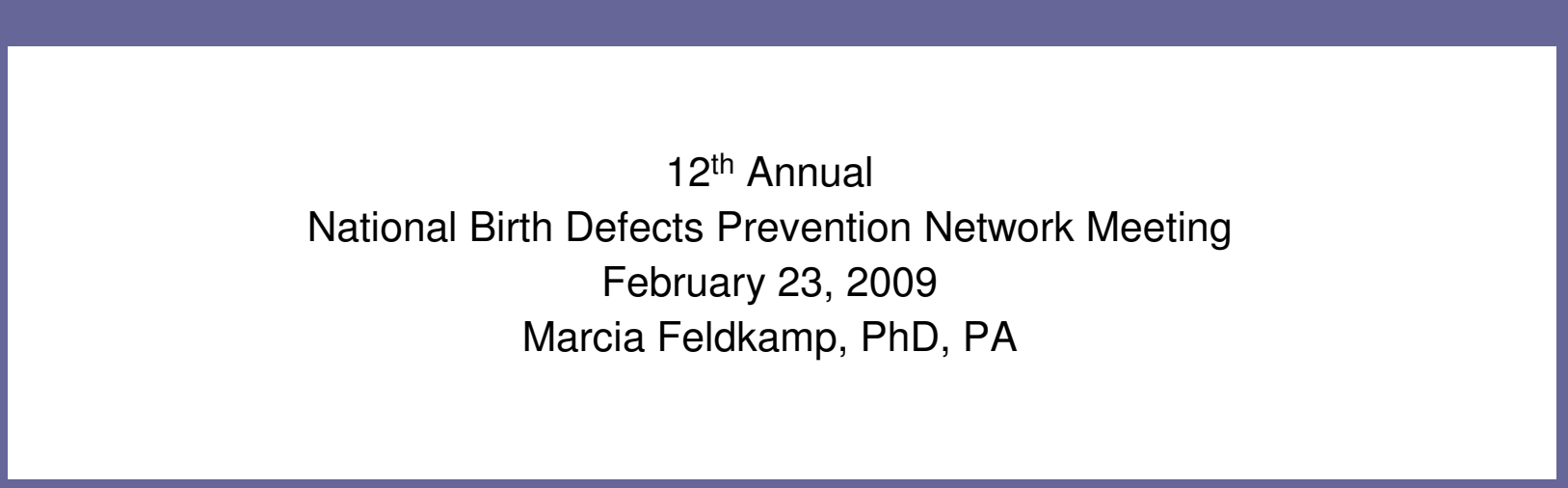




Evolution of the Utah Birth Defect Network

Different Strokes:

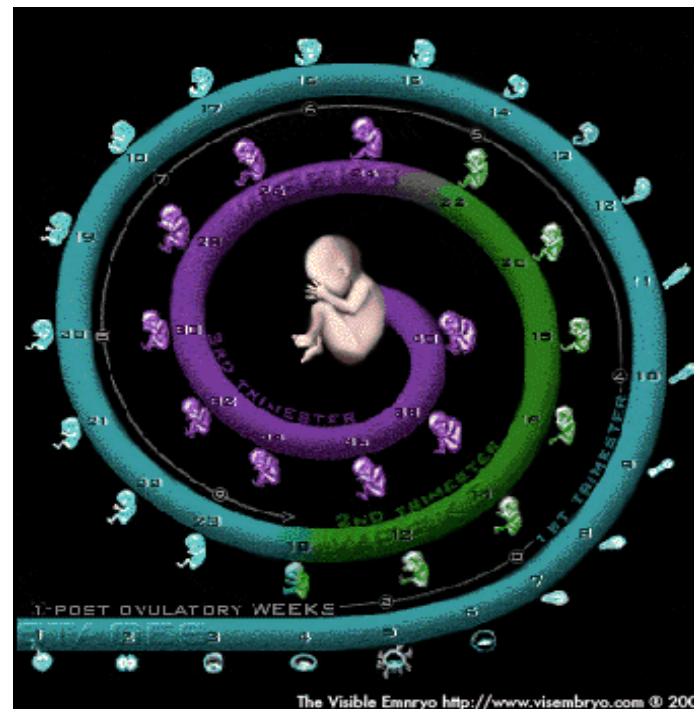
How the Mission and Objectives of a Birth Defect
Program Shapes its Data Collection and Uses



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

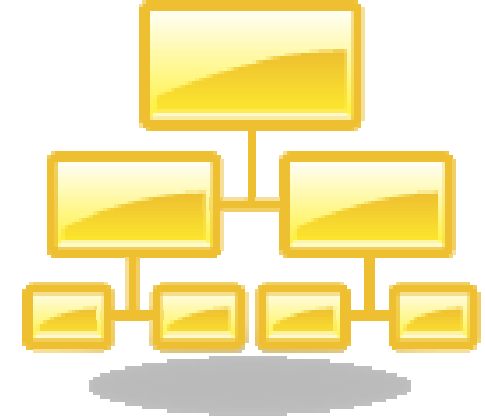
The Utah Birth Defect Network seeks to prevent birth defects and secondary disabilities through public health surveillance, outreach to families and health care providers, and epidemiologic studies.

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

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

Transferred to CDC

-
-
-
-
-
-
-
-
-
-

Birth Defect Code	<input type="button" value="Check Codes"/>	Birth Defect	Prenatally Diagnosed	Birth Defect Description	NBDPS Eligible	
<input type="button" value="▶"/> Trisomy 21/Down syndrome with <input type="button" value="v"/>		Major <input type="button" value="v"/>	Yes <input type="button" value="v"/>		No <input type="button" value="v"/>	1003
<input type="button" value="▶"/> Hirschsprung's disease, NOS <input type="button" value="v"/>		Major <input type="button" value="v"/>	No <input type="button" value="v"/>		No <input type="button" value="v"/>	1003
<input type="button" value="▶"/> Simian crease / Transverse palm <input type="button" value="v"/>		Minor <input type="button" value="v"/>	No <input type="button" value="v"/>	Bilateral simian crease	No <input type="button" value="v"/>	1003
<input type="button" value="*"/> <input type="button" value="v"/>		<input type="button" value="v"/>	<input type="button" value="v"/>		<input type="button" value="v"/>	<input type="button" value="v"/>

Classification Familial Etiology Known

Birth Defect Category	
<input type="button" value="▶"/> Gastrointestinal <input type="button" value="v"/>	1003
<input type="button" value="▶"/> Trisomy 21 <input type="button" value="v"/>	1003
<input type="button" value="*"/> <input type="button" value="v"/>	<input type="button" value="v"/>

Clinical Case Review Comments

Clinical Case Reviewer

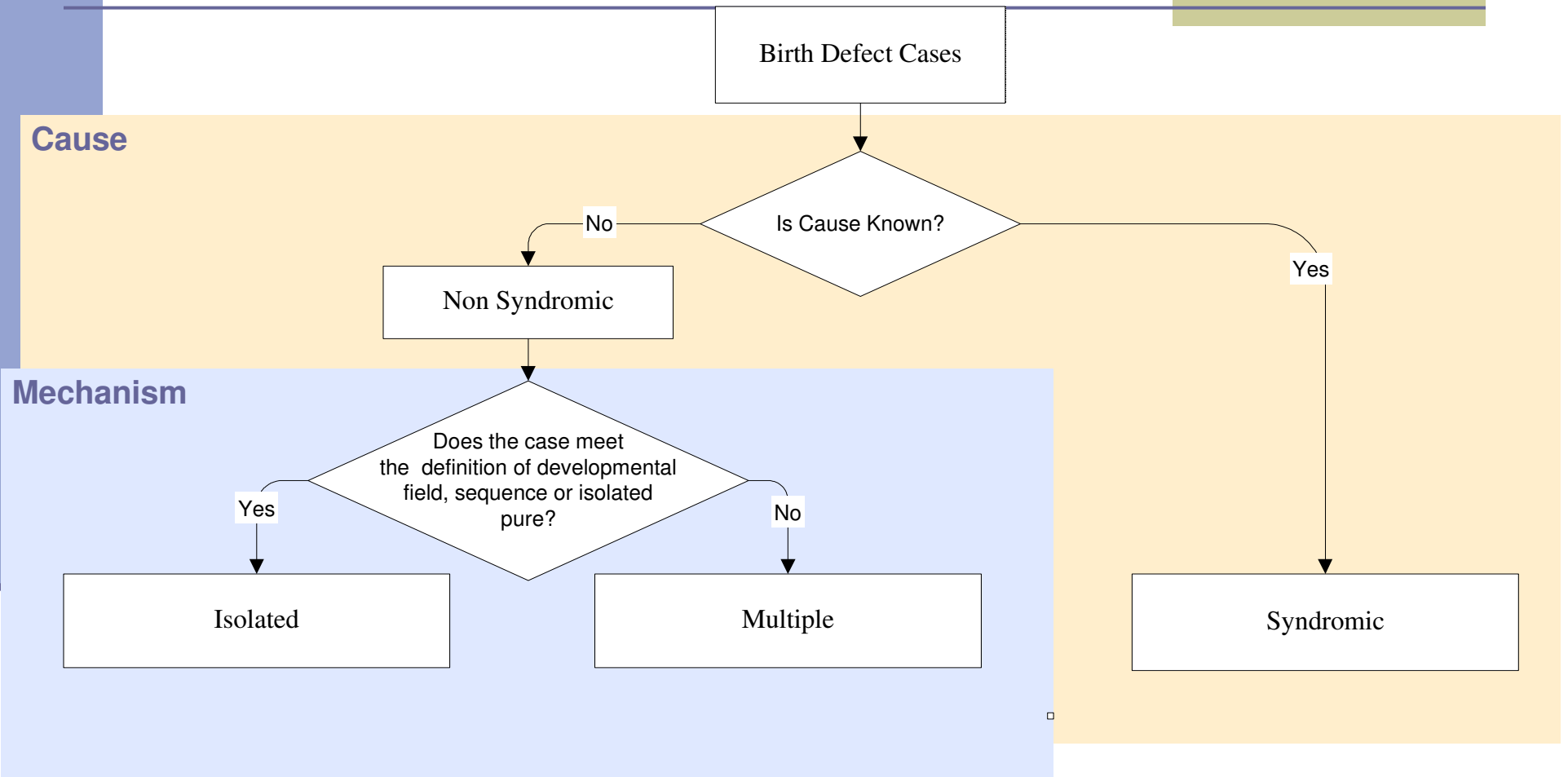
Classification of Birth Defects

- 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)

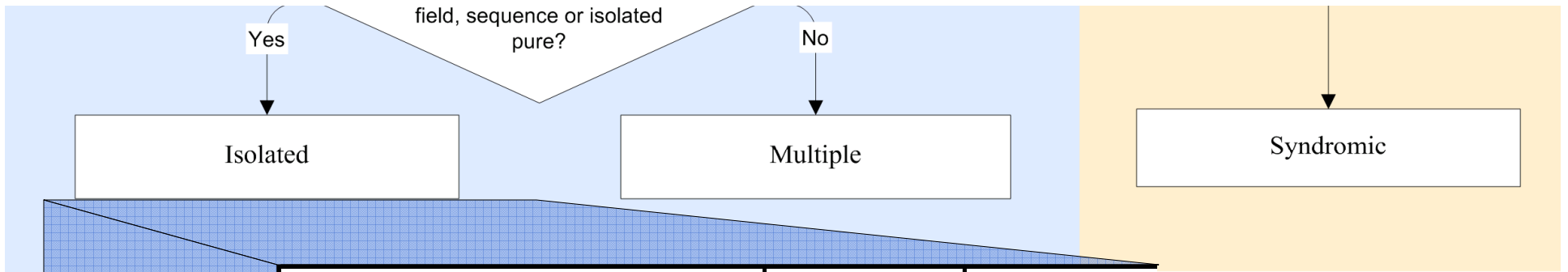
Classification tool in action



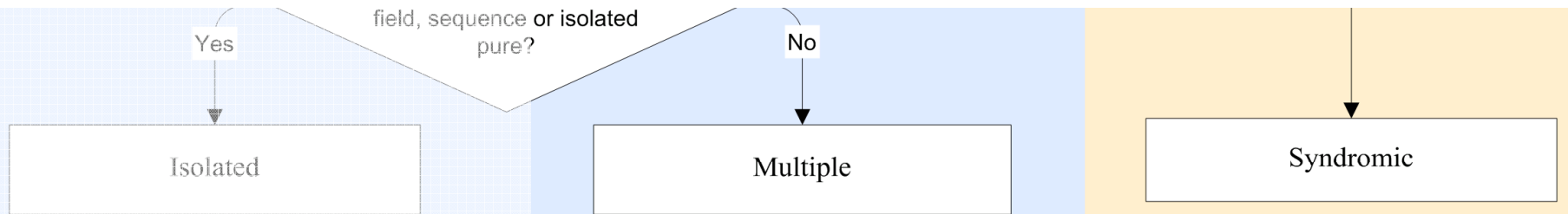
SURVEILLANCE

Classification Tool in Action

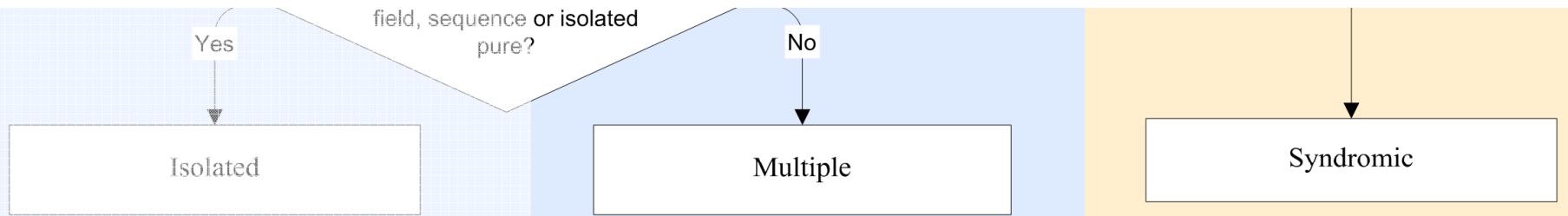
Classification	1994-2006		1999-2006	
	Frequency (%)	Prevalence	Frequency (%)	Prevalence
Isolated	6884(69.4)	1 in 88	6281(72.1)	1 in 63
Multiple	1041 (10.5)	1 in 585	882 (10.1)	1 in 450
Syndromic	1997 (20.1)	1 in 304	1544 (17.7)	1 in 258
Total	9922	1 in 61	8707	1 in 46



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



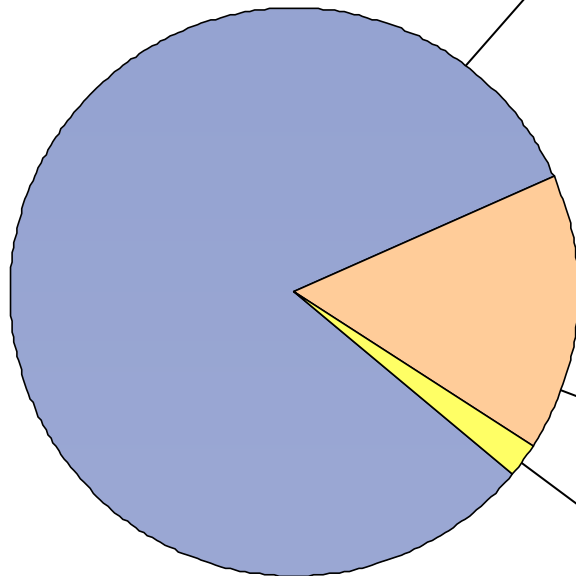
<i>Classification</i>	<i>Cases (%)</i>		<i>Familial (%)</i>	
Multiple				
2+ Majors	708	68.0	17	2.4
1 Major/Minors	194	18.6	8	4.1
Provisionally Unique Association	95	9.1	13	13.7
Recognizable Pattern	24	2.3		
	20	1.9	1	5.0



<i>Classification</i>	<i>Cases (%)</i>		<i>Familial (%)</i>	
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)



<i>Chromosomal 82.2%:</i>	<i>Cases (%)</i>	
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	

<i>Genetic 15.8%:</i>	<i>Cases (%)</i>	
Total # of cases	315	(1.0)

<i>Teratogen 2.0%:</i>	<i>Cases (%)</i>	
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

The screenshot shows the Utah Birth Defect Network (UBDN) website. At the top, there is a navigation bar with links for "Online Services", "Agency List", and "Business". A search box with "Google™ Custom Search" and a "Search" button is also present. Below this is the "Utah Department of Health" logo and a secondary navigation bar with links for "News", "A to Z Index", "Health Data", "FAQs", "Training", and "Local Health". The main header features the "Utah Birth Defect Network" logo on the left and a large photograph of children playing with balloons on the right. The tagline "Striving to Prevent Birth Defects" is centered below the main header. A dark blue navigation bar contains links for "Defect Information", "Prevention", "Services", "Providers and Members", and "About UBDN". On the left side, there is a "Search UBDN:" box with a "Go" button and a vertical menu with links for "Monitoring", "What Causes Birth Defects?", "Studies", and "Cost and Impact". The main content area features a "Spotlight article" titled "Improving a child's lifelong health by preventing alcohol-related birth defects:" with a small image of hands. Below the title is a bulleted list of key points. At the bottom of the article is a link to "Click here to read full article". On the right side, there are three boxes with links to "March of Dimes", "CDC: National Center on Birth Defects and Developmental Disabilities", and "Centers for Disease Control and Prevention: MMWR".

utah.gov Online Services Agency List Business Google™ Custom Search Search

Utah Department of Health health.utah.gov | News | A to Z Index | Health Data | FAQs | Training | Local Health

Utah Birth Defect Network
Striving to Prevent Birth Defects

Defect Information | Prevention | Services | Providers and Members | About UBDN

Search UBDN: Go

Monitoring
What Causes Birth Defects?
Studies
Cost and Impact

Spotlight article
Improving a child's lifelong health by preventing alcohol-related birth defects:

- When a pregnant woman drinks alcohol, so does her baby, and the consequences may be devastating.
- According to the Centers for Disease Control and Prevention, fetal alcohol syndrome is one of the leading preventable causes of birth defects and developmental disabilities.
- The effects of alcohol may cause a recognizable pattern of physical and neurological findings called fetal alcohol syndrome (FAS).
- FAS does not occur only in children of mothers who are heavy drinkers: anyone who drinks wine, wine coolers, beer or liquor during pregnancy may put her baby at risk.
- There is no known safe amount of alcohol during pregnancy.
- The effects of alcohol on the developing baby are 100% preventable.

[Click here to read full article](#)

March of Dimes
CDC: National Center on Birth Defects and Developmental Disabilities
Centers for Disease Control and Prevention: MMWR

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 & AM-820 Radio Personality
Saturday, January 21 • 1-5 p.m.
Spencer F. and Cleone Eccles Health Sciences Education Building
The University of Utah



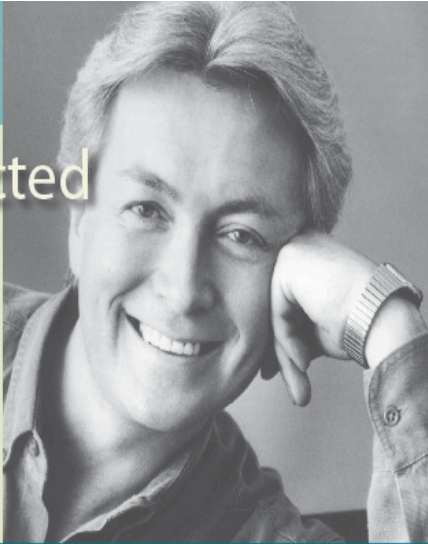
Every year, more than 1400 Utah children are born with birth defects. So begins a life-changing journey that impacts families, communities, and society. Come to Utah's first conference on birth defects, to learn and network with others affected by birth defects. Parents and parents-to-be, family members, 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 Cleone Eccles Health Sciences Education Building
The University of Utah



Every year, more than 1400 Utah children are born with birth defects. So 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 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

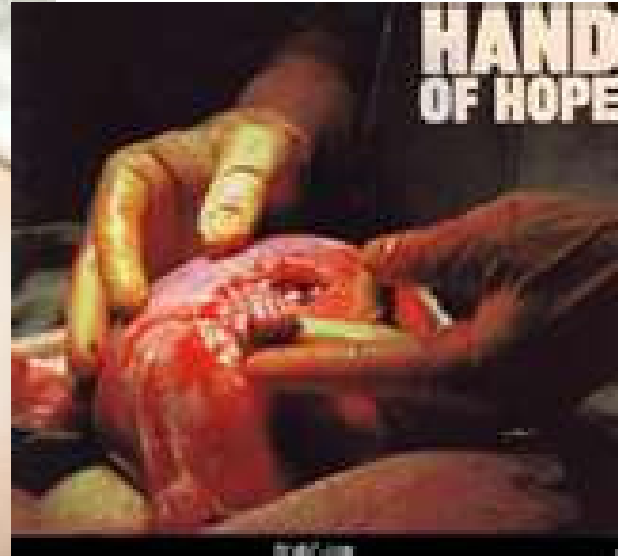
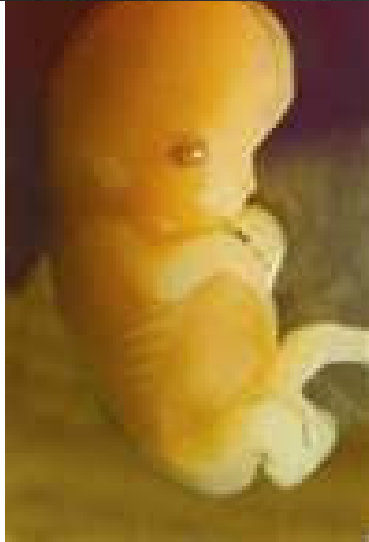
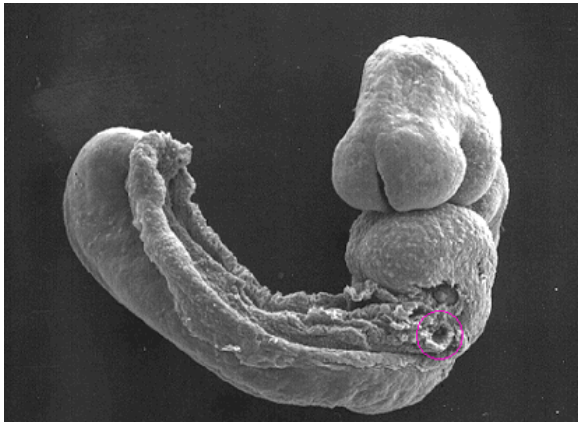
The Utah Birth Defect Network seeks to prevent birth defects and secondary disabilities through public health surveillance, outreach to families and health care providers, and epidemiologic studies.

Acknowledgments

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

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





Thank You!