to environmental contaminants is one of the reasons for increased pediatric cancer incidence in the United States. Although several studies have been conducted to determine the relationship between environmental contaminants and carcinogenesis among children, research gaps exist in developing a meaningful association between them. For this study, pediatric cancer data was provided by the Cancer Data Registry of Idaho, concentrations of metals and metalloids in groundwater were collected from the Idaho Department of Water Resources, and pesticide use data were collected from the United States Geological Survey. Most environmental variables were significantly intercorrelated at an adjusted *P*-value <0.01 (97 out of 153 comparisons). Hence, a principal component analysis was employed to summarize those variables to a smaller number of components. An environmental burden index (EBI) was constructed using these principal components, which categorized the environmental burden profiles of counties into low, medium, and high. EBI was significantly associated with pediatric cancer incidence (*P*-value <0.05). The rate ratio of high EBI profile to low EBI profile for pediatric cancer incidence was estimated as 1.196, with lower and upper confidence intervals of 1.061 and 1.348, respectively. A model was also developed in the study using EBI to estimate the county-level pediatric cancer incidence in Idaho (Nash-Sutcliffe Efficiency = 0.97).

Realities and metaphors

GeoHealth

RESEARCH ARTICLE

10.1029/2021GH000548

Key Points:

- This study analyzed the relationships of pediatric cancer rates to an aggregate of several potentially carcinogenic metals and pesticides
- An environmental burden index (EBI) was constructed and categorized the environmental profiles of counties in Idaho as low, medium, and high
- A statistical model was developed using the EBI to estimate pediatric cancer incidence in Idaho

Supporting Information:

Supporting Information may be found in the online version of this article.

Correspondence to:

A. S. Kolok,
akolok@uidaho.edu

Citation:

Joseph, N., & Kolok, A. S. (2022). Assessment of pediatric cancer and its relationship to environmental contaminants: An ecological study in

Assessment of Pediatric Cancer and Its Relationship to Environmental Contaminants: An Ecological Study in Idaho

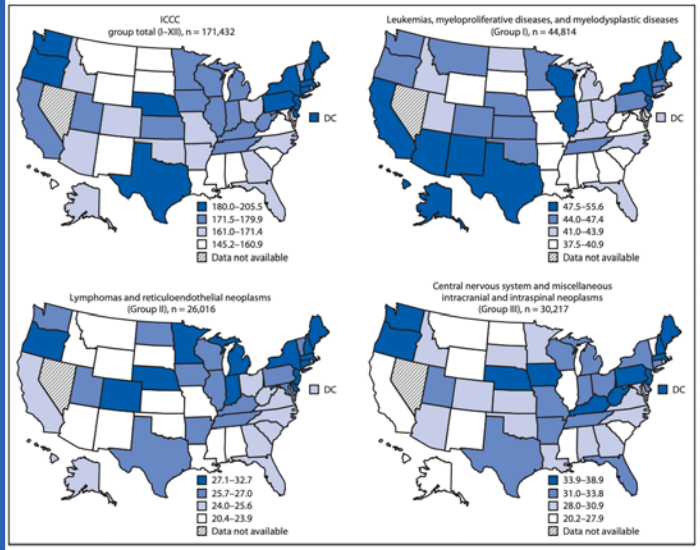
Naveen Joseph[1] and Alan S. Kolok[1]

[1]Idaho Water Resources Research Ins

Abstract The primary aim o
component model based on sever
level pediatric cancer rates acros
is one of the reasons for increase
have been conducted to determin
among children, research gaps ex
pediatric cancer data was provid
metalloids in groundwater were
data were collected from the Uni
intercorrelated at an adjusted *P*-v
analysis was employed to summa
burden index (EBI) was construc
burden profiles of counties into l
incidence (*P*-value <0.05). The r
was estimated as 1.196, with low
was also developed in the study u
(Nash-Sutcliffe Efficiency = 0.9

Geographic Variation in Pediatric Cancer Incidence — United States, 2003–2014

David A. Siegel, MD[1,2]; Jun Li, MD, PhD[2]; S. Jane Henley, MSPH[2]; Reda J. Wilson, MPH[2]; Natasha Buchanan Lunsford, PhD[2]; Eric Tai, MD[2]; Elizabeth A. Van Dyne, MD[1,2]



See figure footnotes on next page.

Realities and metaphors

GeoHealth

RESEARCH ARTICLE
10.1029/2021GH000548

Assessment of Pediatric Cancer and Its Relationship to Environmental Contaminants: An Ecological Study in Idaho

Naveen Joseph[1] and Alan S. Kolok[1]

[1]Idaho Water Resources Research Ins

Key Points:

- This study analyzed the relationships of pediatric cancer rates to an aggregate of several potentially carcinogenic metals and pesticides
- An environmental burden index (EBI) was constructed and categorized the environmental profiles of counties in Idaho as low, medium, and high
- A statistical model was developed using the EBI to estimate pediatric cancer incidence in Idaho

Supporting Information:

Supporting Information may be found in the online version of this article.

Correspondence to:

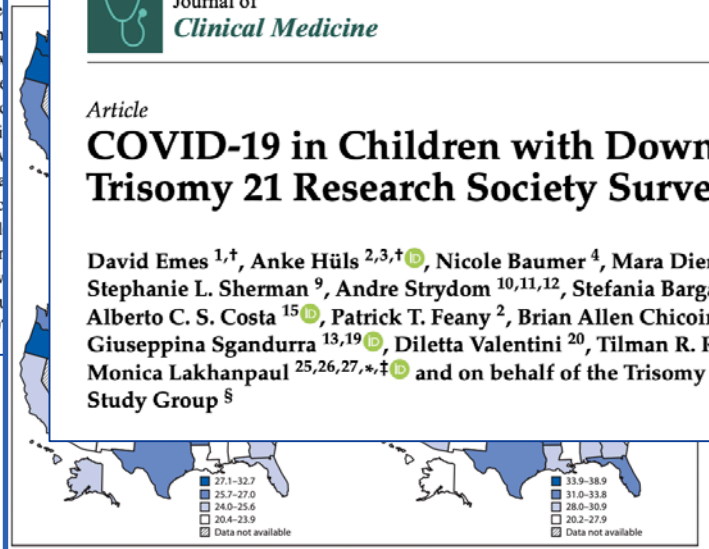
A. S. Kolok,
akolok@uidaho.edu

Citation:

Joseph, N., & Kolok, A. S. (2022). Assessment of pediatric cancer and its relationship to environmental contaminants: An ecological study in

Abstract The primary aim o
component model based on sever
level pediatric cancer rates acros
is one of the reasons for increase
have been conducted to determin
among children, research gaps ex
pediatric cancer data was provid
metalloids in groundwater were
data were collected from the Uni
intercorrelated at an adjusted *P*-v
analysis was employed to summa
burden index (EBI) was construc
burden profiles of counties into l
incidence (*P*-value <0.05). The r
was estimated as 1.196, with low
was also developed in the study u
(Nash-Sutcliffe Efficiency = 0.9

Geographic Variation in Pediatric Cancer Incidence —



See figure footnotes on next page.

Journal of *Clinical Medicine*

MDPI

Article

COVID-19 in Children with Down Syndrome: Data from the Trisomy 21 Research Society Survey

David Emes [1,†], Anke Hüls [2,3,†], Nicole Baumer [4], Mara Dierssen [5,6,7], Shiela Puri [8], Lauren Russell [2], Stephanie L. Sherman [9], Andre Strydom [10,11,12], Stefania Bargagna [13], Ana Cláudia Brandão [14], Alberto C. S. Costa [15], Patrick T. Feany [2], Brian Allen Chicoine [16], Sujay Ghosh [17], Anne-Sophie Rebillat [18], Giuseppina Sgandurra [13,19], Diletta Valentini [20], Tilman R. Rohrer [21], Johannes Levin [22,23,24], Monica Lakhanpaul [25,26,27,*,‡] and on behalf of the Trisomy 21 Research Society COVID-19 Initiative Study Group [§]

Realities and metaphors

GeoHealth

RESEARCH ARTICLE
10.1029/2021GH000548

Assessment of Pediatric Cancer and Its Relationship to Environmental Contaminants: An Ecological Study in Idaho

Naveen Joseph[1] and Alan S. Kolok[1]

[1]Idaho Water Resources Research Ins

Key Points:

- This study analyzed the relationships of pediatric cancer rates to an aggregate of several potentially carcinogenic metals and pesticides
- An environmental burden index (EBI) was constructed and categorized the environmental profiles of counties in Idaho as low, medium, and high
- A statistical model was developed using the EBI to estimate pediatric cancer incidence in Idaho

Supporting Information:

Supporting Information may be found in the online version of this article.

Correspondence to:

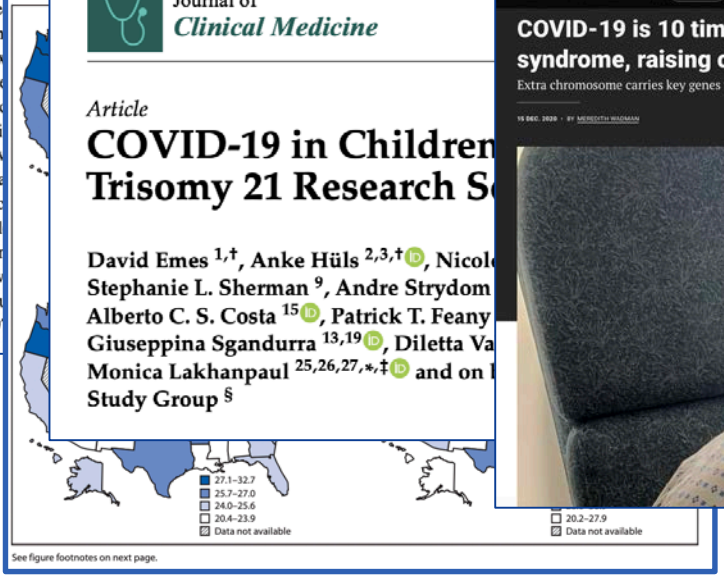
A. S. Kolok,
akolok@uidaho.edu

Citation:

Joseph, N., & Kolok, A. S. (2022). Assessment of pediatric cancer and its relationship to environmental contaminants: An ecological study in

Abstract The primary aim o
component model based on sever
level pediatric cancer rates acros
is one of the reasons for increase
have been conducted to determin
among children, research gaps ex
pediatric cancer data was provid
metalloids in groundwater were
data were collected from the Uni
intercorrelated at an adjusted *P*-v
analysis was employed to summa
burden index (EBI) was construc
burden profiles of counties into l
incidence (*P*-value <0.05). The r
was estimated as 1.196, with low
was also developed in the study u
(Nash-Sutcliffe Efficiency = 0.9

Geographic Variation in Pediatric Cancer Incidence —



Journal of
Clinical Medicine

Article

**COVID-19 in Children
Trisomy 21 Research S**

David Emes [1,†], Anke Hüls [2,3,†], Nicol
Stephanie L. Sherman [9], Andre Strydom
Alberto C. S. Costa [15], Patrick T. Feany
Giuseppina Sgandurra [13,19], Diletta Va
Monica Lakhanpaul [25,26,27,*,‡] and on
Study Group §

COMMENTARY JOURNALS COVID-19 Science LOG IN

COVID-19 is 10 times deadlier for people with Down syndrome, raising calls for early vaccination

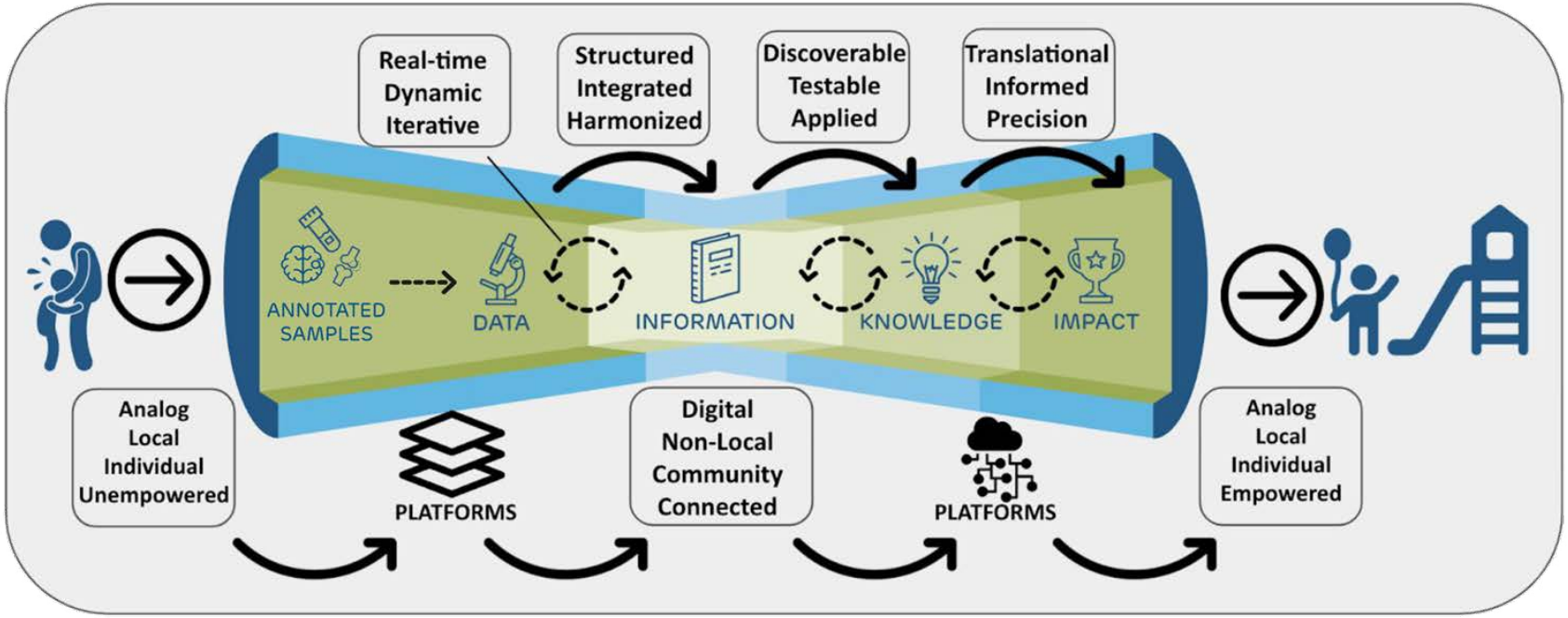
Extra chromosome carries key genes that may work against them

15 DEC. 2020 · BY MEREDITH WADMAN

An abstraction for shared approaches for pediatric disaster research

Multi-sourced, Multi-modal, Multi-omic, Multi-disease, Multi-User, Asynchronous Data

Gabriella Miller Kids First Data Resource Center

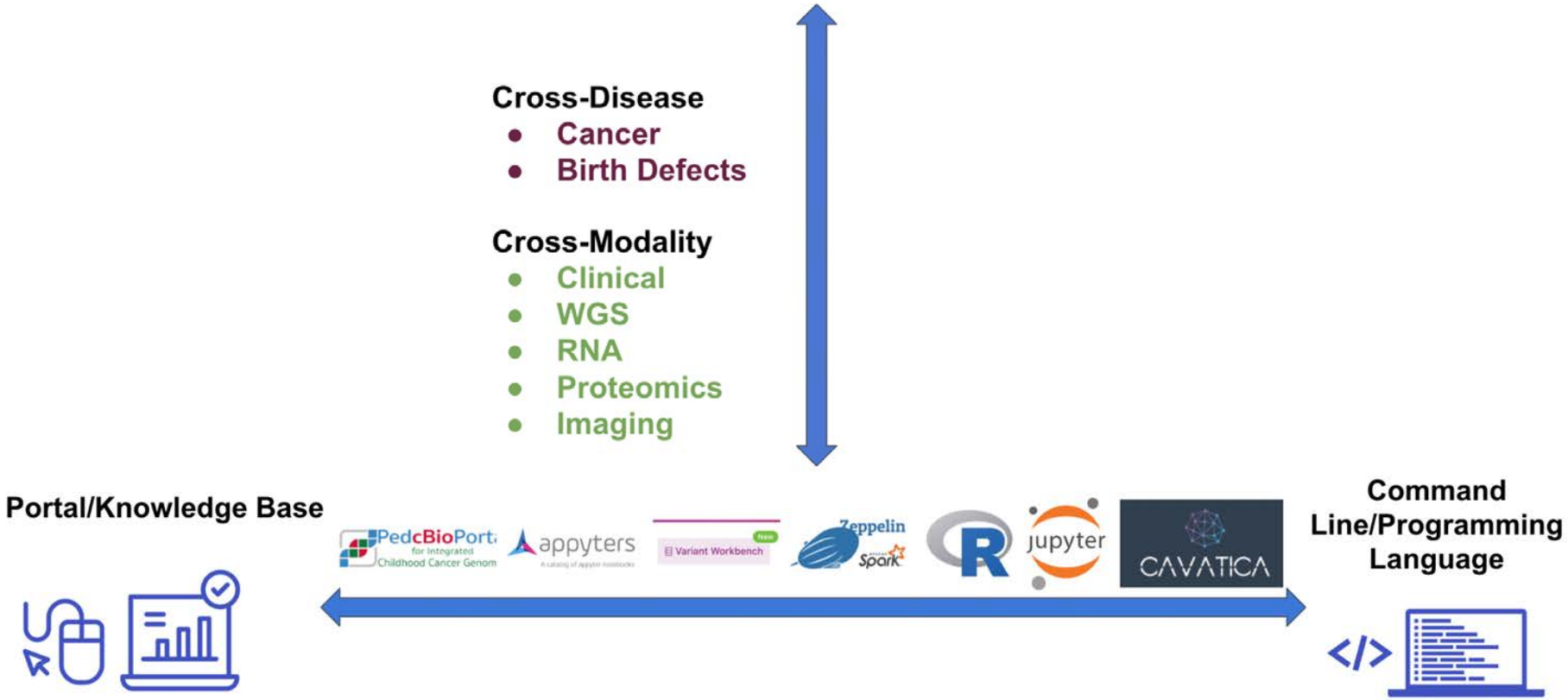


Platforms as a Service supporting FAIR Ecosystems

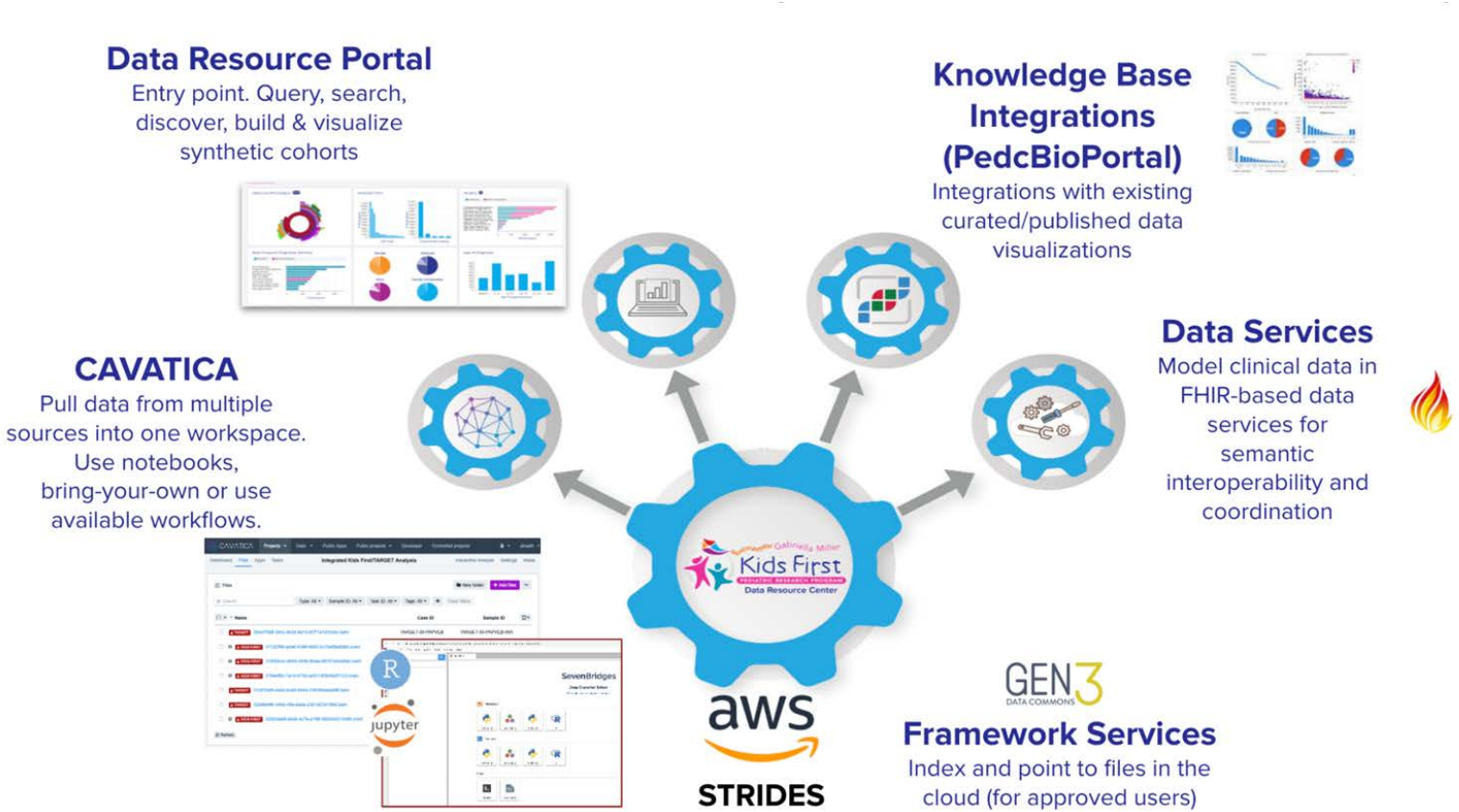
Rigor through community use:

- **Source**
- **Metadata**
- **Derived**
- **Workflows/Tools**

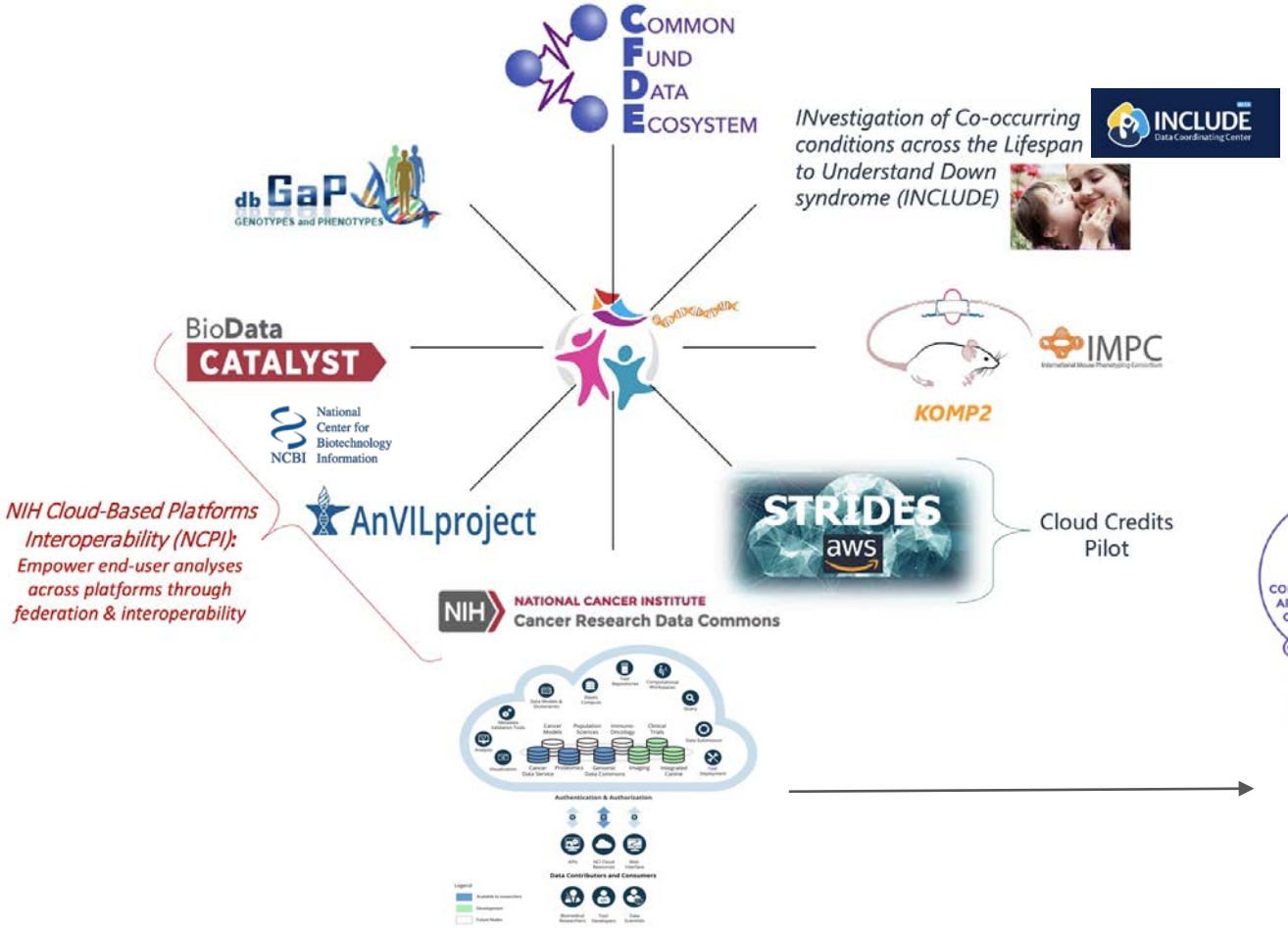
Big Data for Different Users (and their collaborations)

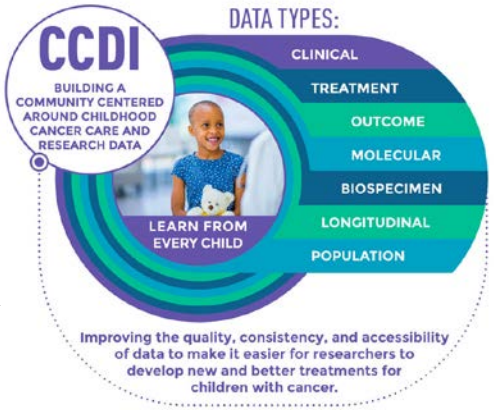


Platforms as a Service supporting FAIR Ecosystems



Cross-Platform Interoperability and Governance



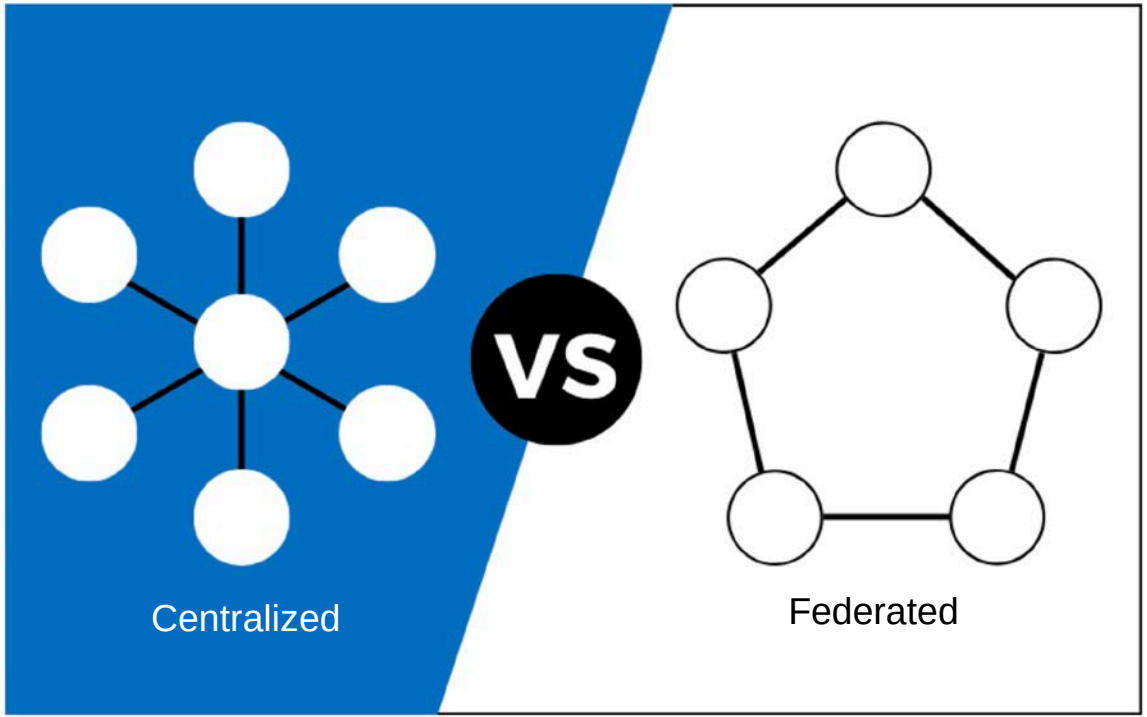


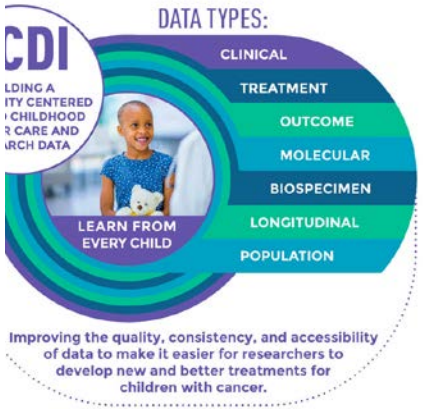
Cross-Platform Interoperability and Governance

COMMON FUND DATA ECOSYSTEM

INvestigation of Co-occurring INCLUDE

NIH Cloud-Based Pla
Interoperability (N
Empower end-user an
across platforms thr
federation & interope

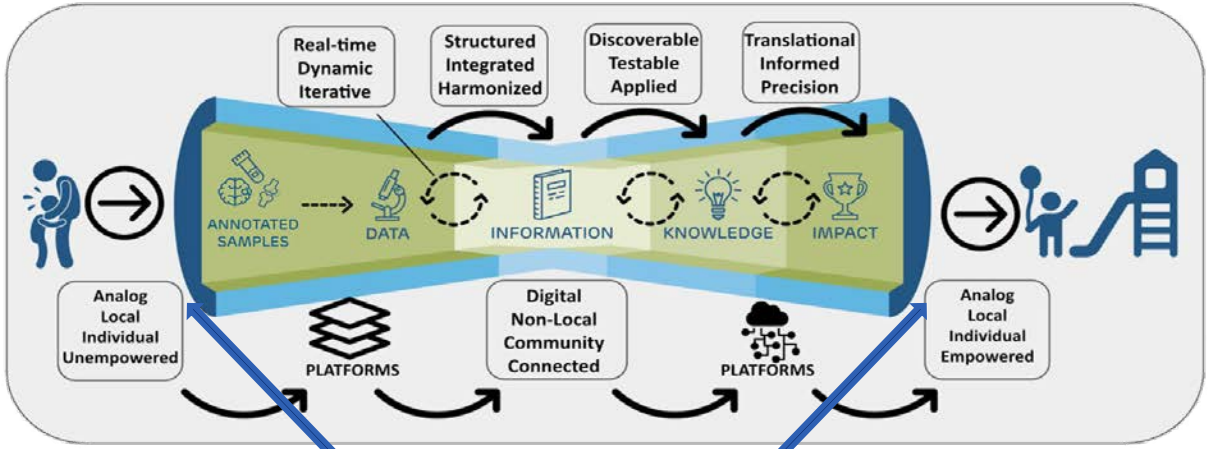


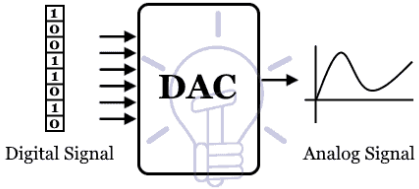


Final Thoughts

Lessons and Opportunities

The Analog to Digital Transition is the hardest part . . .





Intersection across:

1) State/federal data registries and repositories

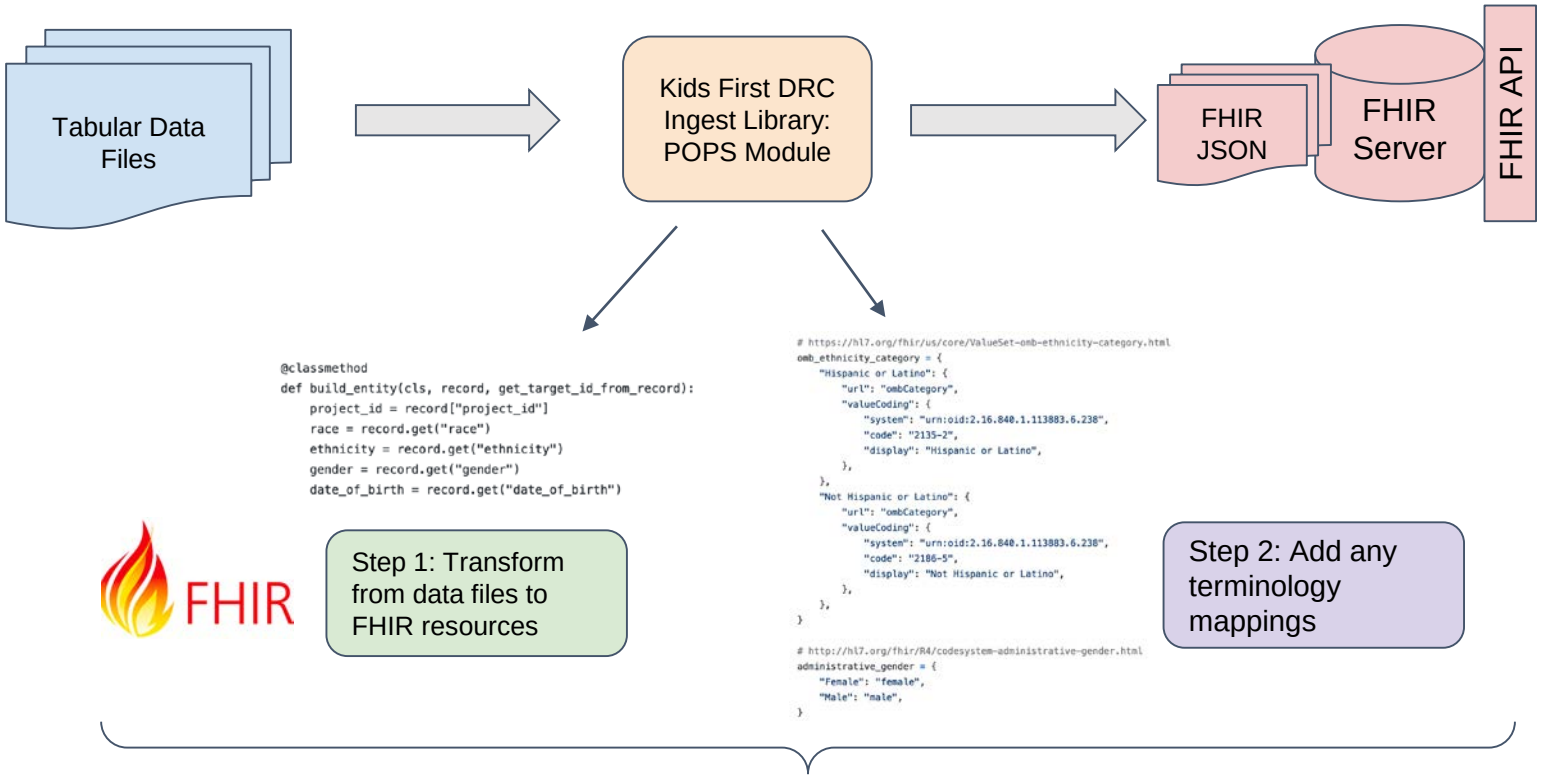
&

2) Healthcare Learning Systems

&

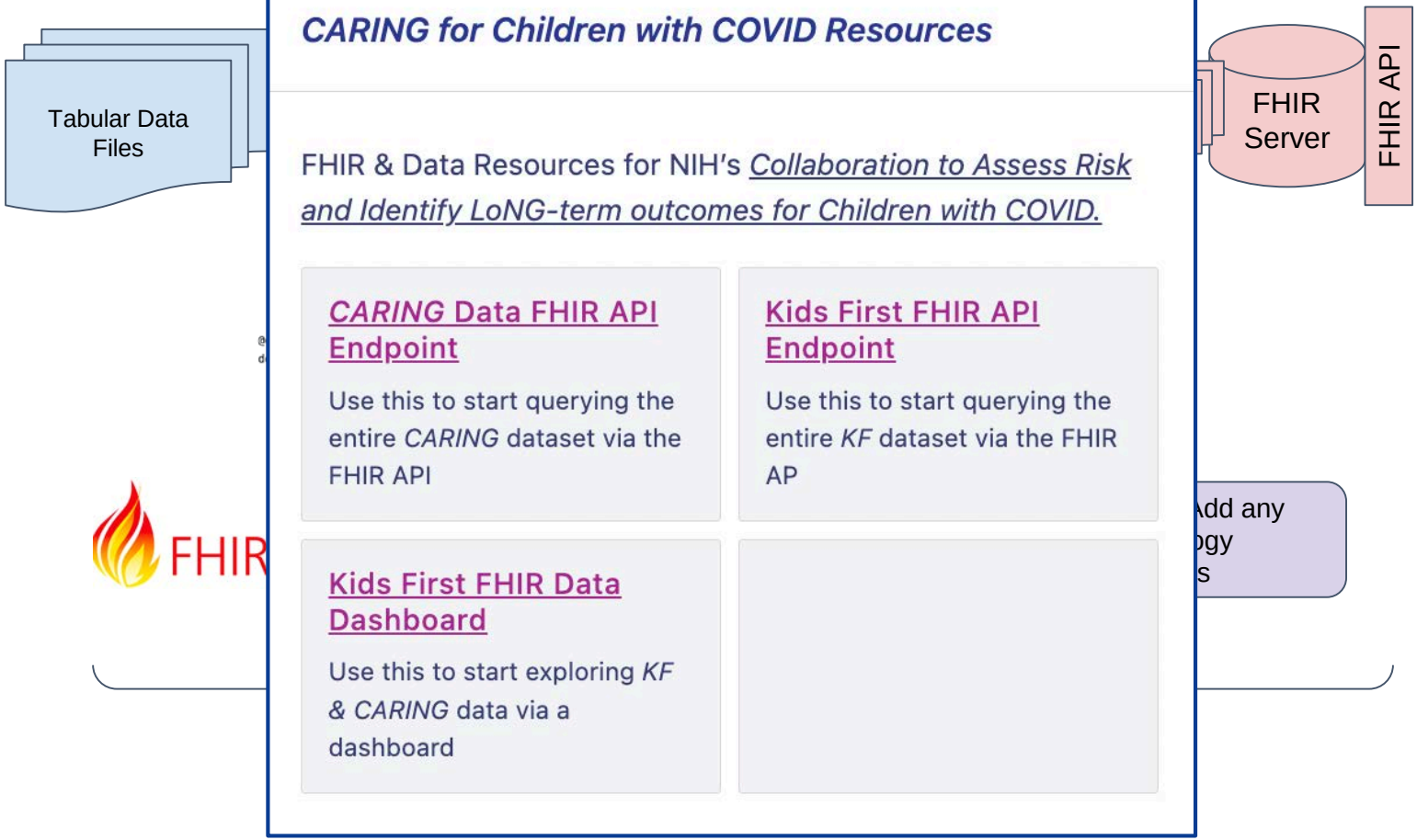
3) “Protocolized” academic research frameworks

Leveraging Community Standards as Paired with Tools

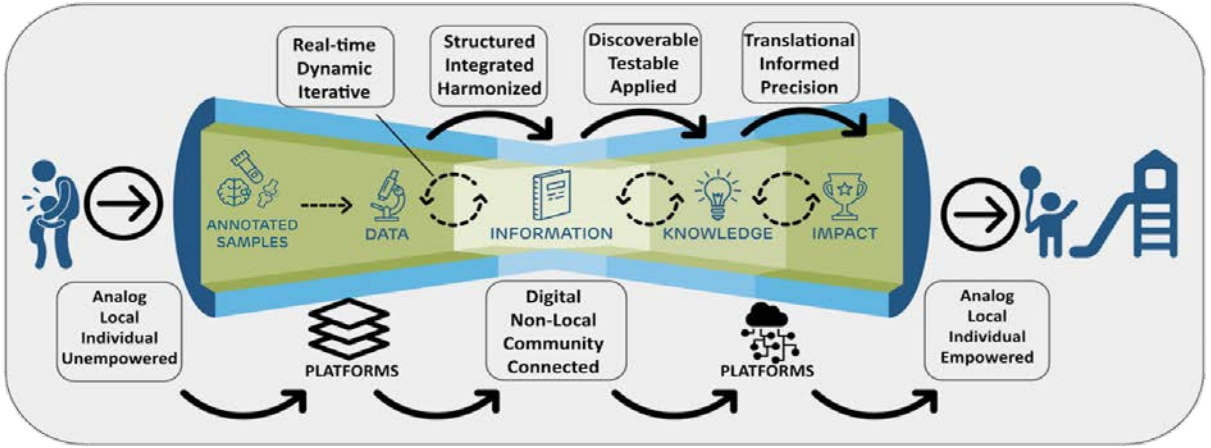


Code once, then automated as new data arrives:
From data deposition to data “streams”

Leveraging Community Standards as Paired with Tools



Disaster Research Infrustructure Opportunity



$$a = \frac{\Delta v}{\Delta t}$$

Leverage existing frameworks while **optimizing the ecosystem** around *ACCELERATED ACTIONABILITY* as the variable of impact is a shared need across all translational research, on behalf of all children