Evolution of the Utah Birth Defect Network

Different Strokes:
How the Mission and Objectives of a Birth Defect Program Shapes its Data Collection and Uses

12th Annual
National Birth Defects Prevention Network Meeting
February 23, 2009
Marcia Feldkamp, PhD, PA
Birth Defects Surveillance

- How do we improve our understanding of birth defects, so, ultimately, we can prevent them?
Goals of Surveillance in Utah

- Statewide and population-based
- Capture all pregnancy outcomes
- Clinically well-defined
  - Reviewed by a geneticist
- Case base for research
MISSION STATEMENT

The Utah Birth Defect Network seeks to prevent birth defects and secondary disabilities through public health surveillance, outreach to families and health care providers, and epidemiologic studies.
Challenges of Surveillance

- Getting surveillance started
- Co-agency program
- Building the surveillance team
- Developing & maintaining reporting sources
- Getting surveillance funded
- Improving data over time – not a perfect system
- Assessing data quality
- Database requires constant improvements
- Keeping surveillance funded during this economic crisis
Circa 1990

- Began discussing a birth defect registry
- Worked within the Division of Community and Family Health Services, UDOH
- Critical to the success
  - Dr. George Delavan
    - CSHCN Bureau Director, UDOH
    - CFHS Division Director
  - Dr. John Carey
Circa 1990

- Early Challenges

- Competition for leadership
  - Different views expressed on developing a birth defect surveillance system
  - UDOH leadership given opportunity to determine direction

- Viewed as an outsider

- Data collection tool
  - Too much vs. too little
Circa 1993

- Developed NTD pilot project under the wings of the Developmental Disability Grant from CDC.

- Pilot - used the usual suspects for identification of potential cases and:
  - Genetic counselors
  - Champion model

- Created ability to identify all pregnancy outcomes
Circa 1994

- NTD pilot project went well
  - Surveillance team = 1
  - Data collection
  - Developed database (EpiInfo)

- Began to add other birth defects
  - Based on conditions obvious either prenatally or postnatally
Permission granted to submit a proposal to CDC for funding of birth defects surveillance

UDOH Program received MOD award (1995-1996)
- Hired their own person to:
  - Assist with data abstraction
  - Develop database

Utah received a CDC surveillance award (1995-1998)
- Permitted leadership to be determined
- Process to be better defined
- Surveillance team = 3
- Clinical expertise = 0.05
Circa 1997

- Flying by the seat of my pants
- Collecting data on unsuspecting mothers, fathers and infants
- Worked with the UDOH attorneys to draft an Administrative Rule
1999 - Reporting Rule

- State of Utah - Administrative Rule
  - Rule R398-5. Birth Defects Reporting

- What does this mean?
  - Mandated reporting for birthing hospitals
  - Mandated reporting for laboratories
  - Protects providers that report voluntarily
  - Allows the UBDN to collect information from the medical records of affected infants and their mothers
  - All pregnancy outcomes were covered by using broad terminology
    - One cannot forget that we live in Utah!
Funding Surveillance

- Challenge

  - Annual submission of building block
    - 1999 - 2005
  - UDOH building block submitted to Governor
    - 2005 submitted for 2006 session
      - Legislature did not approve funding
    - 2006 submitted for 2007 session
      - Approved *ongoing* funding
  - 2008 – free and clear
  - 2009 – funding questioned – no cuts
  - 2010 – still remains to be seen
Who We Are

- Utah Birth Defect Network
  - Co-agency program
    - Children with Special Health Care Needs
      - Utah Department of Health
    - Division of Medical Genetics
      - Department of Pediatrics, University of Utah
  - Surveillance
    - Mother is a Utah resident at delivery
    - All pregnancy outcomes ascertained
      - Live births, stillbirths, pregnancy terminations, miscarriages
    - Major structural malformations
    - Adding new conditions
Surveillance - Research - Prevention

Utah Birth Defect Network
Population-based Surveillance of Major Birth Defects
>50,000 births and >1,100 cases / year

National Birth Defects Prevention Study
Quality of Life for Craniofacial Defects
Birth Defects and Childhood Cancer Study
Utah Population Database Linkage

NTD Primary Prevention – Statewide Education & WIC Project
NTD Recurrence Prevention
Education and Outreach
Birth Defects Ascertained 1994 - 1997

1994
- Neural tube defects

1995
- Oral facial clefts
- Common trisomies (13, 18 and 21)

1997
- Abdominal wall defects
- Limb reduction defects
- Skeletal dysplasias
- Arthrogryposis
- Congenital heart defects
  - Conotruncal
  - Left sided obstructive lesions
- Chromosomal abnormalities
  - Unbalanced
  - Deletions
Birth Defects Ascertained - January 1999

- Congenital heart defects (excluding VSDs)
- Craniosynostosis
- Dandy-Walker
- Holoprosencephaly
- Hydranencephaly
- Microcephaly
- Other reduction deformities
- Hydrocephalus
- Congenital cataracts/glaucoma
- Aniridia
- Anophthalmia/microphthalmia
- Anotia/microtia
- Choanal atresia
- Lung agenesis/hypoplasia
- Diaphragmatic hernia
- TEF/esophageal atresia
- Pyloric stenosis
- Biliary atresia
- Intestinal atresia/stenosis
- Imperforate anus
- Hirschsprung’s
- Renal agenesis/dysgenesis
- Cloacal/bladder extrophy
- Obstructive GU defects
- Hypospadias/epispadias
UBDN Reporting Sources

- Hospital Champions
- Vital Records
  - Birth Certificates
  - Fetal Death Certificates
  - Death Certificates
- Cytogenetic Laboratories
- Hospital Discharge Data
- Delivery Hospitals
- Tertiary Care Facilities
- Community Craniofacial and Plastic Surgeons
- Community Urologists
- PCMC Specialty Clinics

- Pathology
  - University of Utah
  - Primary Children’s Medical Center
  - Community Hospitals
- Log Books
  - Newborn Nursery
- Prenatal Ultrasound
- Labor and Delivery
- NICU
- PCMC NICU
- Prenatal Diagnostic Centers
  - Genetic Counselors
  - Diagnostic Conference

Most reporting sources require constant vigilance.
### Case Data Entry Form

#### Location of Case Record Form
- **Case Completed - Filed**
- **Surveillance**

#### Status
- **First Data Entry Complete**
- **1003 Transferred to CDC**

#### Child Info
- **Source**
- **Address**
- **Family History**
- **Physician**
- **Previous Preg Info**
- **Index Preg/ PN Comp**
- **Procedures Exams**
- **Infant Info / Lab Test**
- **Birth Defects**

#### Birth Defects
- **Birth Defect Code**: Trisomy 21/Down syndrome with
  - **Check Codes**: Major
  - **Prenatally Diagnosed**: Yes
  - **Birth Defect Description**:
  - **NBDPS Eligible**: No

- **Birth Defect Code**: Hirschsprung's disease, NOS
  - **Check Codes**: Major
  - **Prenatally Diagnosed**: No
  - **Birth Defect Description**:
  - **NBDPS Eligible**: No

- **Birth Defect Code**: Simian crease / Transverse palm
  - **Check Codes**: Minor
  - **Prenatally Diagnosed**: No
  - **Birth Defect Description**: Bilateral simian crease
  - **NBDPS Eligible**: No

#### Enroll in NBDPS

**Classification**: Multiple Chromosomal
- **Familial**: No
- **Etiology Known**: Yes

#### Birth Defect Category
- **Gastrointestinal**: 1003
- **Trisomy 21**: 1003

#### Clinical Case Review Comments

**Clinical Case Reviewer**:

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*Note: The form contains various fields for data entry, including check codes for birth defects and options for enrollment and classification.*
Classification of Birth Defects

- A classification tool that mirrors how normal structures develop

Coding
- ICD9 and BPA
  - Common classification schemes have many benefits
  - Not targeted to studying birth defect causes or trends
  - Split or lump defects based on anatomy rather than embryology

Classification
- Dr. John Carey devised a classification tool
- Data abstractors do not code any birth defect data
Classification of Birth Defects

Clinical geneticists consider:

- mechanism = pure defects, sequences, developmental field defects
- cause = chromosome abnormalities, genetic conditions, teratogens, in-utero events
- family history = first degree relative with same defect
- morphology = descriptive, anatomical (e.g., oral facial anomalies)
Classification tool in action

Birth Defect Cases

Is Cause Known?

Non Syndromic

Does the case meet the definition of developmental field, sequence or isolated pure?

Yes

Isolated

No

Multiple

Syndromic
### SURVEILLANCE Classification Tool in Action

<table>
<thead>
<tr>
<th>Classification</th>
<th>1994-2006</th>
<th>1999-2006</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Frequency (%)</td>
<td>Prevalence</td>
</tr>
<tr>
<td>Isolated</td>
<td>6884 (69.4)</td>
<td>1 in 88</td>
</tr>
<tr>
<td>Multiple</td>
<td>1041 (10.5)</td>
<td>1 in 585</td>
</tr>
<tr>
<td>Syndromic</td>
<td>1997 (20.1)</td>
<td>1 in 304</td>
</tr>
<tr>
<td>Total</td>
<td>9922</td>
<td>1 in 61</td>
</tr>
</tbody>
</table>
### Classification

<table>
<thead>
<tr>
<th>Classification</th>
<th>Cases (%)</th>
<th>Familial (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Pure</strong></td>
<td>6631</td>
<td>202 3.0</td>
</tr>
<tr>
<td>Pure</td>
<td>96.3</td>
<td>3.0</td>
</tr>
<tr>
<td><strong>Sequences</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Pierre Robin</td>
<td>45 0.7</td>
<td>4 8.9</td>
</tr>
<tr>
<td>Amniotic Band</td>
<td>43 0.6</td>
<td></td>
</tr>
<tr>
<td>ABS &amp; Limb-Body Wall</td>
<td>14 0.2</td>
<td></td>
</tr>
<tr>
<td>Limb-Body Wall</td>
<td>19 0.3</td>
<td></td>
</tr>
<tr>
<td>Frontonasal Dysplasia</td>
<td>1 0.0</td>
<td></td>
</tr>
<tr>
<td>Oligohydramnios</td>
<td>40 0.6</td>
<td></td>
</tr>
<tr>
<td>Urethral Obstruction</td>
<td>20 0.3</td>
<td></td>
</tr>
<tr>
<td>Twinning Abnormality</td>
<td>29 0.4</td>
<td></td>
</tr>
<tr>
<td><strong>Developmental Field</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Sirenomelia</td>
<td>1 0.0</td>
<td></td>
</tr>
<tr>
<td>Holoprosencephaly</td>
<td>9 0.1</td>
<td>2 22.2</td>
</tr>
<tr>
<td>Cloaca</td>
<td>4 0.1</td>
<td></td>
</tr>
<tr>
<td>Cantrell Pentology</td>
<td>3 0.0</td>
<td></td>
</tr>
<tr>
<td>Heterotaxia</td>
<td>25 0.4</td>
<td>3 12.0</td>
</tr>
</tbody>
</table>

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### Diagram

- **Yes:** field, sequence or isolated pure?
  - **Isolated**
  - **Multiple**
  - **Syndromic**
<table>
<thead>
<tr>
<th>Classification</th>
<th>Cases (%)</th>
<th>Familial (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Multiple</td>
<td></td>
<td></td>
</tr>
<tr>
<td>2+ Majors</td>
<td>708</td>
<td>17</td>
</tr>
<tr>
<td>1Major/Minors</td>
<td>194</td>
<td>8</td>
</tr>
<tr>
<td>Provisionally Unique</td>
<td>95</td>
<td>13</td>
</tr>
<tr>
<td>Association</td>
<td>24</td>
<td>1</td>
</tr>
<tr>
<td>Recognizable Pattern</td>
<td>20</td>
<td>1</td>
</tr>
</tbody>
</table>

The diagram illustrates the classification of cases into isolated, multiple, or syndromic categories based on whether the field, sequence, or isolated condition is present.
<table>
<thead>
<tr>
<th>Classification</th>
<th>Cases (%)</th>
<th>Familial (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Syndromic</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Chromosomal</td>
<td>1642 82.2</td>
<td>31 1.9</td>
</tr>
<tr>
<td>Genetic</td>
<td>315 15.8</td>
<td>96 30.5</td>
</tr>
<tr>
<td>Teratogen</td>
<td>40 2.0</td>
<td>3 7.5</td>
</tr>
</tbody>
</table>

**Diagram:**
- Yes: Isolated
- No: Multiple, Syndromic
# Syndromic cases

(Known cases)

<table>
<thead>
<tr>
<th>Chromosomal 82.2%:</th>
<th>Cases (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Trisomy21</td>
<td>856 (52.1)</td>
</tr>
<tr>
<td>Trisomy18</td>
<td>199 (12.1)</td>
</tr>
<tr>
<td>Trisomy13</td>
<td>92  (5.6 )</td>
</tr>
<tr>
<td>Turner</td>
<td>137 (43.5)</td>
</tr>
<tr>
<td>Deletion 22q11</td>
<td>73  (4.4 )</td>
</tr>
<tr>
<td>Prader-Willi 15q deletion</td>
<td>21  (1.3 )</td>
</tr>
<tr>
<td>Wolf-Hirschhorn 4p deletion</td>
<td>4   (0.2 )</td>
</tr>
<tr>
<td>Other conditions</td>
<td>260 (15.8)</td>
</tr>
<tr>
<td><strong>Total # of cases</strong></td>
<td><strong>1642</strong></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Genetic 15.8%:</th>
<th>Cases (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Total # of cases</strong></td>
<td><strong>315</strong> (1.0)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Teratogen 2.0%:</th>
<th>Cases (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>IDDM</td>
<td>20 (50.0 )</td>
</tr>
<tr>
<td>Cytomegalovirus</td>
<td>6  (15.0)</td>
</tr>
<tr>
<td>Valproic Acid</td>
<td>11 (27.5 )</td>
</tr>
<tr>
<td>Accutane</td>
<td>1   (2.5 )</td>
</tr>
<tr>
<td>SLE</td>
<td>1   (2.5 )</td>
</tr>
<tr>
<td>Varicella</td>
<td>1   (2.5 )</td>
</tr>
<tr>
<td><strong>Total # of cases</strong></td>
<td><strong>40</strong></td>
</tr>
</tbody>
</table>
PREVENTION
UBDN Web Site

www.health.utah.gov/birthdefect
PREVENTION
Outreach: Family Meetings

2006

A Utah Conference for
Everyone Affected by Birth Defects

Birth Defects: A Lifetime Journey for Children and Families
Keynote Speaker: Kurt Bestor
Musician/Composer & KSTAR Radio Personality
Saturday, January 21 - 1-3 p.m.
Spencer F. and Clerna Eccles Health Sciences Education Building
The University of Utah

Every year, more than 1400 Utah children are born with birth defects. This begins a life-changing journey that impacts families, communities, and society. Come to Utah's first conference on birth defects, to learn and network with others affected by birth defects. Parents and parents-to-be, family members, providers, educators, and legislators are especially invited.

2007

A Utah Conference for
Everyone Affected by Birth Defects

Birth Defects: A Lifetime Journey for Children and Families
Keynote Speaker: Dr. Michael Ballam
Musician/Parent
Saturday, January 20 - 1-4 p.m.
Spencer F. and Clerna Eccles Health Sciences Education Building
The University of Utah

Every year, more than 1400 Utah children are born with birth defects. This begins a life-changing journey that impacts families, communities, and society. Come to Utah's second annual conference on birth defects, to learn and network with others affected by birth defects. Parents and parents-to-be, family members, providers, educators, and legislators are especially invited.
Lessons Learned

- Parallels to parenting – pick your battles that are worth fighting
- UDOH support critical
  - Varied over time, depending on who was at the helm
- Cohesive (internal) program better than multiple programs trying to work together
- Advantages and disadvantages to co-agency program
- Takes a long time to evolve a system
- Start small and think big
  - Data collection - instrument
  - Training and keeping staff a critical element to success
  - Database development and tweaking
  - QA issues take a long time to implement
- Finding the right people that work well together
- Having enough money to do all that you want will always be a challenge
- Never give up!
The Utah Birth Defect Network seeks to prevent birth defects and secondary disabilities through public health surveillance, outreach to families and health care providers, and epidemiologic studies.
Acknowledgments

- Dr. George Delavan
  - Division Director, Community and Family Health Services

- UBDN Group
  - Clinical Team
    - Dr. John Carey
    - Dr. Lorenzo Botto
    - Dr. Jan Byrne
  - Surveillance Team
    - Miland Palmer
    - Kara Lecheminant
    - Toni Fightmaster
    - Kristin Wiley
    - Tricia Rawson
    - Aliese Franck
  - Research Team
    - Jane Johnson
    - Sergey Krikov
    - Mary Bishop Stone
    - Sivithee Srisukumbornchaei
    - Amy Nance
    - Patty Smith
    - Marcella Montoya
    - Denise Spicer
Thank You!