Iowa Registry for Congenital and Inherited Disorders

Paul A Romitti, PhD

Professor, Department of Epidemiology, Department of Biostatistics, and the Interdisciplinary Program in Toxicology, University of Iowa

Director, Iowa Registry for Congenital and Inherited Disorders (IRCID)

Director, Reproductive Molecular Epidemiology Research and Education Program (RMEREP)

Director, Iowa Center for Birth Defects Research and Prevention (CBDRP)

Iowa Registry for Congenital and Inherited Disorders

IDPH Iowa Department of Public Health

The University of Iowa

Legislation – Chapter 136A

- **136A.2 DEFINITIONS**

- "Congenital disorder" means an abnormality existing prior to or at birth, including a stillbirth, that adversely affects the health and development of a fetus, newborn, child, or adult, including a ***structural malformation or a genetic, chromosomal, inherited, or biochemical disorder.***

Legislation – Chapter 136A

- **136A.3 ESTABLISHMENT OF CENTER FOR CONGENITAL AND INHERITED DISORDERS -- DUTIES**

- Perform ***surveillance and monitoring of congenital and inherited disorders*** to determine the occurrence and trends of the disorders, to conduct thorough and complete epidemiological surveys, to assist in the planning for and provision of services to children with congenital and inherited disorders and their families, and to identify environmental and genetic risk factors for congenital and inherited disorders.

Legislation – Chapter 136A

- **136A.6 CENTRAL REGISTRY**

- The center for congenital and inherited disorders shall ***maintain a central registry or shall establish an agreement with a designated contractor to maintain a central registry***, to compile, evaluate, retain, and disseminate information on the occurrence, prevalence, causes, treatment, and prevention of congenital disorders. Congenital disorders shall be considered reportable conditions in accordance with rules adopted by the department and shall be abstracted and maintained by the registry.

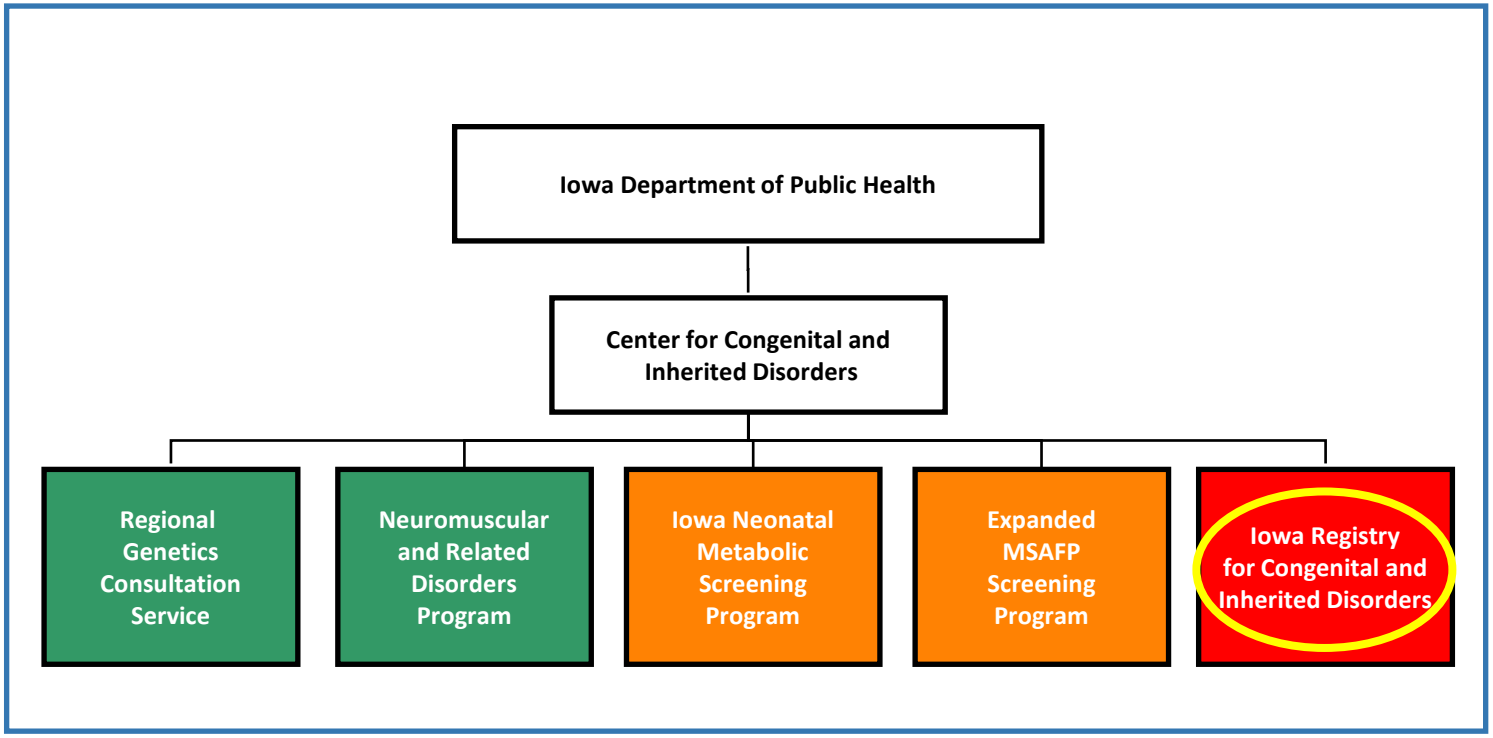
IRCID

- **Established in 1983 as Iowa Birth Defects Registry by:**
  - Iowa Department of Human Services
  - Iowa Department of Public Health (IDPH)
  - The University of Iowa (UI)

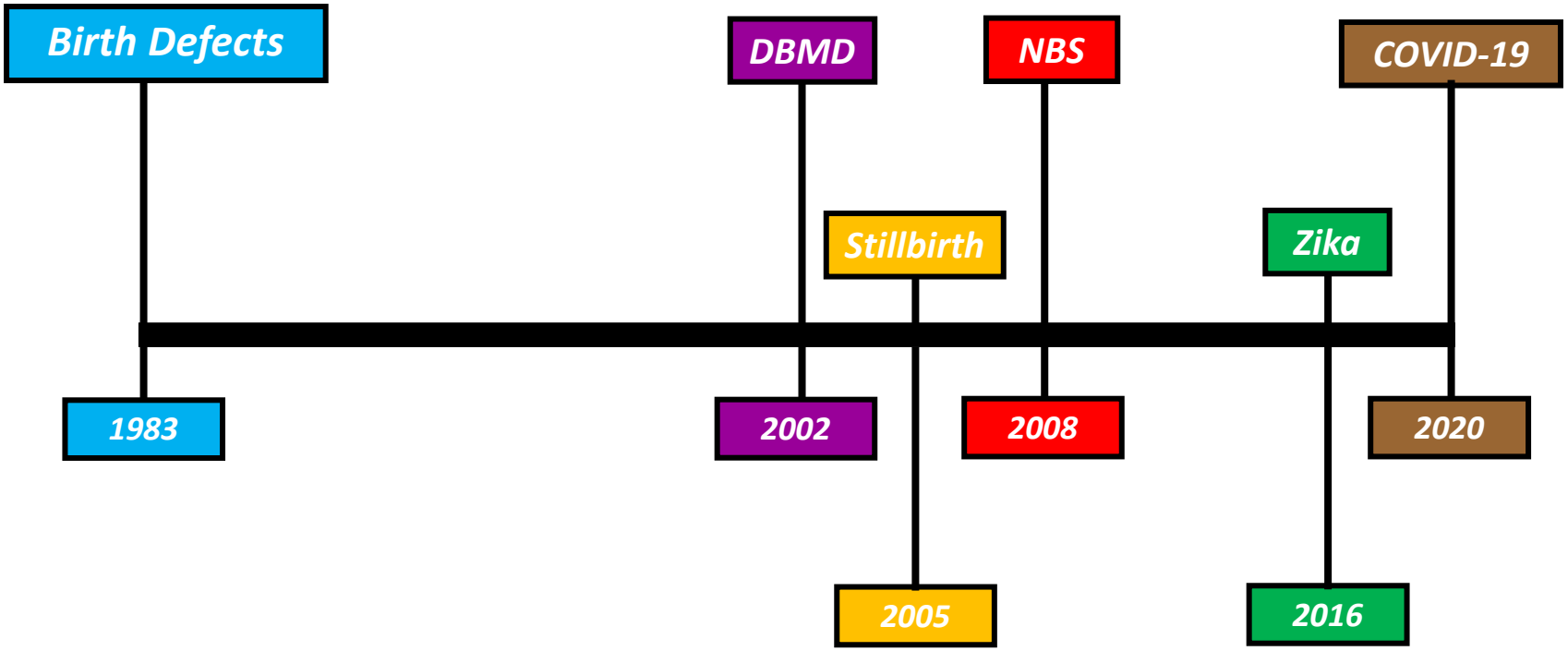
- **Instituted surveillance by change in state code that made birth defects reportable conditions in Iowa**

- **Program of IDPH whose operations are chartered to UI College of Public Health**

IRCID


Iowa Department of Public Health
Center for Congenital and Inherited Disorders
Regional Genetics Consultation Service
Neuromuscular and Related Disorders Program
Iowa Neonatal Metabolic Screening Program
Expanded MSAFP Screening Program
Iowa Registry for Congenital and Inherited Disorders

Surveillance Timeline

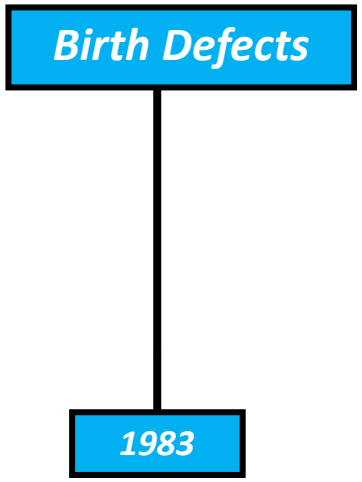


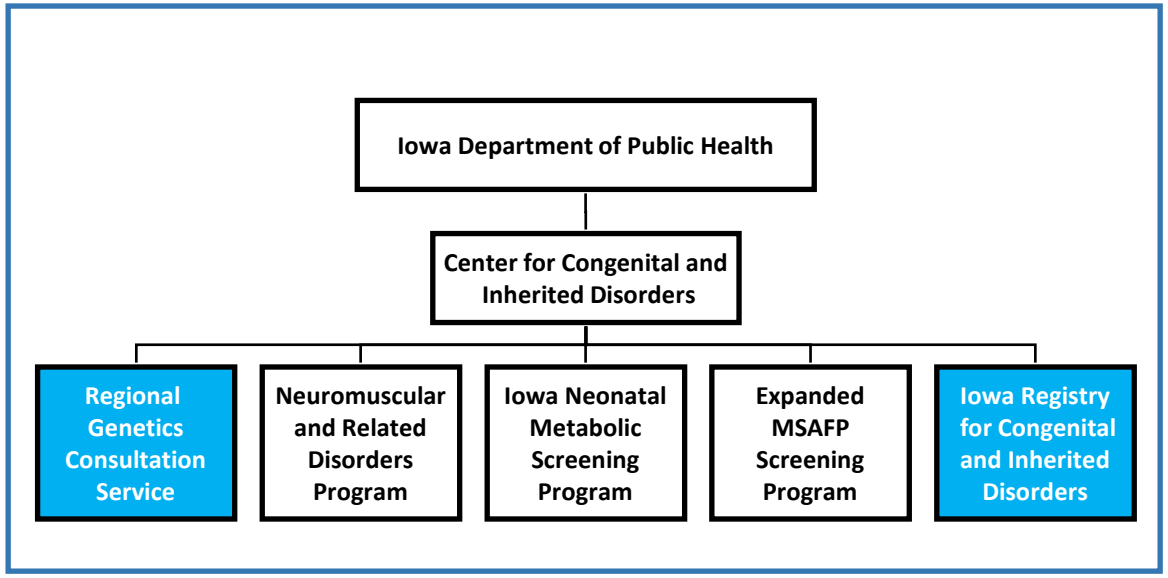
DBMD=Duchenne/Becker Muscular Dystrophy; NBS=Newborn Screening

Goals – Birth Defect Surveillance

- Maintain statewide *surveillance* and monitor trends in birth defect occurrence and mortality
- Provide data to conduct *research* studies to identify genetic and environmental risk factors for birth defects
- Promote *education* activities for the prevention of birth defects
- Provide *outreach* through referral of patients and their families to clinical, educational and social services

Birth Defect Surveillance





Birth Defect

- A **major birth defect** is an abnormality of **organ structure** or **function**, including metabolism, present at birth that results in physical disability, mental disability, or death
- A **minor birth defect** is an abnormality that results in **medically insignificant departure** from normal development

PKU

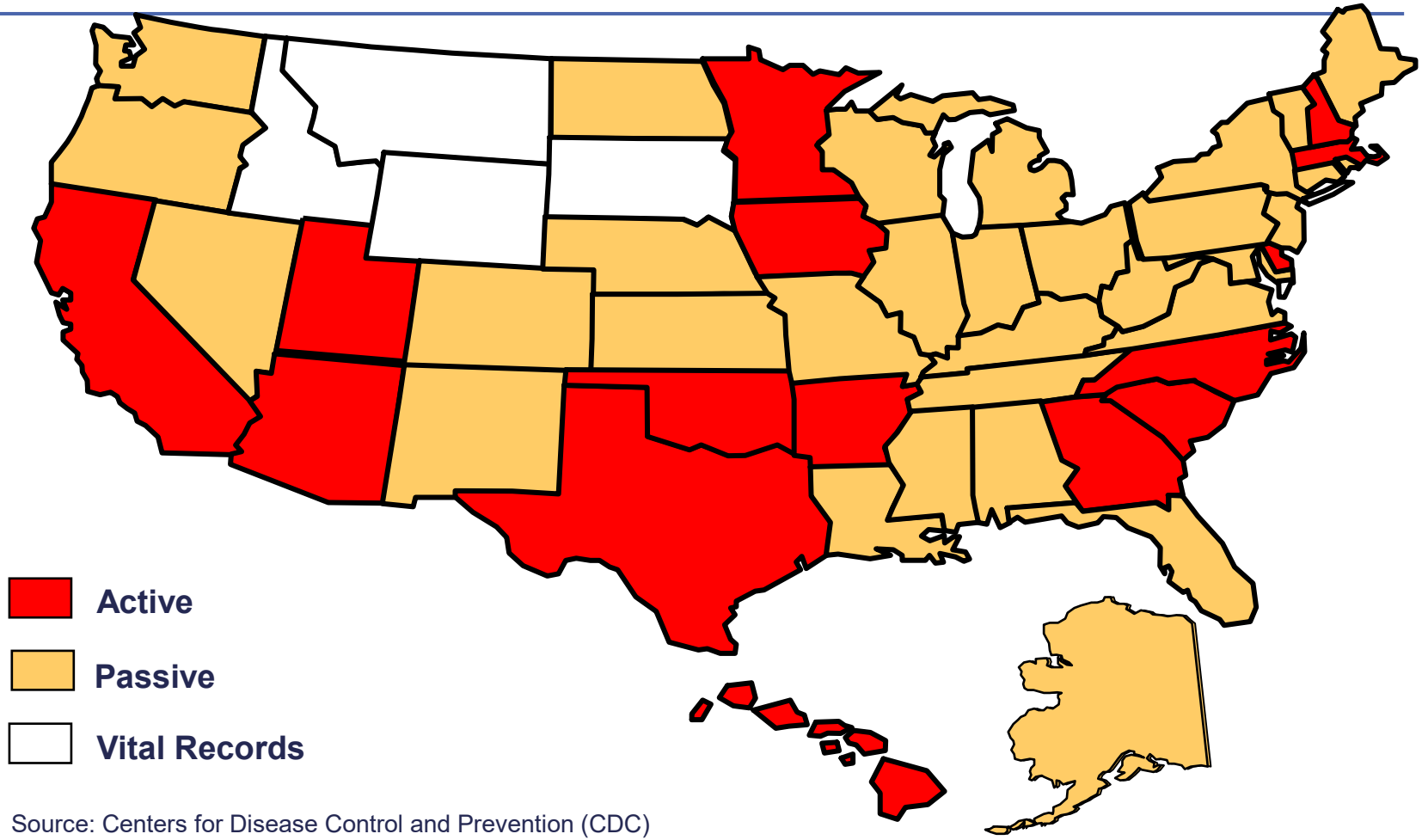
Eligible Case Child

- Mother is Iowa resident at time of delivery
- Delivery date January 1, 1983 – present
- Live birth, stillbirth, or elective termination with one or more reportable defects
- For live births, reportable diagnosis made within child's first year of life

Birth Defect Surveillance Approaches

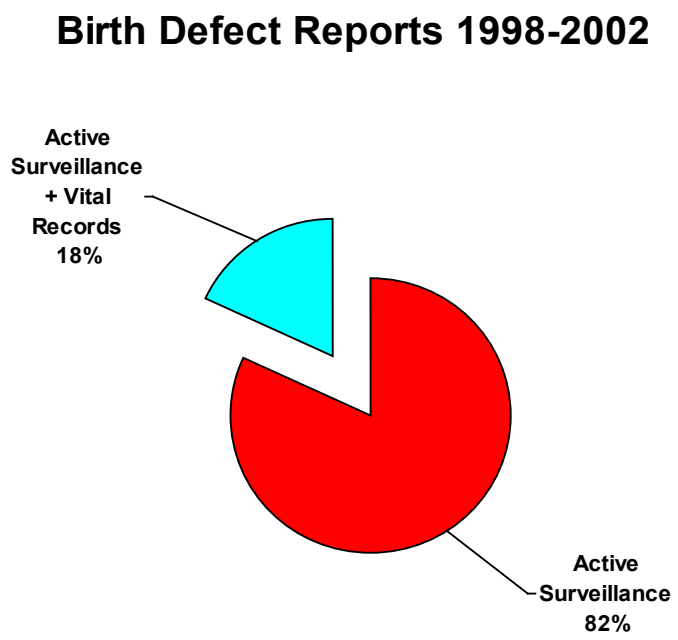
- **Vital Records (Poor)**
  - Use of birth and fetal death certificates provided by state department of health
- **Passive Surveillance (Fair-Good)**
  - Use of medical reports submitted by staff from hospitals, clinics, or other facilities
- **Active Surveillance (Best)**
  - Use of trained personnel who systematically review records in hospitals, clinics, and other facilities

BD Surveillance – United States



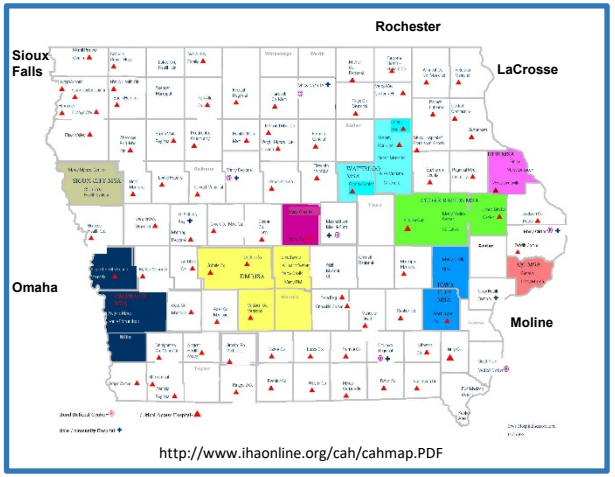
Source: Centers for Disease Control and Prevention (CDC)

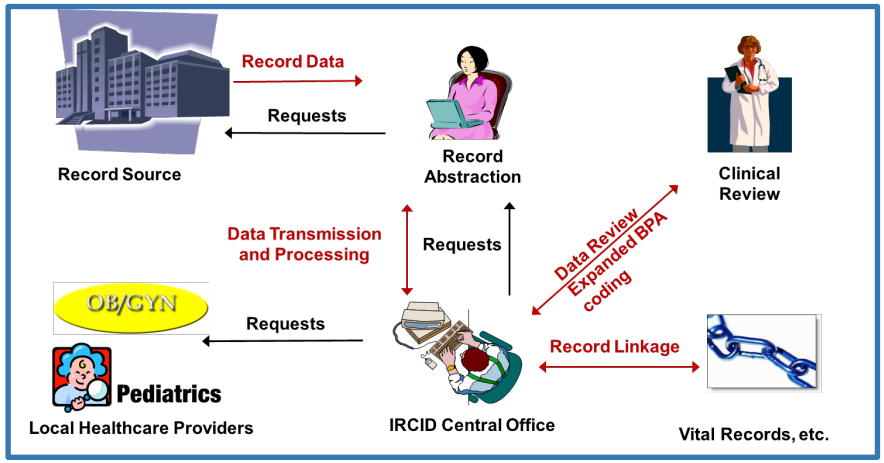
Iowa Birth Defects Ascertainment



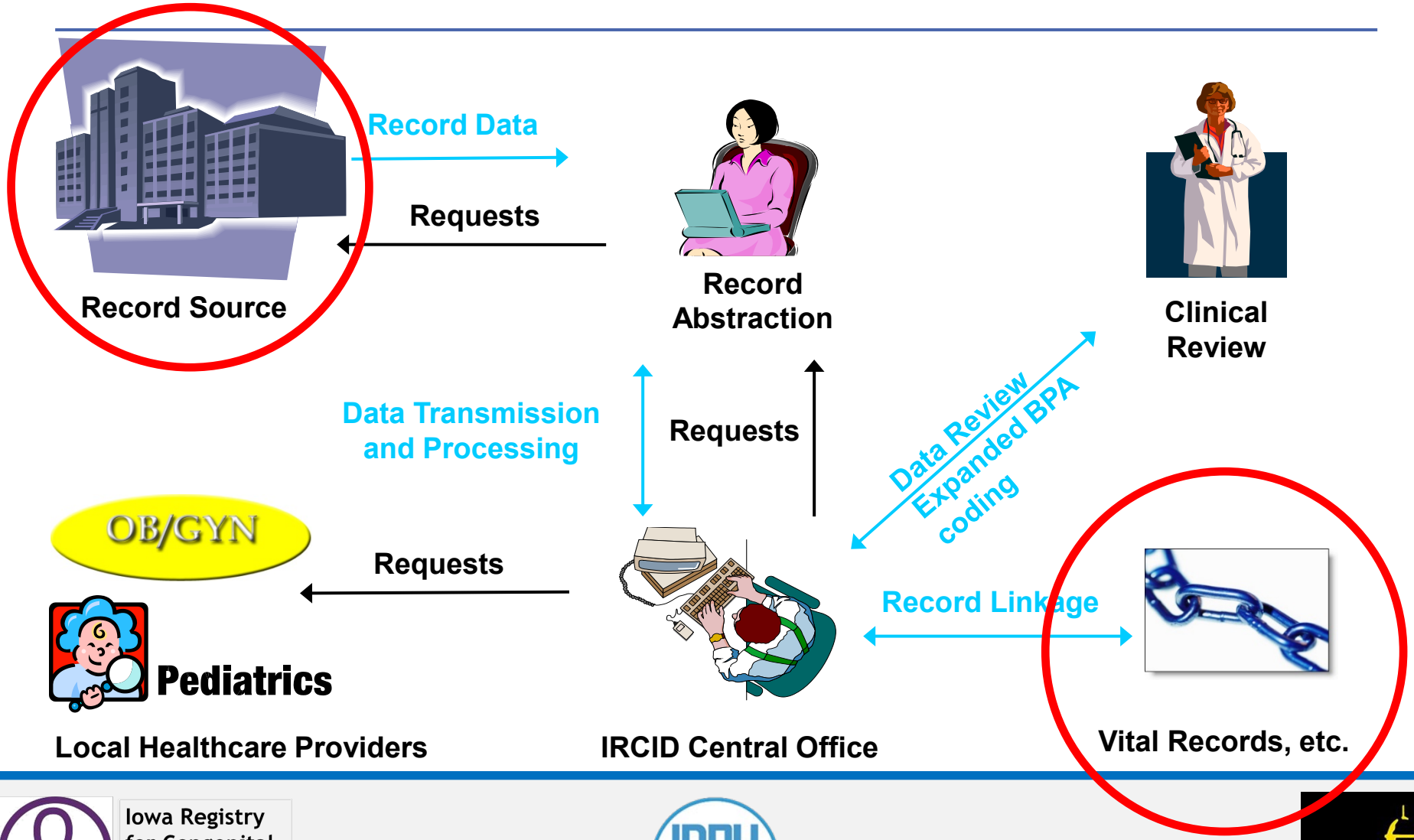
- A record review of 9,080 infants identified by the Registry's active surveillance system showed that vital records would have identified only 1,644 or 18% of these infants. Therefore, for these years, if the Registry relied on vital records only for birth defect surveillance, on average, four out of every five infants diagnosed with a birth defect would not have been identified.

Surveillance Methods





Birth Defect Surveillance



Regulations – Chapter 4 IAC

- **641—4.7(136A) Iowa registry for congenital and inherited disorders (IRCID)**

- **4.7(3) IRCID Activities**
  - c. The IRCID staff shall review ***hospital records, clinical charts, physician's records, vital records, prenatal records, and fetal death evaluation protocols*** pursuant to 641—1.3(139A), information from the INMSP, RGCS, NMP, and the IMPSP, and any other information that the IRCID deems necessary and appropriate for congenital and inherited disorders surveillance.

Case Finding Sources

- **Vital records**
  - Birth, death, fetal death certificates
- **Medical records**
  - Case lists generated by hospitals based on ICD-10-CM (diagnostic/billing) codes
- **Administrative databases**
  - Hospital discharge data
  - DMV
  - Voter registration

Birth Defect Surveillance

Record Source

Record Data

Requests

Record Abstraction

Clinical Review

Data Transmission and Processing

Requests

Data Review
Expanded BPA coding

Requests

Record Linkage

Local Healthcare Providers

IRCID Central Office

Vital Records, etc.

Surveillance Data – Birth Defect

- Diagnostic information
- Demographic data
- Prenatal history
- Risk factor information
- Cytogenetic and laboratory data
- Family history

Birth Defect Surveillance

Record Source

Record Data

Requests

Record Abstraction

Clinical Review

Data Transmission and Processing

Requests

Data Review
Expanded BPA coding

OB/GYN

Requests

Pediatrics

Local Healthcare Providers

IRCID Central Office

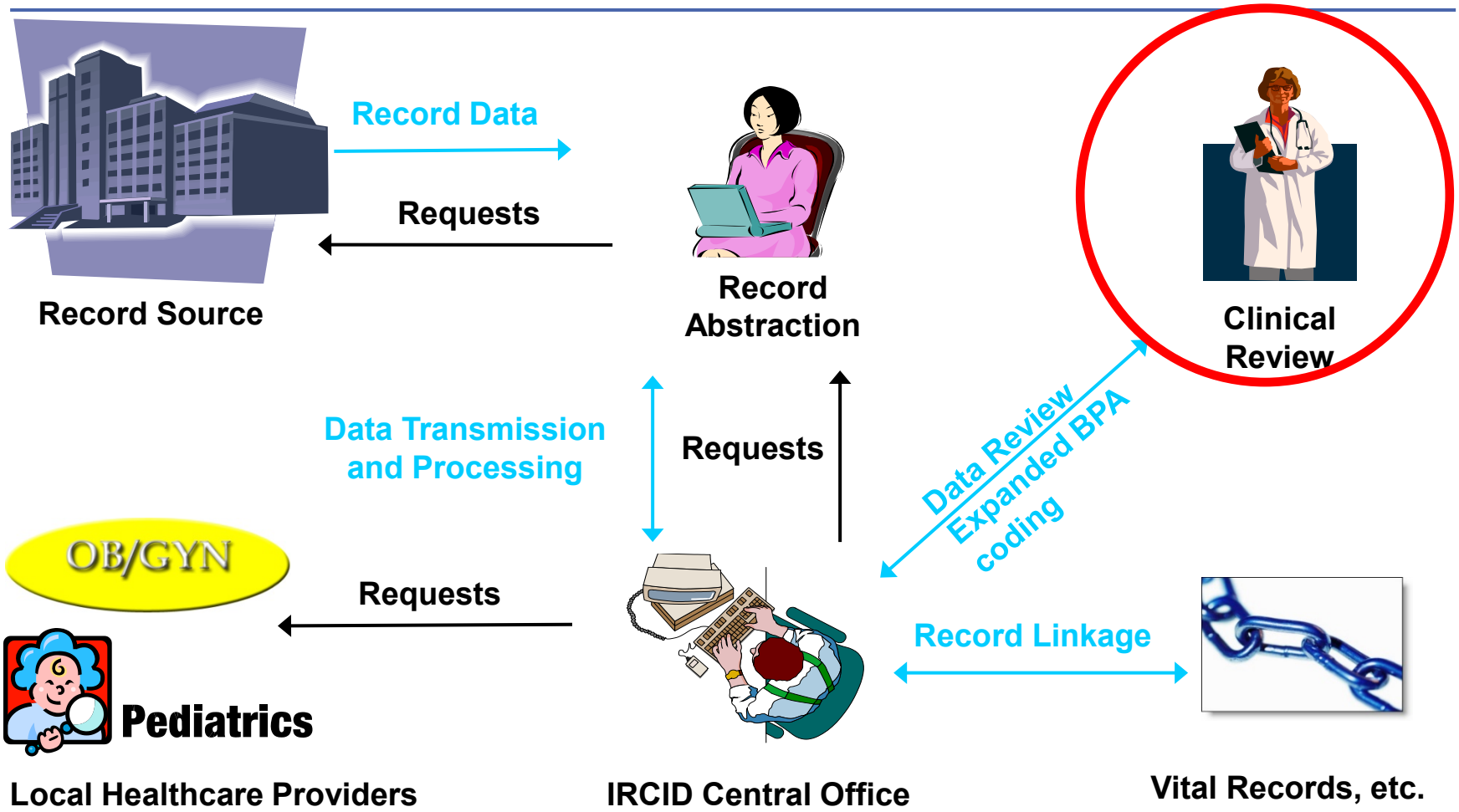
Record Linkage

Vital Records, etc.

Editing

- Ensure abstract is internally consistent
- Ensure diagnostic codes are correctly assigned and documented
- Reconcile information about one child across multiple hospitals (if applicable)

Birth Defect Surveillance



Clinical Case Review

- **Conducted by physician reviewers**
  - Pediatric Geneticist (all birth defects cases)
  - Pediatric Cardiologist (cardiac cases only)
  - Pediatric Neurologist (Zika-related conditions)

Birth Defect Surveillance

Record Source

Record Data

Requests

Record Abstraction

Clinical Review

Data Transmission and Processing

Requests

Data Review
Expanded BPA coding

OB/GYN

Requests

Pediatrics

Local Healthcare Providers

IRCID Central Office

Record Linkage

Vital Records, etc.

Data Quality

- Available data sources
- Timeliness vs. Completeness
- Quality Control and Quality Assurance

Surveillance Mission

Identify

Classify

IRCID Surveillance Data

Iowa Registry for Congenital and Inherited Disorders

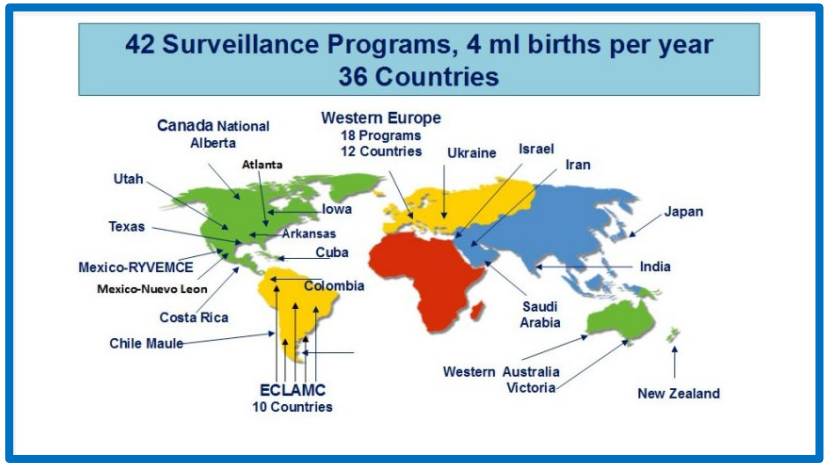
NBDPN

- **Created to establish and maintain a national network of state and population-based programs for birth defects surveillance and research**

- **Mission:**
  - Improve access and application of information about the prevalence and trends of birth defects
  - Increase collaboration between members within the birth defects community
  - Advance science through birth defects surveillance and its application to public health efforts and resource allocation

ICBDSR

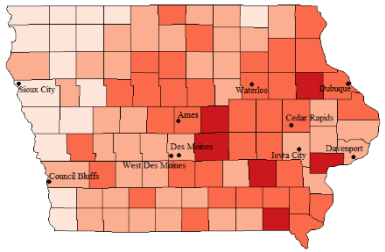
- International organization affiliated with WHO
- Brings together birth defect programs globally to conduct worldwide surveillance and research into the occurrence and possible causes of birth defects



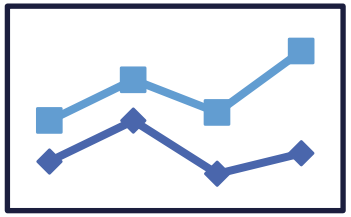
Public Health Surveillance

Person

Place



Time



Iowa Study

RESEARCH ARTICLE

AMERICAN JOURNAL OF medical genetics PART A

Epidemiology of Congenital Idiopathic Talipes Equinovarus in Iowa, 1997–2005

Vijaya Kancherla,[1] Paul A. Romitti,[1]* Kristin M. Caspers,[1] Soman Puzhankara,[1] and Jose A. Morcuende[2]

[1]Department of Epidemiology, College of Public Health, The University of Iowa, Iowa City, Iowa

[2]Department of Orthopedic Surgery, Carver College of Medicine, The University of Iowa, Iowa City, Iowa

Multistate Study

WILEY Birth Defects Research

ORIGINAL ARTICLE

Prevalence and descriptive epidemiology of infantile hypertrophic pyloric stenosis in the United States: A multistate, population-based retrospective study, 1999–2010

Renuka Kapoor[1] | Vijaya Kancherla[1] | Yanyan Cao[2] | Jacob Oleson[3] | Jonathan Suhl[2] | Mark A Canfield[4] | Charlotte M Druschel[5] | Russell S Kirby[6] | Robert E Meyer[7,8] | Paul A Romitti[2,3]

Surveillance Data

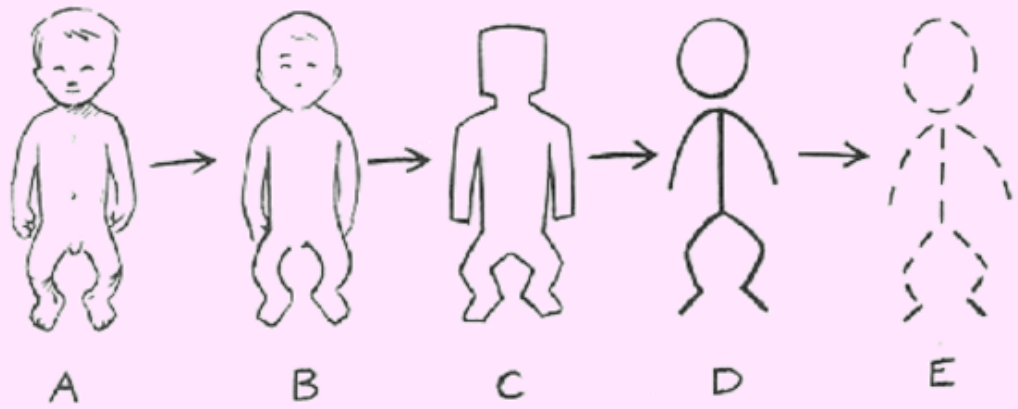


Diagram illustrating the distortion of the information in a reporting system, from the infant to the coded data. (A) shows the actual infant, (B) is the doctor's picture of it and what is written down in the medical records, (C) is the content of the report form to the surveillance registry, (D) is the interpretation of that form in the registry, and (E) is the coded data which are stored in the computer.

Kallen: The Epidemiology of Human Reproduction

Goals – Birth Defect Surveillance

- Maintain statewide *surveillance* and monitor trends in birth defect occurrence and mortality
- Provide data to conduct *research* studies to identify genetic and environmental risk factors for birth defects
- Promote *education* activities for the prevention of birth defects
- Provide *outreach* through referral of patients and their families to clinical, educational and social services

Surveillance

Research

Iowa Registry for Congenital and Inherited Disorders

Reproductive Molecular Epidemiology Research and Education Program

RMEREP

Types of Studies

- Record linkage
- (Molecular) Epidemiologic
- Genomic
- Life Course

National Birth Defects Prevention Network NBDPN

Agricultural Health Study

MD STARnet

ELEA Early Life Exposures in Agriculture

National Down Syndrome Project

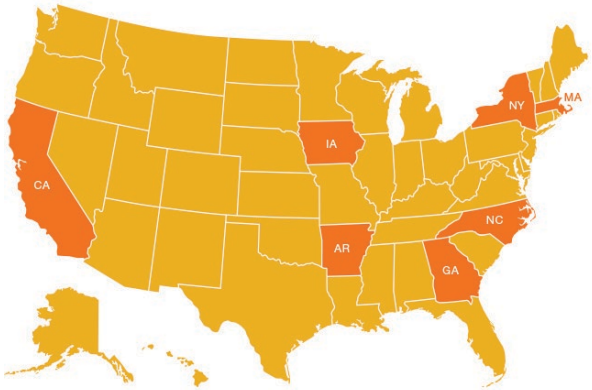
Craniosynostosis Network

Centers for Birth Defects Research & Prevention

nbdps national • birth • defects • prevention • study

bdsteps Centers for Birth Defects Research and Prevention

Centers for Birth Defects Research & Prevention

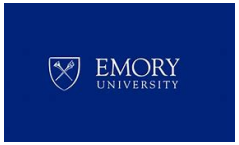


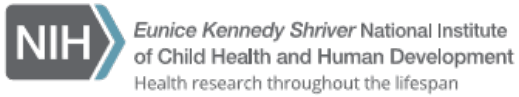
- Began in 1996
- Conducted multistate studies, including the NBDPS, BD-STEPS I, and BD-STEPS II
- Published over 250 NBDPS manuscripts on environmental exposures, genes, and gene-environment interaction effects
- Conducted local studies, including all study types mentioned previously
- Trained over 30 scholars, including post-baccalaureate, masters, doctoral, and postdoctoral scholars

RMEREP

Iowa CBDRP

Iowa Registry for
Congenital and
Inherited Disorders


EMORY
UNIVERSITY


NIH
Eunice Kennedy Shriver National Institute
of Child Health and Human Development
Health research throughout the lifespan

UNIVERSITY OF IOWA
HEALTH CARE

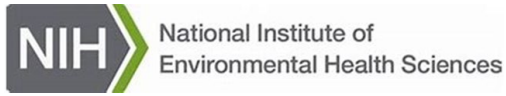
THE
UNIVERSITY
OF IOWA

Icahn School of Medicine
at Mount Sinai

NIH
National Human Genome
Research Institute

PennState

NIH
NATIONAL CANCER INSTITUTE


NIH
National Institute of
Environmental Health Sciences

IDPH

UCDAVIS

CDC

THE
UNIVERSITY
OF UTAH®

State
Hygienic
Laboratory
at The University of Iowa

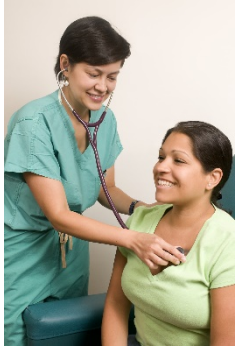
Wake Forest®
School of Medicine
HealthPartners®

Centers for
Birth Defects Research & Prevention

International Clearinghouse
for Birth Defects
Surveillance and Research

eurocat

Multistate Study (NBDPS)



www.freepik.com/free-photo https://beyondins.net www.freestockphotos.biz www.publicdomainfiles.com

Iowa Registry for Congenital and Inherited Disorders

IDPH Iowa Department of Public Health

The University of Iowa

Disinfection By-Products

ORIGINAL RESEARCH ARTICLE

WILEY Birth Defects Research THE TERATOLOGY SOCIETY EST. 1960

Drinking water disinfection byproducts and risk of orofacial clefts in the National Birth Defects Prevention Study

Peter Weyer[1] | Anthony Rhoads[2] | Jonathan Suhl[2] | Thomas J. Luben[3] | Kristin M. Conway[2] | Peter H. Langlois[4] | Dereck Shen[2] | Dong Liang[5] | Soman Puzhankara[2] | Marlene Anderka[6] | Erin Bell[7] | Marcia L. Feldkamp[8] | Adrienne T. Hoyt[4] | Bridget Mosley[9] | Jennita Reefhuis[10] | Paul A. Romitti[2] | The National Birth Defects Prevention Study

Iowa Registry for Congenital and Inherited Disorders

IDPH IOWA Department of PUBLIC HEALTH

THE UNIVERSITY OF IOWA

OTC Cough Medications

Boston University Slone Epidemiology Center
Slone Drug Dictionary

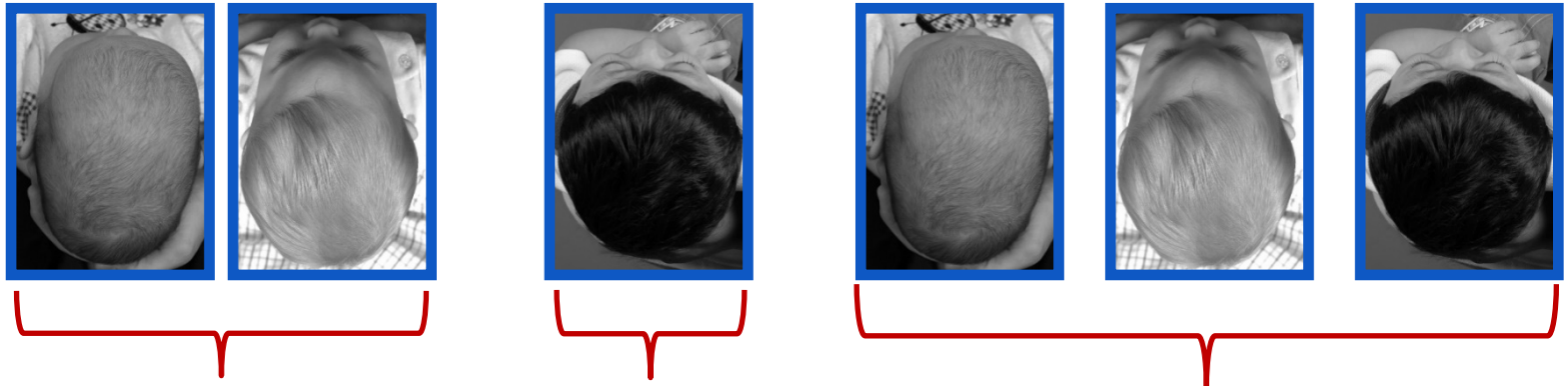
Open access | Original research

BMJ Open

Maternal use of cough medications during early pregnancy and selected birth defects: a US multisite, case–control study

Yanyan Cao,[1] Anthony Rhoads,[1] Trudy Burns,[1] Ryan M Carnahan,[1] Kristin M Conway,[1] Martha M Werler,[2,3] Allen Mitchell,[2] Paul Romitti [1]

Leveraged NBDPS - Craniosynostosis



R01
(Boyd/Romitti PIs
Sheffield Co-I)

BMP2
BBS9

BMP7

P01
(Jabs/Romitti/
Other PIs)

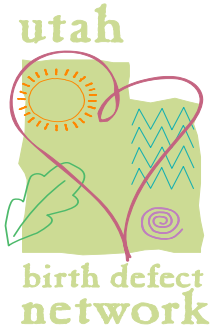
In preparation

Kid's First
(Boyd/Romitti PIs)

Data Analysis

Multistate Multiregistry Study

UTAZIA: Utah Arizona Iowa



OPEN ACCESS Freely available online

PLOS | ONE

Cancer Risk in Children and Adolescents with Birth Defects: A Population-Based Cohort Study

Lorenzo D. Botto[1]*, Timothy Flood[2], Julian Little[3], Mark N. Fluchel[4], Sergey Krikov[1], Marcia L. Feldkamp[1], Yuan Wu[5], Rhinda Goedken[5], Soman Puzhankara[5], Paul A. Romitti[5]

Goals – Birth Defect Surveillance

- Maintain statewide *surveillance* and monitor trends in birth defect occurrence and mortality
- Provide data to conduct *research* studies to identify genetic and environmental risk factors for birth defects
- Promote *education* activities for the prevention of birth defects
- Provide *outreach* through referral of patients and their families to clinical, educational and social services

Education

- **Publications**
  - Manuscripts
  - Reports
  - Press releases
- **Presentations**
  - Communities
  - Schools
  - Professionals
- **Other**
  - Brochures
  - Posters
  - Radio/TV clips

Goals – Birth Defect Surveillance

- Maintain statewide *surveillance* and monitor trends in birth defect occurrence and mortality
- Provide data to conduct *research* studies to identify genetic and environmental risk factors for birth defects
- Promote *education* activities for the prevention of birth defects
- Provide *outreach* through referral of patients and their families to clinical, educational and social services

Parent Notification

- **Identified priority issue in State Genetics Plan of Iowa**

- **Project development**
  - Administrative Rules
  - Physician/Nurse-Practitioner Survey
  - Parent/Professional Work Group
  - Statewide Parent Focus Groups (6)
  - Physicians will be informed of process

- **Notification began with January 1, 2003 births**

Parent Notification

- **Family letter**
  - Eligibility determined by Clinician during Case Review
  - Terminology: lay terms used for medical diagnosis

    Pyloric stenosis—Narrowing of part of the stomach
  - Includes information about available resources

- **Physician letter**
  - Sent to primary care provider
  - Includes copy of family letter

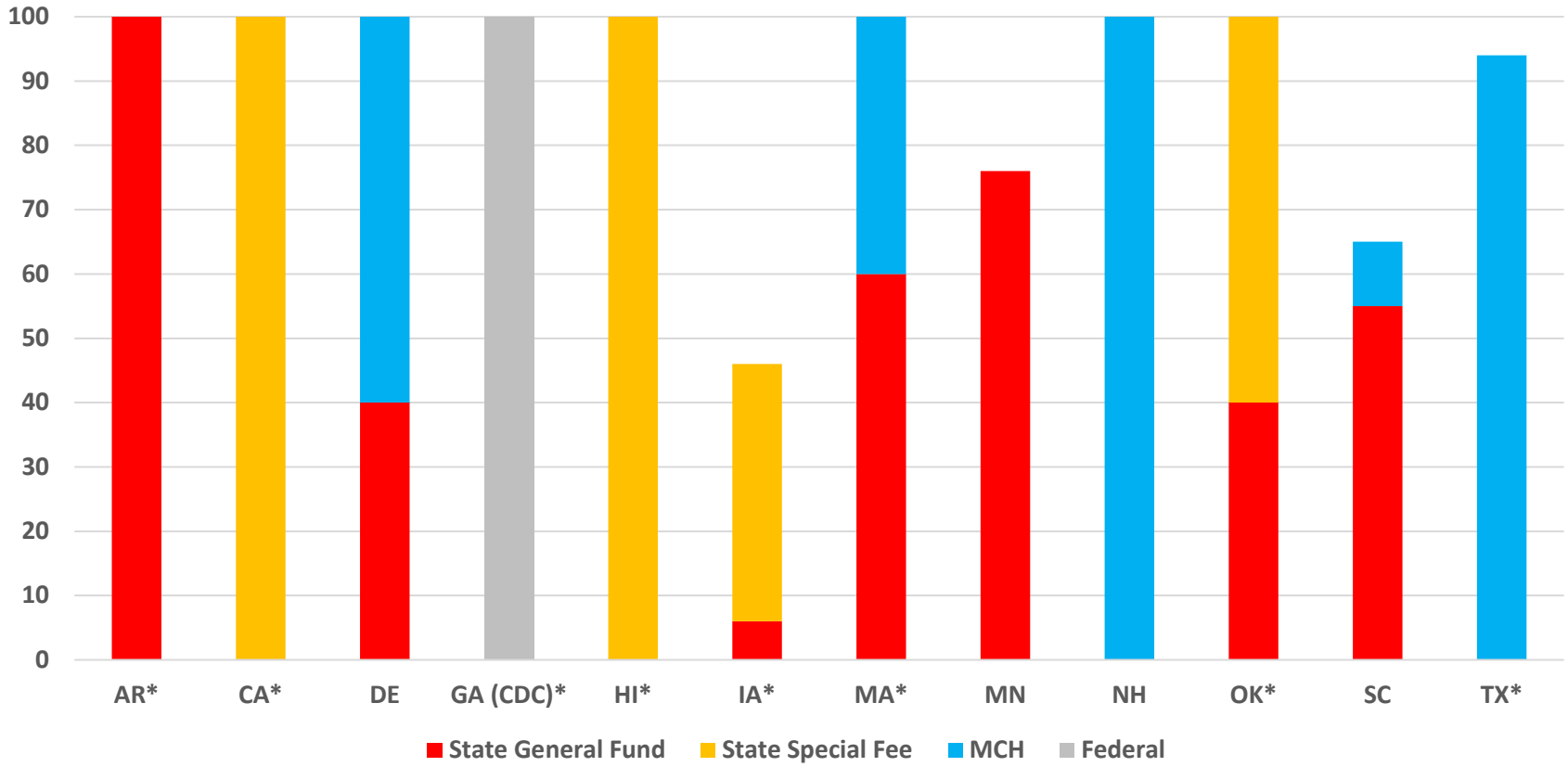
Awards

- Assigned letter grade to birth defect programs
- Grade reflected program's ability to conduct surveillance, research, and prevention activities

A

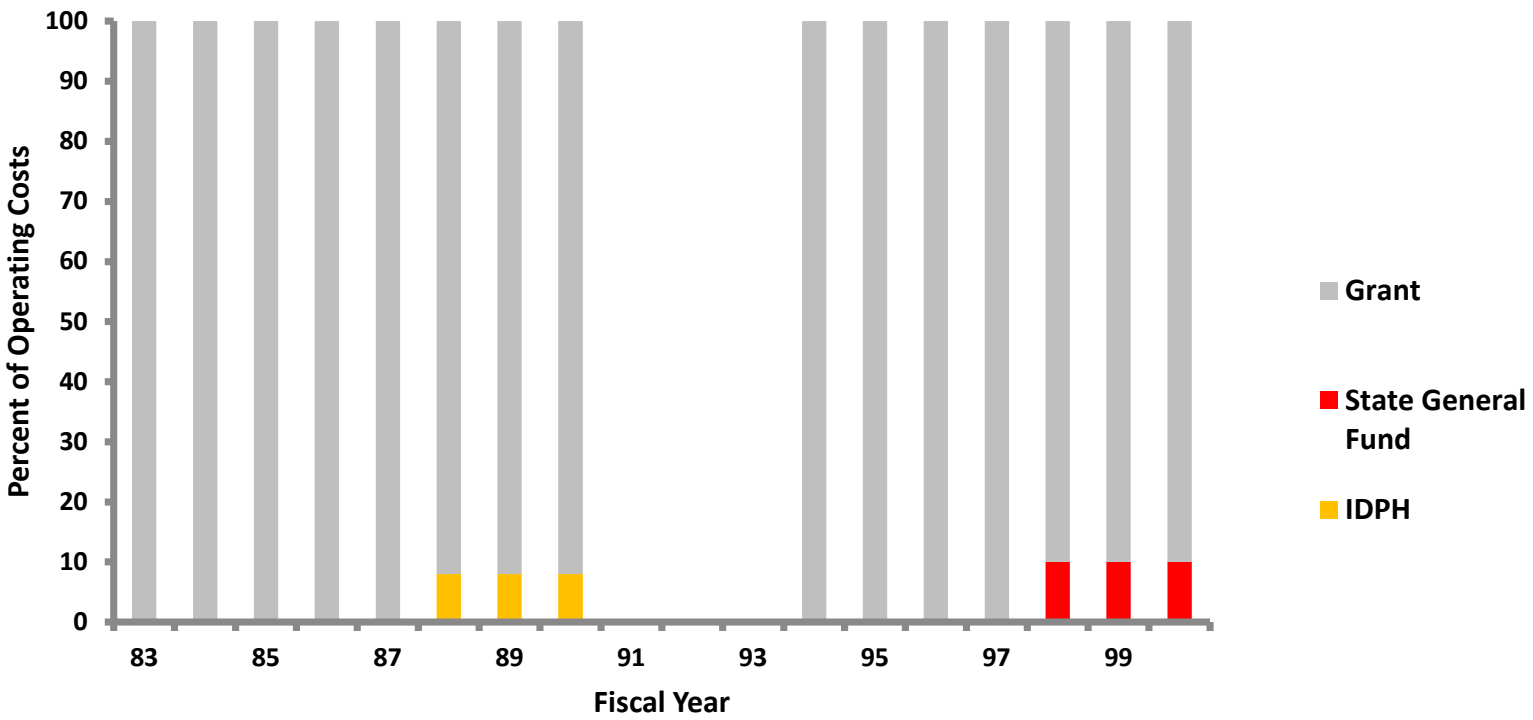
State Leadership Award

Funding for Active Registries

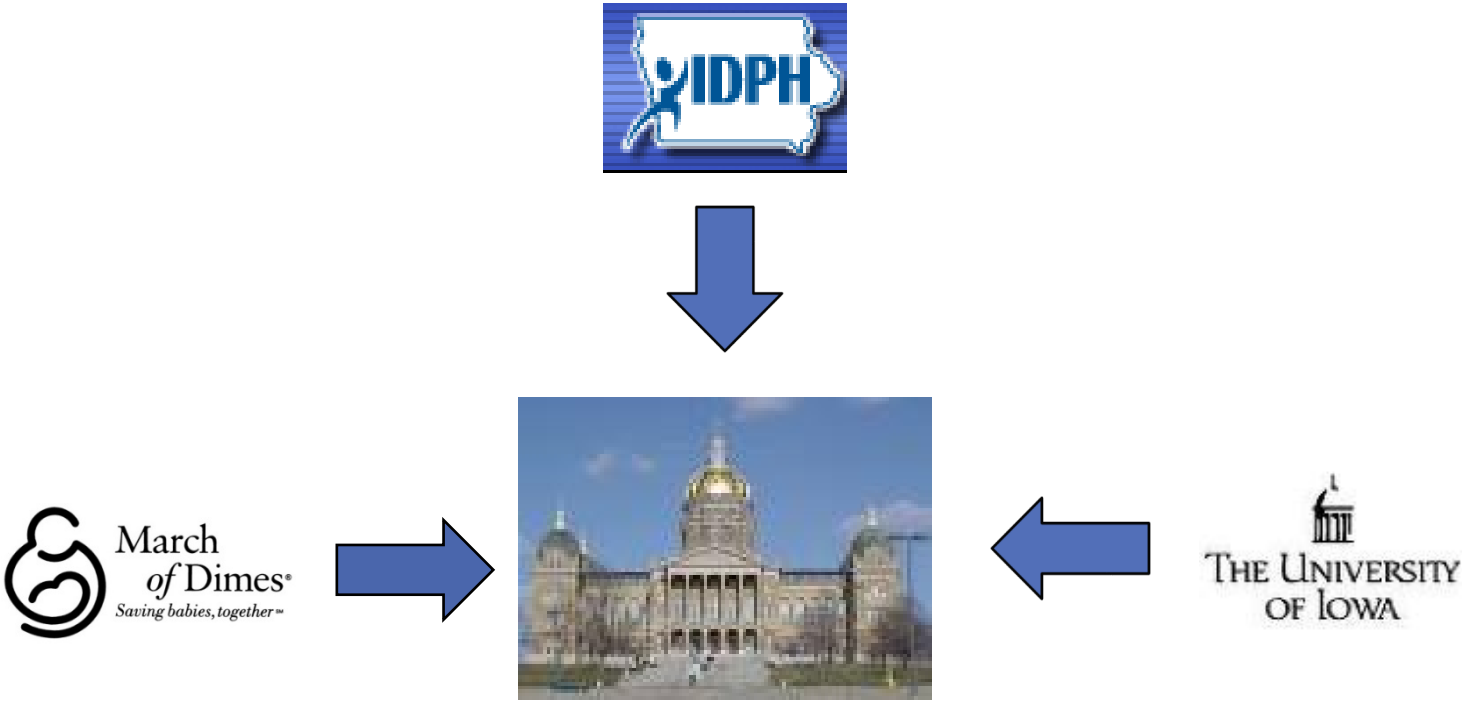


Funding data not available for AZ and NC

Funding Timeline FY1983-2000



Coalition-Building



Birth Defects Surveillance

- Shared field representatives with Iowa Cancer Registry
- Supervised by Iowa Cancer Registry staff
- Lacked adequate time for birth defects training / abstracting
- Required increased training and rapid ascertainment
- Needed to identify funds for dedicated field staff

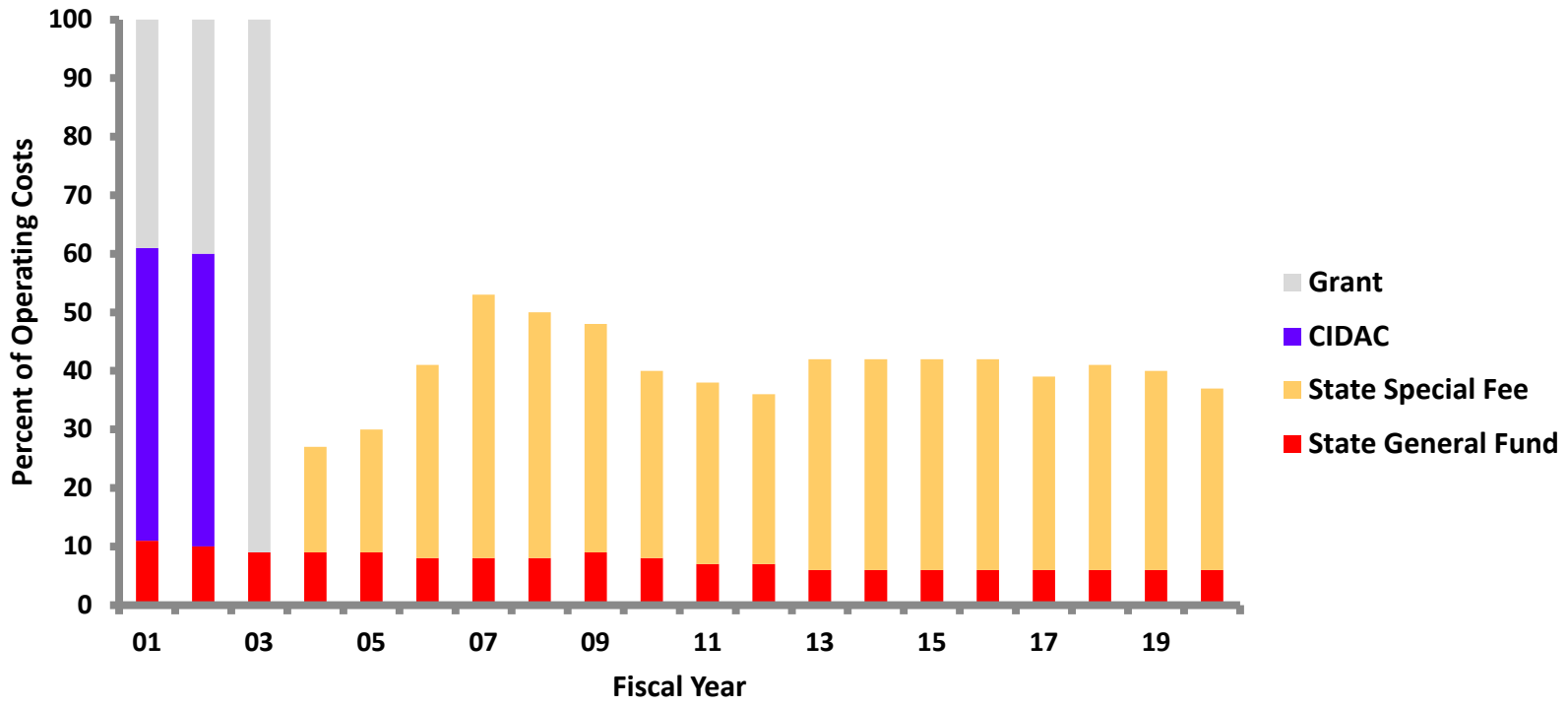
Developmental Funds

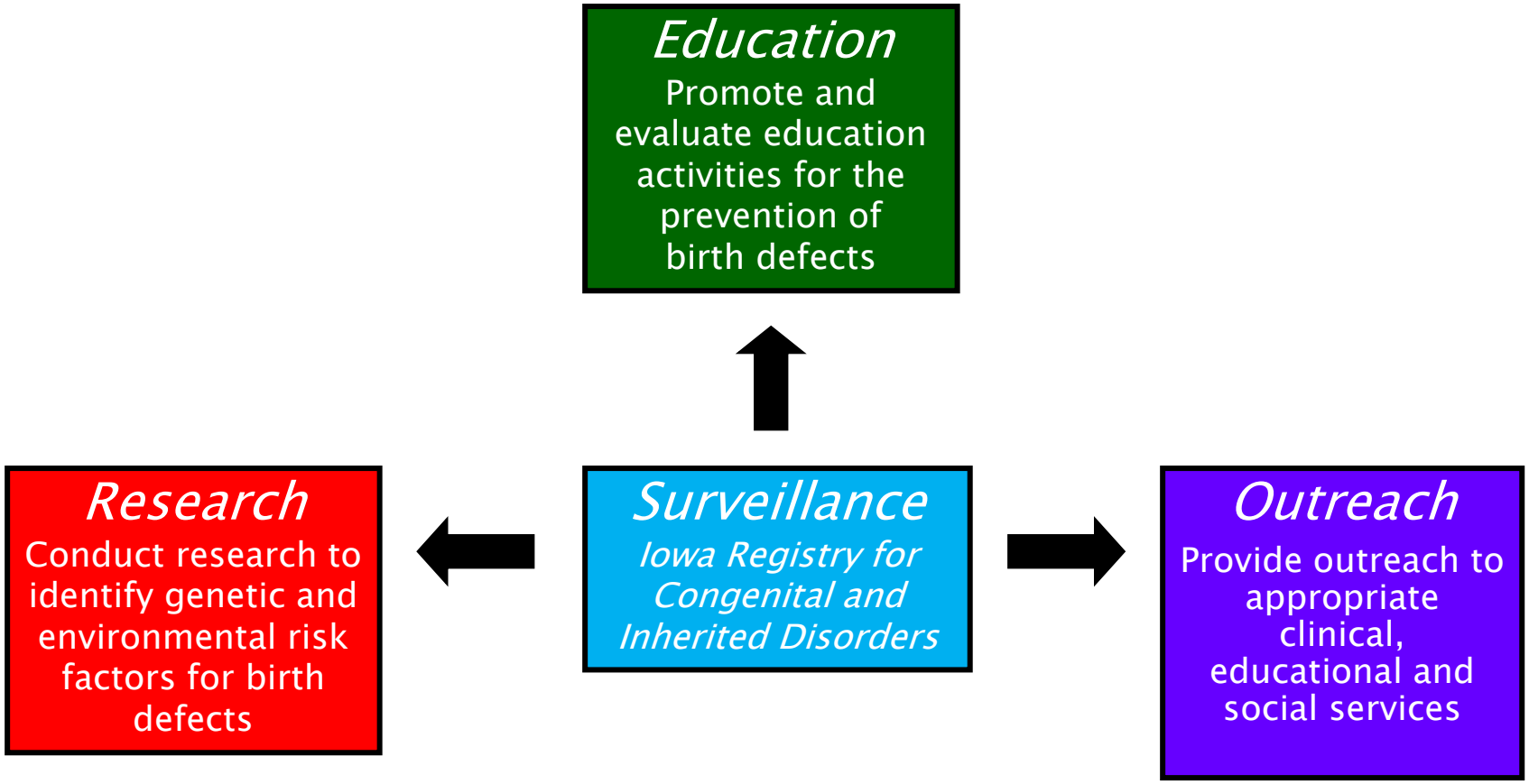
- Received funds from Iowa Birth Defects Institute
- Support for one-year (7/01-6/02) with renewal for one-year
- Sustain funding using phase-in plan with IDPH
- Fund Field Coordinator and Representatives

Birth Certificate Fee Increase

- Four-year effort headed by MOD with assistance from IDPH and UI
- Obtain increased and sustained funding for Registry
- In 2003 and 2004, legislation passed and revised to add fee increase on birth certificates

Funding Timeline FY2001-2020




Education
Promote and evaluate education activities for the prevention of birth defects
Research
Conduct research to identify genetic and environmental risk factors for birth defects
Surveillance
Iowa Registry for Congenital and Inherited Disorders
Outreach
Provide outreach to appropriate clinical, educational and social services

For more information…

- **Iowa Department of Public Health**
  - https://idph.iowa.gov/genetics/public/iowa-registry
- **National Birth Defects Prevention Network**
  - http://www.nbdpn.org/
- **International Clearinghouse for Birth Defects Surveillance and Research**
  - http://www.icdbsr.org/

Thank You!