Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STAR*net*)

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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

- **2001 passage of the Muscular Dystrophy Community Assistance, Research, and Education (MD CARE Act)**

- **MD CARE Act authorized the National Institutes of Health (NIH) to:**
  - Expand and intensify research and related activities for various forms of muscular dystrophy by selected Institutes
  - Create Centers of Excellence for basic and clinical research for various forms of muscular dystrophy

- **MD CARE Act authorized the Centers for Disease Control and Prevention (CDC) to:**
  - Create a National Muscular Dystrophy Epidemiology Program – MD STAR*net*

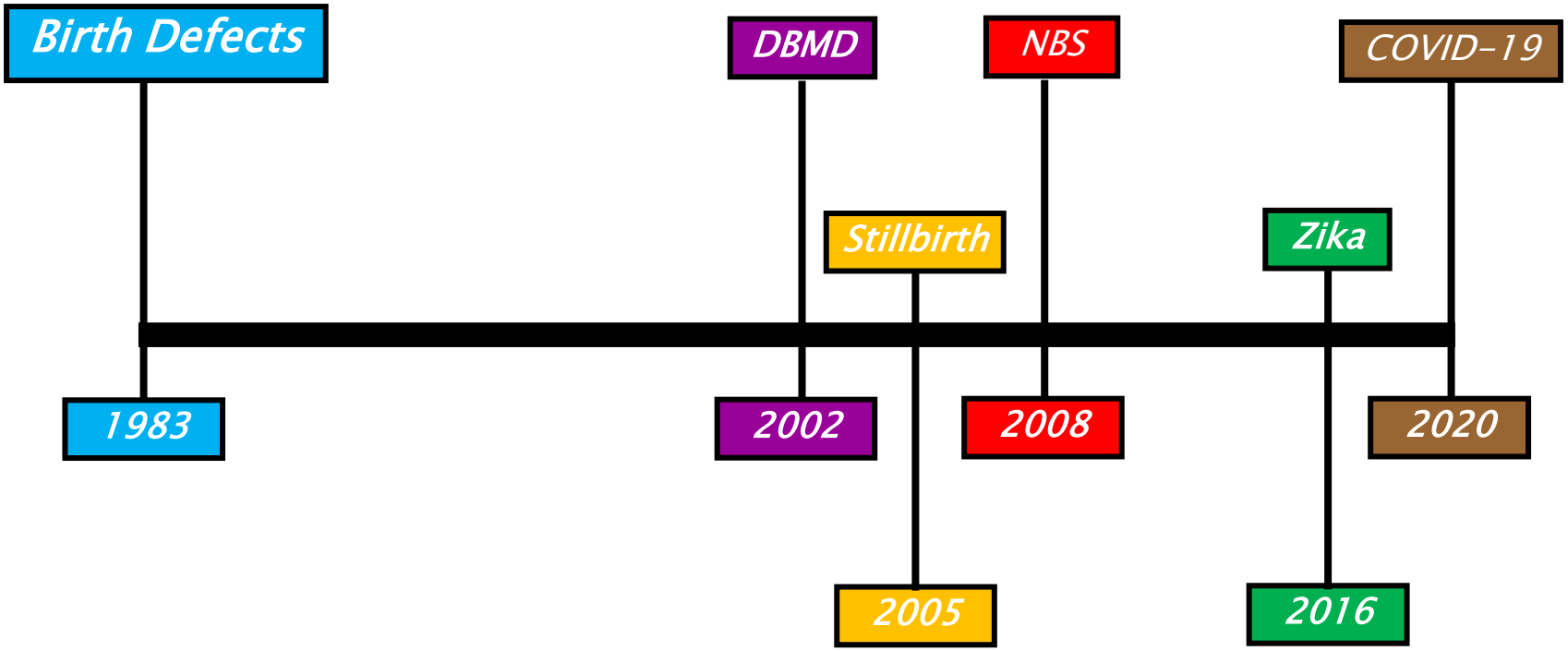
- **MD CARE Act re-authorized in 2008 and 2014**

Approach

- **Expand population-based birth defect surveillance programs to conduct active case finding and record abstraction for muscular dystrophies**

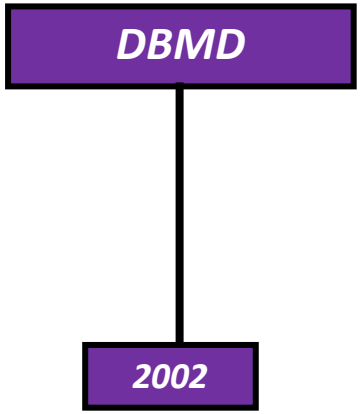
Public Health Surveillance

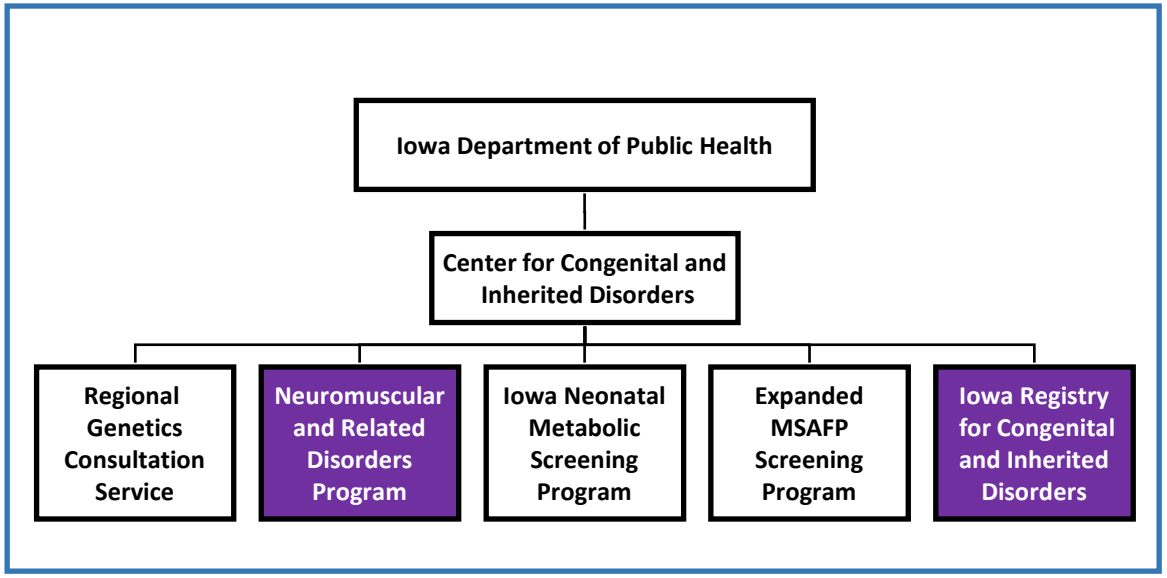
Surveillance Timeline



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

MD Surveillance





DBMD=Duchenne/Becker Muscular Dystrophy

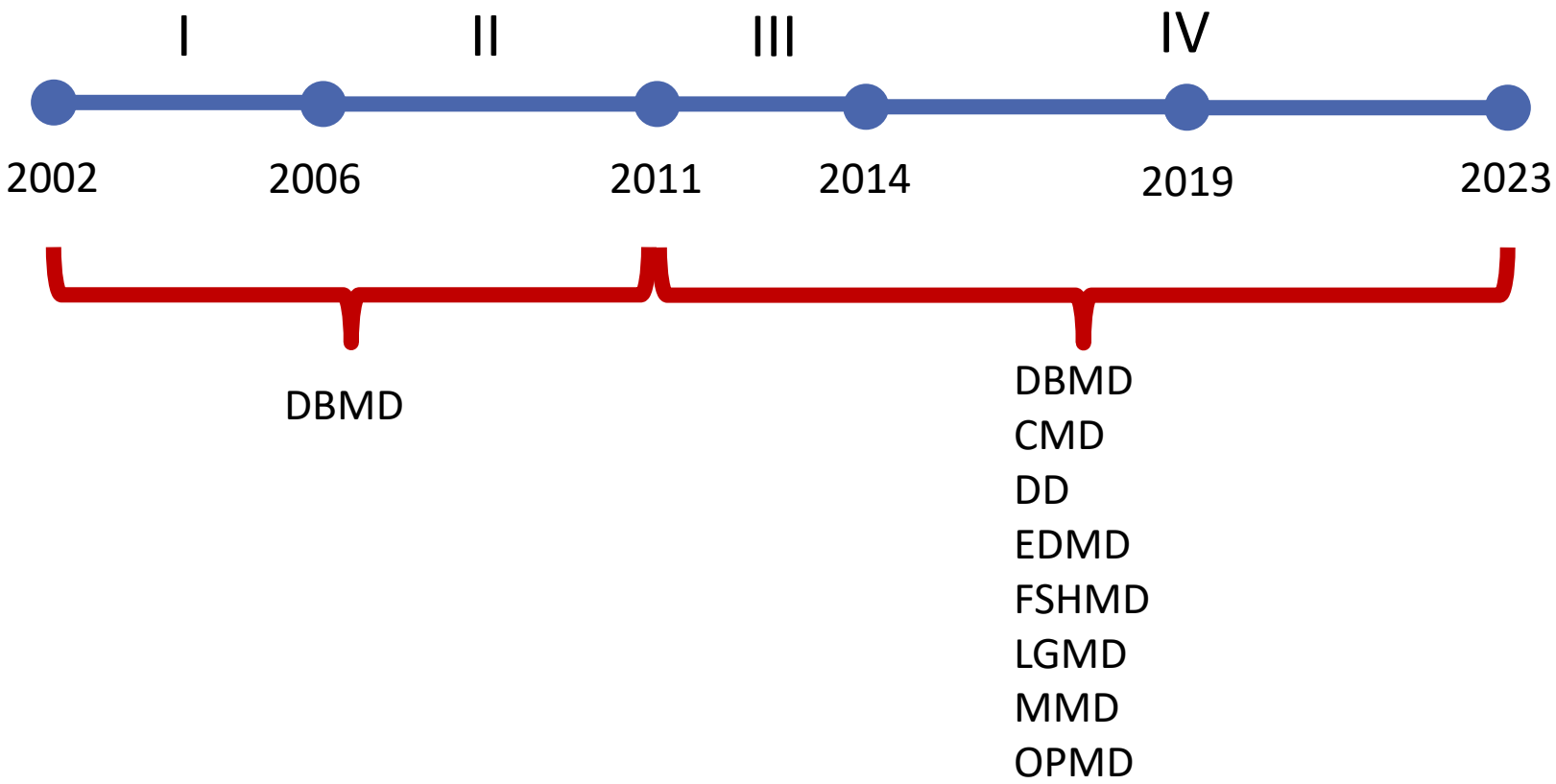
Objectives

- **Characterize prevalence, natural history, healthcare service use and costs, and disparities in access to care, initially for:**
  - Duchenne and Becker Muscular Dystrophy (DBMD)

  and subsequently for:
  - Congenital Muscular Dystrophy (CMD)
  - Distal Muscular Dystrophy (DD)
  - Emery-Dreifuss Muscular Dystrophy (EDMD)
  - Facioscapulohumeral Muscular Dystrophy (FSHMD)
  - Limb-Girdle Muscular Dystrophy (LGMD)
  - Myotonic Muscular Dystrophy (MMD)
  - Oculopharyngeal Muscular Dystrophy (OPMD)

- **Assess whether treatments, interventions, or changes in healthcare use are associated with disease progression and survival**

MD Types


I
II
III
IV
2002
2006
2011
2014
2019
2023
DBMD
DBMD
CMD
DD
EDMD
FSHMD
LGMD
MMD
OPMD

MD STAR*net* Approach

- Core Sites
- Abstractor Coordinating Center
- Data Coordinating Center
- Evaluation Committee

MD STAR*net* Surveillance

Record Source

Raw Data

Requests

Record Abstraction

Clinical Review

Data Transmission and Processing

Requests

Data Review

MD STAR*net* Data Coordinating Center

Transmit Data

MD STAR*net* Site

Record Linkage

Vital Records, Medicaid Records, etc.

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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.

Regulations – Chapter 4 IAC

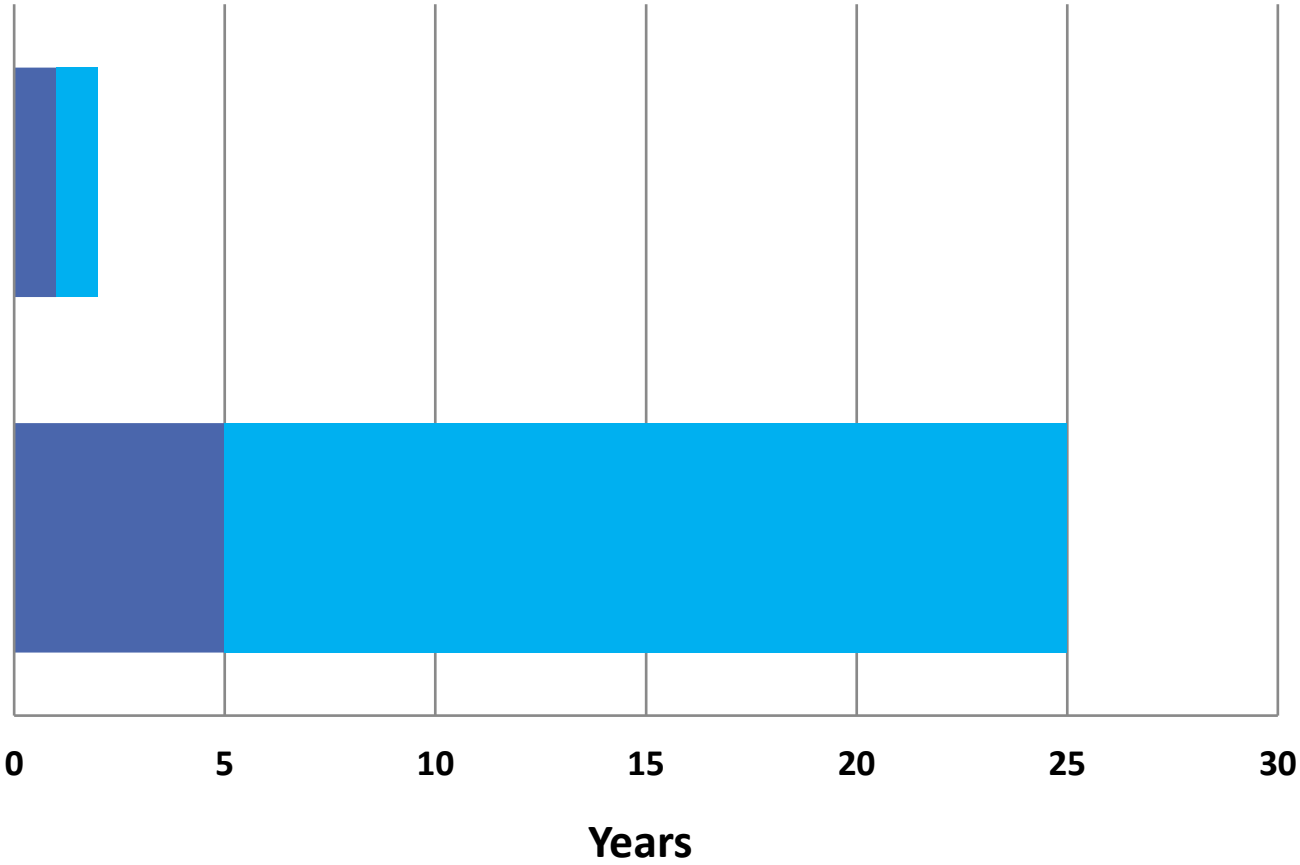
- **641—4.7(136A) Iowa registry for congenital and inherited disorders (IRCID)**

- **4.7(1) Definitions**
  - b. Neuromuscular disorders shall be defined as Duchenne, Becker, congenital, distal, Emery-Dreifuss, fascioscapulohumeral, limb-girdle, myotonic, and oculopharyngeal muscular dystrophies.

DBMD Surveillance Eligibility

- Diagnosis of childhood-onset DBMD
- Residence in an MD STAR*net* site at some time point following diagnosis
- Birthdate on or after January 1, 1982 and on or before December 31, 2011

Age of Onset



Photos: MDA.org; NEFR.org

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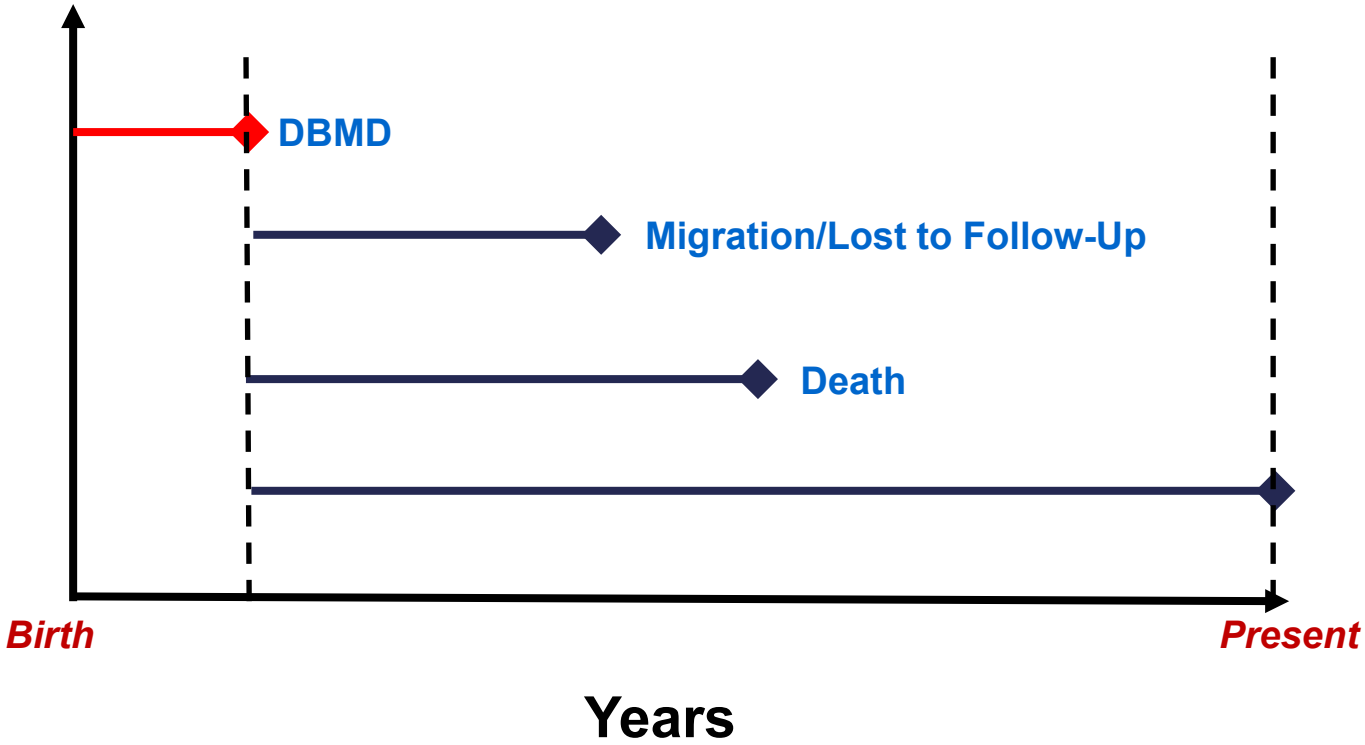
Surveillance Follow-Up



Regulations – Chapter 4 IAC

- **4.7(2) Surveillance policy**
  - b. Records for neuromuscular disorders shall be abstracted pursuant to 641-1.3 (139A) and maintained in the IRCID. Neuromuscular disorders surveillance for individuals of all ages shall be performed in order to determine the ***occurrence and trends of the selected neuromuscular disorders***, to determine ***co-occurring conditions and treatments through annual follow-up abstraction,*** to conduct thorough and complete epidemiological surveys through annual long-term follow-up, and to ***assist in the planning for and provision of services to individuals with selected neuromuscular disorders and their families.***

Surveillance Mission

Identify

Classify

Characterize

Publications

Prevalence of Duchenne and Becker Muscular Dystrophies in the United States

Paul A. Romitti, PhD[a], Yong Zhu, PhD[a], Soman Puzhankara, MS[a], Katherine A. James, PhD[b], Sarah K. Nabukera, MD, DrPH[a], Gideon K.D. Zamba, PhD[a], Emma Ciafaloni, MD[c], Christopher Cunniff, MD[d], Charlotte M. Druschel, MD, MPH[e,f], Katherine D. Mathews, MD[a], Dennis J. Matthews, MD[b], F. John Meaney, PhD[d], Jennifer G. Andrews, MBA[d], Kristin M. Caspers Conway, PhD[a], Deborah J. Fox, MPH[e], Natalie Street, MS, CGC[g], Melissa M. Adams, PhD[g], Julie Bolen, PhD, MPH[g], on behalf of the MD STAR*net*

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Healthcare Use

Co-Morbidities

Delayed Diagnosis in Duchenne Muscular Dystrophy: Data from the Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STARnet)

Emma Ciafaloni, MD, Deborah J. Fox, MPH, Shree Pandya, PT, MS, Christina P. Westfield, RN, BSN, Soman Puzhankara, MS, Paul A. Romitti, PhD, Katherine D. Mathews, MD, Timothy M. Miller, MD, Dennis J. Matthews, MD, Lisa A. Miller, MD, MSPH, Christopher Cunniff, MD, Charlotte M. Druschel, MD, MPH, and Richard T. Moxley, MD

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Surveillance Data

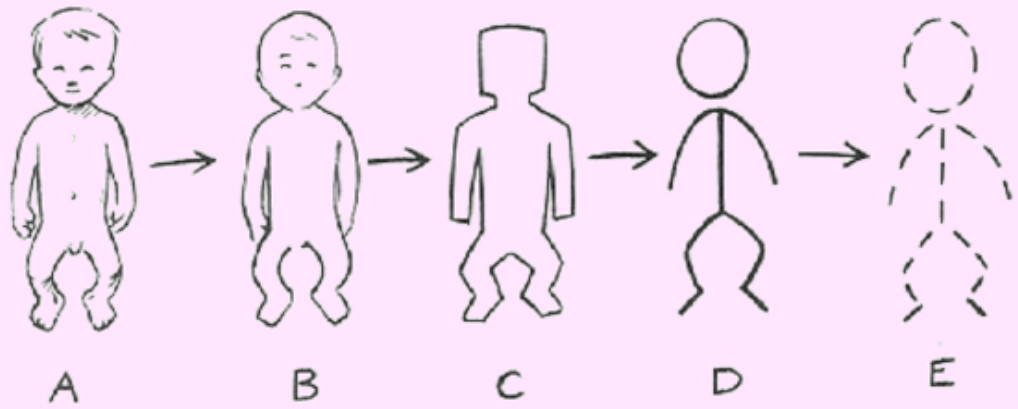

Diagram illustrating the distortion of the information in a reporting system, from the infant to the coded data. (A) shows the actual infant, (B) is the doctor's picture of it and what is written down in the medical records, (C) is the content of the report form to the surveillance registry, (D) is the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

Survey Studies

- Baseline Interview (Caregiver)
- Needs Assessment Survey (Caregiver and Oldest Affected Male)
- Quality of Life Survey (Caregiver)
- Health Care and Other Life Transitions (Caregiver and Oldest Affected Male)

Strengths

- Multi-disciplinary expertise (clinical and epidemiologic)
- Population-based data collection with multi-record source case ascertainment and annual follow-up abstraction
- Systematic data collection procedures for surveillance and survey studies
- Centralized and rigorous quality assurance/quality control procedures

Future Directions

- Estimate prevalence and describe mortality
- Track access to and coverage for new FDA-approved medications and compare outcomes of individuals taking and not taking these drugs
- Evaluate care received in accordance with care considerations or guidelines
- Explore the use of surveys and collaborations to answer research questions not addressed through surveillance data
- Continue to provide data to providers, advocates, and policy makers to inform decision making

Iowa MD STAR*net* Core Site

- Continue to conduct surveillance statewide
- Pool coded, de-identified data at MD STAR*net* DCC
- Established data use agreements with all core sites (CDC in-process)
- Conduct pooled analysis of surveillance data

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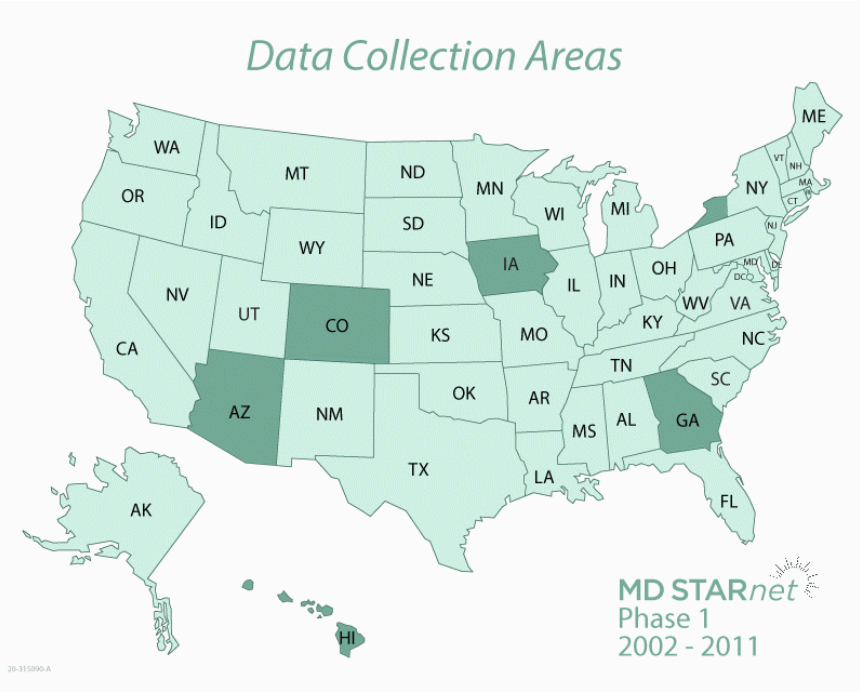

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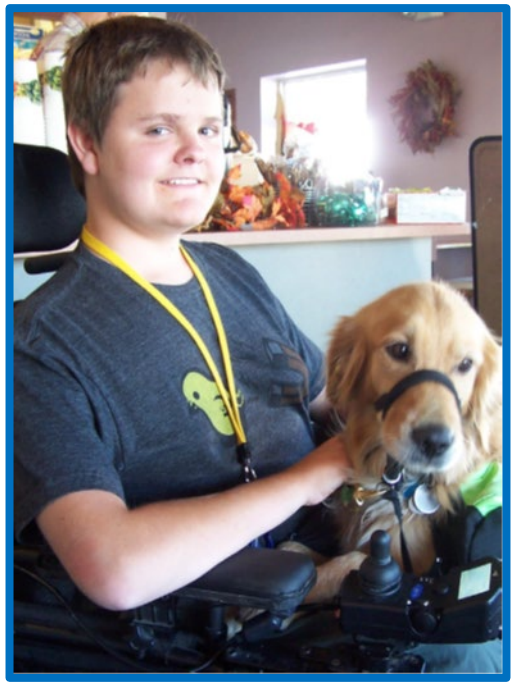
For more information…

- **Centers for Disease Control and Prevention**
  - https://www.cdc.gov/ncbddd/musculardystrophy/research.html

Thank You!



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