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
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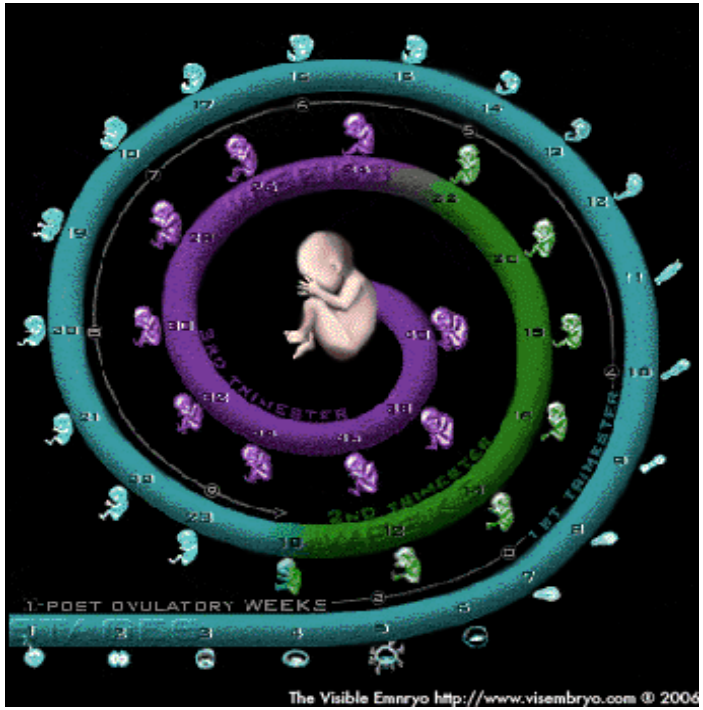
Birth Defects Surveillance

- 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

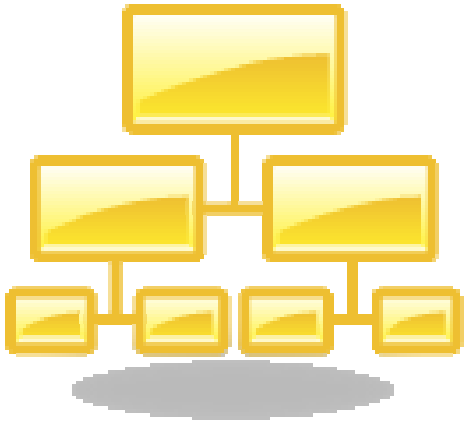
The Utah Birth Defect Network seeks to prevent birth defects and secondary disabilities through public health surveillance, outreach to families and health care providers, and epidemiologic studies.

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

Circa 1994

- 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

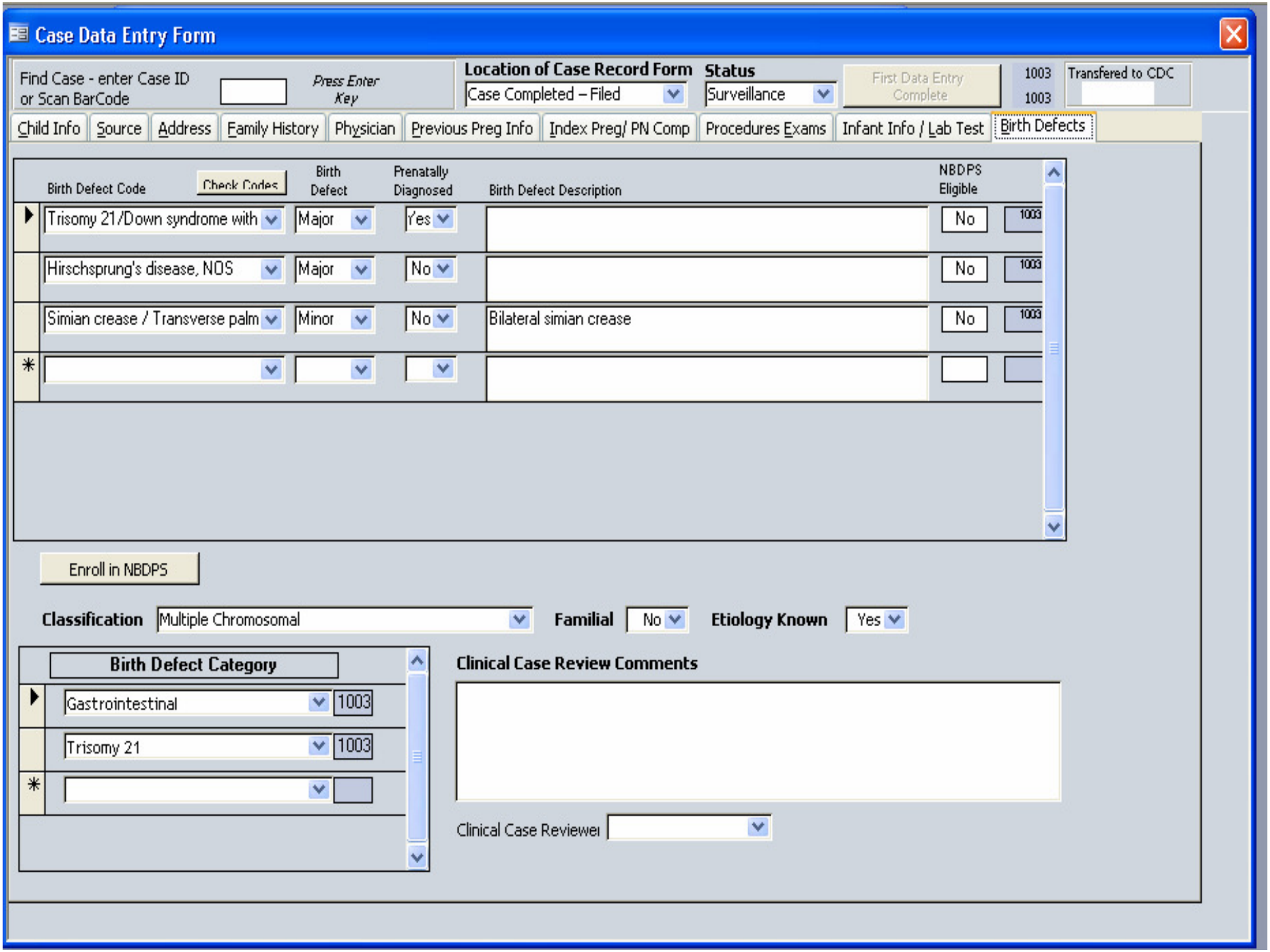
Birth Defects Ascertained - January 1999

- 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 exstrophy
- 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
  - Prenatal Ultrasound
  - Labor and Delivery
  - Newborn Nursery
  - NICU
- PCMC NICU
- Prenatal Diagnostic Centers
  - Genetic Counselors
  - Diagnostic Conference

Most reporting sources require constant vigilance


Case Data Entry Form
Find Case - enter Case ID or Scan BarCode
Press Enter Key
Location of Case Record Form
Case Completed – Filed
Status
Surveillance
First Data Entry Complete
1003
1003
Transfered 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 Defect Code
Check Codes
Birth Defect
Prenatally Diagnosed
Birth Defect Description
NBDPS Eligible
Trisomy 21/Down syndrome with
Major
Yes
No
1003
Hirschsprung's disease, NOS
Major
No
No
1003
Simian crease / Transverse palm
Minor
No
Bilateral simian crease
No
1003
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

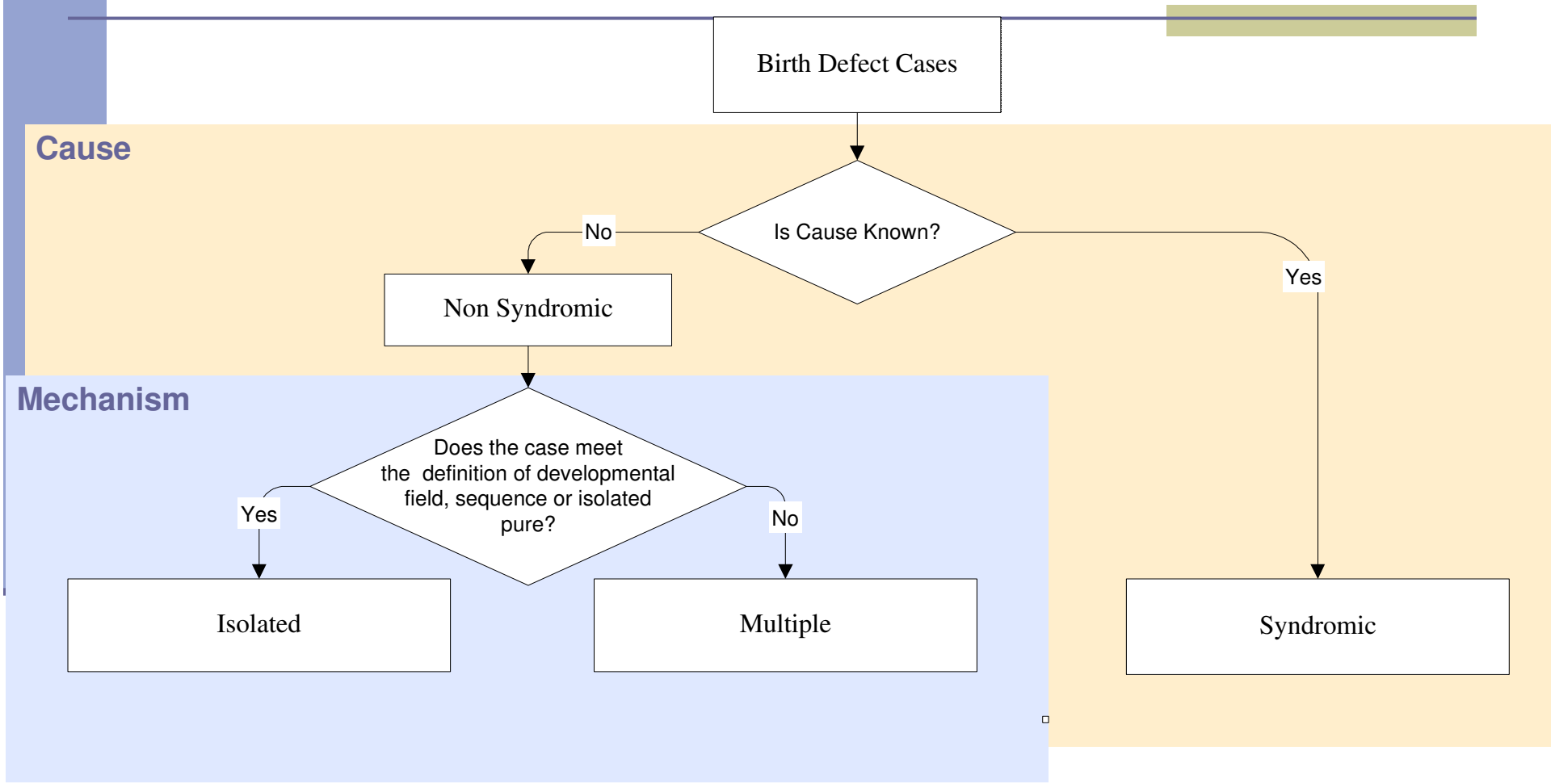
Classification of Birth Defects

- 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

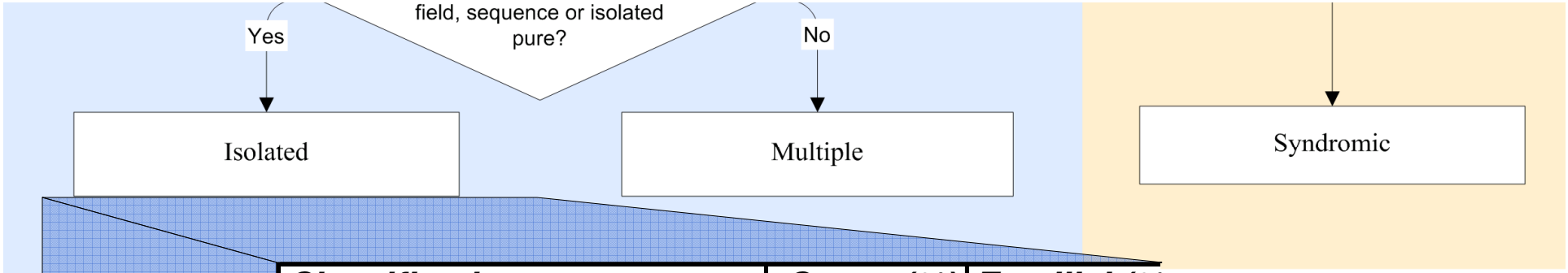
- Clinical geneticists consider:
  - mechanism = pure defects, sequences, developmental field defects
  - cause = chromosome abnormalities, genetic conditions, teratogens, in-utero events
  - family history = first degree relative with same defect
  - morphology = descriptive, anatomical (e.g., oral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

Classification tool in action

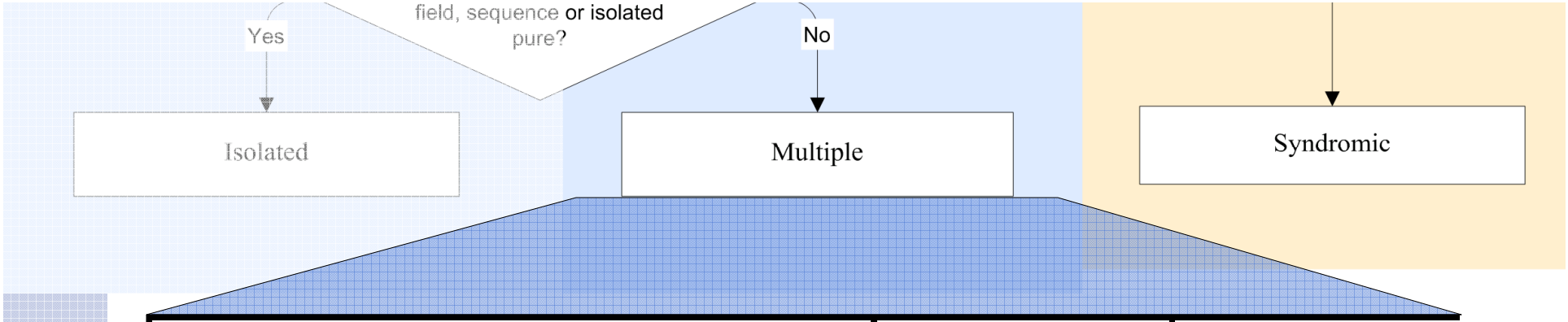


SURVEILLANCE
Classification Tool in Action

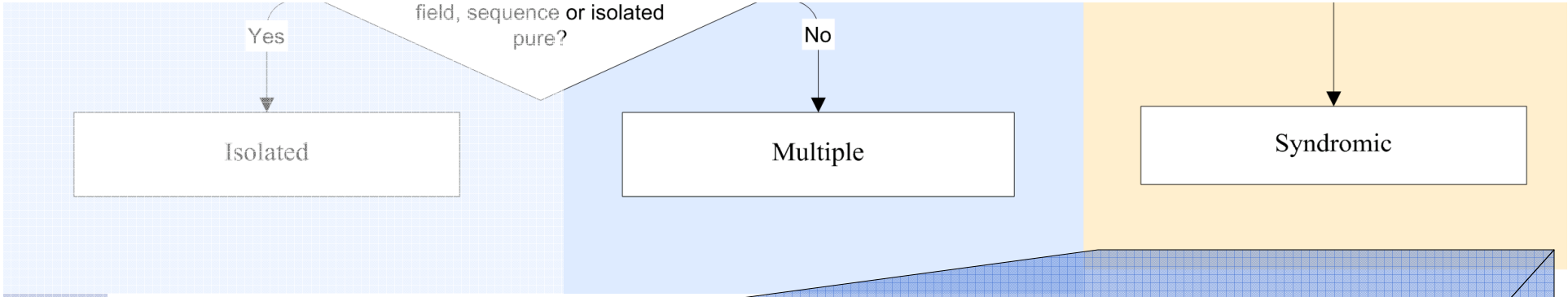
	1994-2006		1999-2006	
Classification	**Frequency (%)**	**Prevalence**	**Frequency (%)**	**Prevalence**
Isolated	6884(69.4)	1 in 88	6281(72.1)	1 in 63
Multiple	1041 (10.5)	1 in 585	882 (10.1)	1 in 450
Syndromic	1997 (20.1)	1 in 304	1544 (17.7)	1 in 258
Total	**9922**	**1 in 61**	**8707**	**1 in 46**



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



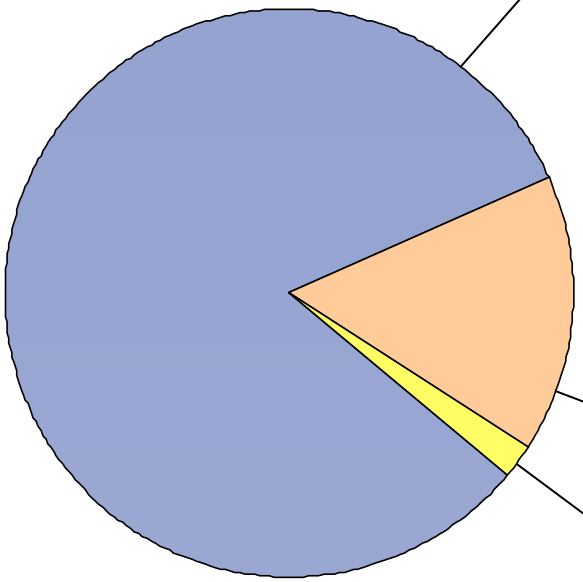
Classification	*Cases (%)*		*Familial (%*	
Multiple				
2+ Majors	708	*68.0*	17	*2.4*
1Major/Minors	194	*18.6*	8	*4.1*
Provisionally Unique	95	*9.1*	13	*13.7*
Association	24	*2.3*		
Recognizable Pattern	20	*1.9*	1	*5.0*



Classification	*Cases (%)*		*Familial (%*	
Syndromic				
Chromosomal	1642	*82.2*	31	*1.9*
Genetic	315	*15.8*	96	*30.5*
Teratogen	40	*2.0*	3	*7.5*

Syndromic cases

(Known cases)



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

Genetic 15.8%:	*Cases (%)*	
Total # of cases	**315**	***(1.0)***

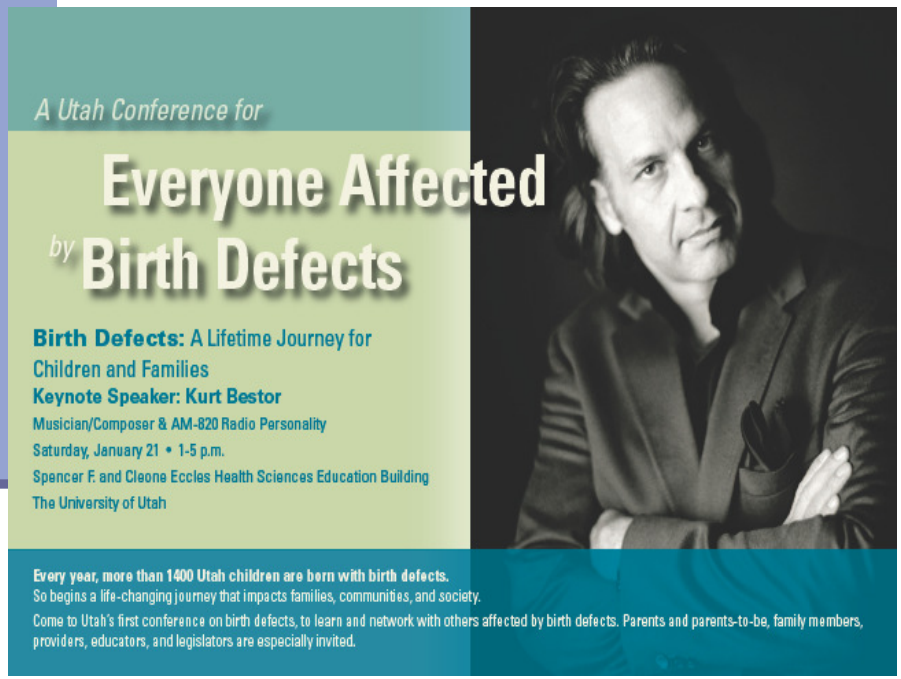
Teratogen 2.0%:	*Cases (%)*	
IDDM	20	*(50.0)*
Cytomegalovirus	6	*(15.0)*
Valproic Acid	11	*(27.5)*
Accutane	1	*(2.5)*
SLE	1	*(2.5)*
Varicella	1	*(2.5)*
Total # of cases	**40**	

PREVENTION
UBDN Web Site

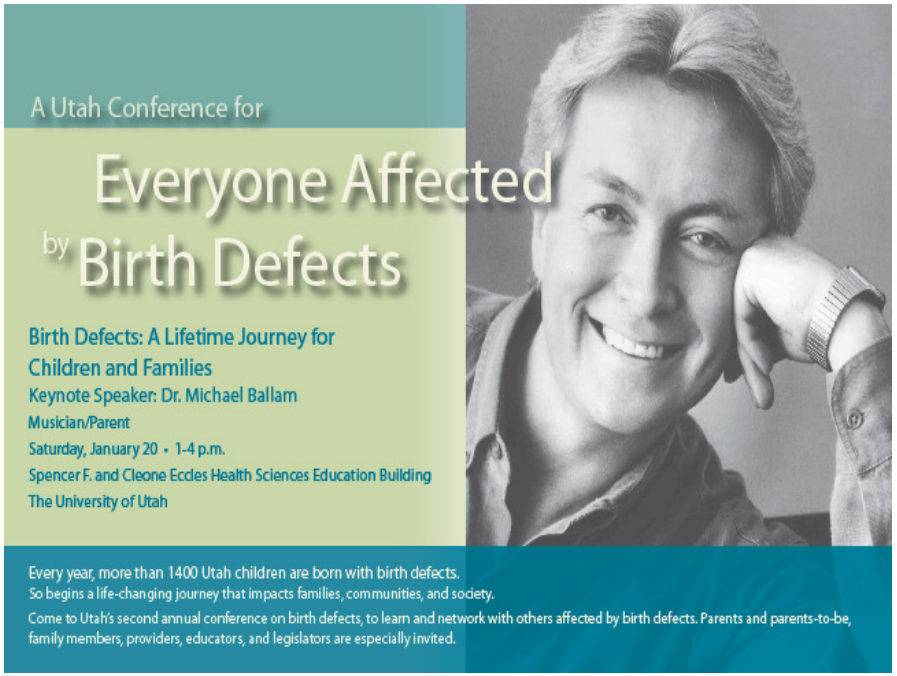
www.health.utah.gov/birthdefect

PREVENTION
Outreach: Family Meetings

2006



2007



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 thing 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!

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.

Acknowledgments

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

- **UBDN Group**
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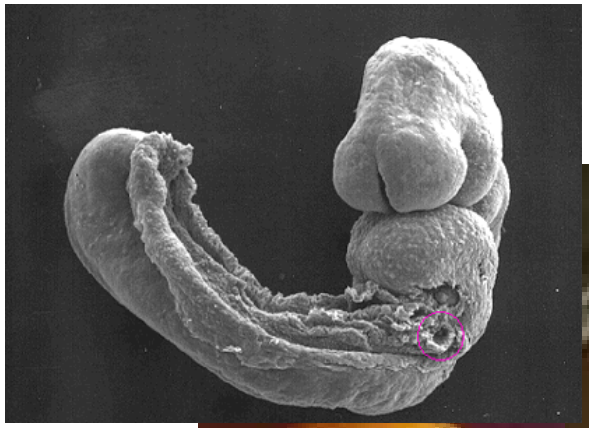

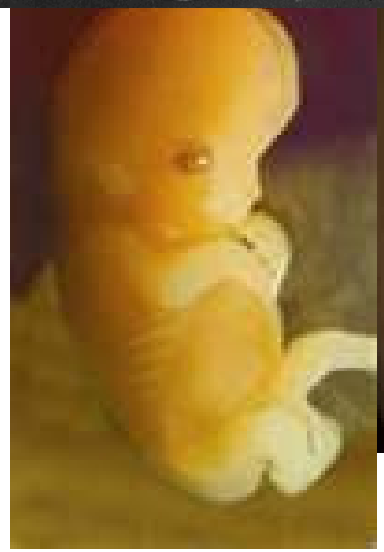

Thank You!