Birth Defects - Data Availability & Other Factors Relevant to Environmental Public Health Tracking: Report Summary

Center for Excellence in Environmental Public Health Tracking, School of Public Health, University of California Berkeley

Sharan L Campleman*, PhD MPH CTR, Amy D. Kyle, PhD MPH, John Balmes, MD

*Contact information: scamplem@berkeley.edu

INTRODUCTION

The environmental public health tracking initiative is working to build capacity to track environmental factors and environmentally mediated disease. The ultimate goal is to use such information to reduce risks and disease. Birth defects have been identified by the western states affiliated with the Berkeley Center for Environmental Public Health Tracking and by the Centers for Disease Control and Prevention (CDC) as an outcome of interest for environmental public health tracking. In response, the Berkeley Center has prepared a report intended to provide information about birth defects and about birth defects data that is relevant to this initiative and to other projects seeking to better understand birth defects, environmental factors that may contribute to them, and how to reduce them.

OBJECTIVES

The goals of this report were to:

(1) Review and summarize the current status of birth defects surveillance in the United States including the range of outcomes included and data sources and methods applied to estimating the burden on the population; and,

(2) Characterize the current surveillance, vital statistics, administrative, and survey data potentially applicable to environmental public health tracking efforts.

Table A outlines the major sections of the report ***"Birth Defects Data Availability & Other Factors Relevant to Environmental Public Health Tracking"***. This document focuses on the birth defects traditionally monitored by surveillance systems, which are the physical malformations and a limited number of chromosomal abnormalities. These birth defects are often considered 'major' congenital conditions that require medical or surgical intervention, with a serious adverse impact on the child's health, development or cosmetic appearance. However, discussion of the approaches and limitations in these various birth defect surveillance systems may be applicable to the characterization of other adverse developmental, neurodevelopmental, sensory or metabolic-related birth outcomes.

Table A

Birth Defects Data Availability & Other Factors Relevant to Environmental Public Health Tracking

1.0 Summary
2.0 Introduction
3.0 Magnitude of Disease Occurrence
- 3.1 Morbidity
- 3.2 Mortality
- 3.3 Economic Costs

4.0 Birth Defect Surveillance & Monitoring
- 4.1 Current Status
- 4.2 Surveillance Methods
- 4.3 Sources of Birth Defects Data – Availability, Limitations & Quality Issues

4.4 Data Linkage to Pool Multiple Sources
- 4.5 Classification Schemes for Birth Defects Reporting
- 4.6 Case Definitions for Reporting

5.0 Summary of Current Knowledge on Risk Factors
- 5.1 Maternal & Birth-Related Risk Factors
- 5.2 Summary Evidence of Environmental Etiology

6.0 Summary of Health Disparities
- 6.1 Variation in Population by Demographics
- 6.2 Geographic Distribution
- 6.3 Temporal Trends

7.0 Data Analyses & Interpretation – Applicability to EPHT
- 7.1 Descriptive Analyses
- 7.2 Temporal Trends
- 7.3 Spatial & Environmental Associations

8.0 References
9.0 Glossary of Terms
10.0 Appendices

METHODS

Data and other information summarized from the peer-reviewed literature, published governmental reports, non-governmental organizations, and data generating agencies.

RESULTS

Several issues in birth defects monitoring may influence its application to EPHT and require sufficient understanding before applying and interpreting such outcomes in regards to their potential environmentally mediated determinants. A few examples of multiple issues discussed in greater detail include:

(1) The completeness and accuracy of birth defects reporting for specific defects by surveillance method influence the selection of defect types to track or link with exposure data.

(2) The relative completeness of individual data items varies substantially by data source and thereby between surveillance programs.

(3) The availability and utilization of prenatal diagnosis and elective termination vary among populations, across geographic regions and over time, influencing the relative reporting of several defects.

(4) The etiologically heterogeneous nature of birth defects influences the degree to which birth defects should be grouped; although the relatively small numbers of specific, individual defects limits the analytical power to investigate potential environmental associations.

(5) The availability of the legislative authority allowing reporting, the extent of data, and the linkage and dissemination of data within and between state databases varies the accessible level of the identifier, the geographic unit, and the population at risk.

Tables B-D summarize the characteristics of the general types of data sources available for birth defects surveillance and the potential positive/negative influence of EPHT-related analyses.

Table B

General Types of Data Sources Used in Birth Defects Surveillance for Case-Finding and/or Case Ascertainment

General Type of Data Source	Type Surveillance			Age of Child at Diagnosis/Ascertainment			
	Case Finding	Case Data	Linkage Add Data	Prenatal Dx	Neonatal Period	Infant <1	Older Child
PRIMARY REPORTING FACILITIES *							
Birth Hospitals	A	A/P[1]					
Pediatric and Tertiary Care Centers		A/P[1]			X	X	X
SPECIALTY CLINICAL SOURCES							
Prenatal Diagnosis (Ultrasound, Maternal Serum Screening)	A	A/P[1]		X			
Cytogenetic Laboratories	A	A/P[1]		X	X	X	X
Genetic Counseling/Screening	A	A/P[1]		X			
Other Specialty Clinics (Heart Ultrasound)	A	A/P[1]				X	X
OTHER SPECIALTY PROGRAMS							
Special Needs Childrens Programs	A	P	A/P			X	X
FAS Support Programs	A	P	A/P			X	X
VITAL RECORDS							
Birth Certificates	P	P	A/P				
Deaths Certificates		P	A/P			X	X
Fetal Death Reports	P	P	A/P		X	X	
Linked Birth-Death files		P	A/P			X	
ADMINISTRATIVE DATABASES (SECONDARY)							
Hospital Discharge (Out/In Patient)		P	P		X	X	X
Medicaid Utilization File		P	P			X	X

*Potential in facility sources: chart review, disease or discharge index, discharge summaries, obstetrics logs, nursery logs, newborn intensive care logs, postmortem/pathology logs.
A=Active Surveillance P=Passive Surveillance
[1] Availability of data for passive surveillance by source varies by legislatively mandated reporting requirements and supporting code.

Table C

General Characteristics of Secondary Data Sources Relevant to Birth Defect Surveillance

VITAL STATISTICS DATA FILES

- \+ Standardized, continuous data collection for a defined population.
- \+ Basic demographics that are often not otherwise available on infant/child and mother.
- \+ Provides limited data on some important maternal and infant characteristics (e.g. birth weight).
- \+ Provide demographic data including residential address at birth.
- \+ Able to manipulate to review differences in population level trends.
- \+ Well established.
- \+ Documentation of instrument changes well documented by authoritative body (National Center for Health Statistics).
- \+ Linkage potential at personal (maternal SS#, name, DOB) and geographic level (address, zip code, county, state).

- \- Adverse birth outcome variables limited and under reported.
- \- Minimal quality control.
- \- Residence at birth not equal to residence at conception (issue for interpretation of exposure associations).

ADMINISTRATIVE DATABASES

- \+ Linkage potential at personal (child SS#, maternal SS#, name, DOB) and geographic level (address, zip code, county, state).
- \+ Often provides relevant clinical data on infant/child not in maternal birth record.
- \+ Able to identify cases/conditions diagnosed post-newborn period.

- \- Diagnostic coding not designed for disease registration.
- \- Variable in population coverage (+ universal coverage).
- \- Denominator for estimate of completeness not always available (hospital, HMO, or geographic level).
- \- Identifies number of contacts not number of cases for a given period.
- \- Requires unique identifiers for infant/child/mother and additional manipulation for case/record consolidation.
- \- Variable content and quality in demographic and other data (non-economic variables).
- \- Minimal quality control in variable of greatest interest to registry (demographics, disease coding).
- \- Limited documentation of coding changes over time (historical documentation of case definitions, codes, or procedures).

Table D

Summary Characteristics of Case Ascertainment Method vs. EPHT-Related/Initiated Analyses

	Time Trends	Spatial Variation	Cluster Analysis	Research
ACTIVE				
+ Near complete case ascertainment	x	x	x	
+ Improved timeliness for data collection			x	x
+ High data quality	x	x	x	x
+ Ability for case follow up /RCA			x	x
+ Availability additional demographic & other risk factors data	x	x	x	x
-Labor intensive	x	x	x	
-High resource/costs	x	x	x	
-Often limited population covered		x	x	
PASSIVE				
+ Lower cost	x	x	x	
+ Larger geographic coverage	x	x	x	
+ Availability additional demographic (linkage)	x	x	x	x
-Lack of diagnostic specificity	x	x	x	x
-Minimal control over timeliness of data submission			x	x
-Minimal control over accuracy and data completeness				x
-Inability or limited ability to track cases for follow-up				x
-Limited availability additional risk factor data				x

Tables E and F illustrate examples of both the current wide variation in prevalence of major birth defects by geography (state), ascertainment method (within active surveillance programs) and case definition (inclusion by birth outcome/gestational age).

Table E

Range of the Estimated Prevalence for Major Birth Defects by Race/Ethnicity in Selected States* With Population-Based Active Surveillance, 1996-2000

Organ System Selected Defect	Non-Hispanic White	Non-Hispanic Black	Hispanic	Asian/PI	AI/AN
CENTRAL NERVOUS SYSTEM					
Anencephaly	0.42-4.63 (8)	0.41-3.08 (3)	0.54-3.96 (6)	** (3)	**
Spina bifida w/out anencephalus	2.01-6.04 (8)	2.27-3.76 (4)	3.26-5.58 (7)	1.33-4.30 (5)	**
Hydrocephalus w/out spina bifida	3.67-4.72 (8)	5.26-10.33 (5)	3.07-9.30 (6)	5.60-9.76	**
CARDIOVASCULAR					
Transposition of great arteries	3.95-5.94 (8)	2.77-5.61 (4)	2.76-6.51 (8)	2.98-7.25 (3)	**
Tetralogy of Fallot	2.31-4.41 (8)	2.64-6.71 (4)	2.43-3.45 (6)	3.74-4.41 (3)	**
Ventricular septal defect	13.44-42.12 (8)	17.28-50.54 (6)	11.82-60.27 (8)	9.52-44.52 (7)	12.92-50.56 (5)
Atrial septal defect	18.04-48.32 (8)	20.29-47.48 (6)	13.95-45.1 (8)	13.06-44.25 (6)	16.11-61.25 (5)
Pulmonary valve atresia & stenosis	0.94-10.94 (7)	6.42-16.41 (4)	0.93-10.97 (6)	4.14-8.15 (5)	**
Coarctation of aorta	4.36-9.69 (8)	2.99-11.23 (5)	2.95-6.56 (6)	1.49-6.99 (4)	**
ALIMENTARY TRACT					
Cleft lip w/ & w/out cleft palate	7.56-14.29 (8)	4.13-17.33 (7)	5.90-32.88 (8)	7.74-15.06 (6)	0.69-24.44 (3)
Cleft palate w/out cleft lip	5.46-8.72 (8)	3.72-5.17 (4)	4.20-6.93 (6)	5.72-9.32 (5)	**
Esophageal atresia/tracheosophageal fistula	1.68-5.35 (8)	1.57-1.87 (3)	1.00-2.94 (4)	**	**
Rectal & large intestinal atresia/stenosis	2.41-5.45 (8)	2.81-5.22 (4)	2.67-6.82 (4)	2.67-6.12 (3)	**
GENITOURINARY TRACT					
Renal agenesis/hypoplasia	3.37-7.02 (7)	2.56-5.97 (4)	0.47-5.21 (5)	**	**
Hypospadias & epispadias	2.62-49.97 (8)	17.33-52.97 (6)	2.00-43.84 (8)	18.63-29.13 (6)	**
MUSCULOSKELETAL					
Reduction deformity, lower limbs	1.11-2.41 (7)	1.49-3.78 (4)	0.96-4.47 (5)	**	**
Reduction deformity, upper limbs	2.18-6.02 (6)	1.40-5.97 (4)	1.80-4.85 (7)	**	**
Omphalocele	1.01-3.78 (8)	1.74-2.78 (3)	0.80-2.68 (4)	0.71-3.14 (3)	**
Gastroschisis	1.27-4.72 (8)	1.20-13.10 (5)	1.30-5.35 (6)	**	**
CHROMOSOMAL ABNORMALITY					
Down Syndrome	9.13-21.6 (8)	7.27-11.94 (6)	9.27-13.86 (7)	7.15-17.14 (6)	**

*Selected states with population-based active surveillance systems, 1996-2000: California, Colorado, Georgia, Texas, Hawaii, Iowa, New York, Utah
Prevalence presented only for registries reporting rates based on > 5 cases in five-year period. (#)=Number of registries reporting rate > 5 cases.
Prevalence per 10,000 live births based on mutually exclusive race/ethnicity categories.
**= < 3 Registries reporting (rate not presented).
Asian/PI=Asian/Pacific Islander
AI/AN=American Indian/Alaskan Native
Source: Birth Defects Research (Part A) 67:729-818 (2003)

Table F

Estimates of the Distribution of Birth Defects by Pregnancy Outcome: Data from the Texas Birth Defects Monitoring Division

Organ System Selected Defect	Percentage of Cases in Registry				
	Life Birth Any GA	Fetal Death ≥20 wk GA	Elective Term <20 wk GA	Elective Term ≥20 wk GA	Unspecified Any GA
CENTRAL NERVOUS SYSTEM					
Anencephaly	27.0	27.0	22.0	21.0	3.0
Spina bifida w/out Anencephalus	75.0	5.0	11.0	9.0	0.0
Encephalocele	57.0	6.0	17.0	14.0	6.0
RESPIRATORY SYSTEM					
Agenesis, Aplasia, or Hyperplasia of the Lung	78.0	12.0	2.0	7.0	1.0
GENITOURINARY TRACT					
Renal agenesis/hypoplasia	80.0	10.0	2.0	8.0	0.0
MUSCULOSKELETAL					
Omphalocele	60.0	9.0	13.0	15.0	3.0
Gastroschisis	88.0	6.0	5.0	1.0	0.0
CHROMOSOMAL ABNORMALITY					
Down syndrome	84.0	2.0	6.0	7.0	1.0
Trisomy 13	53.0	9.0	16.0	16.0	6.0
Trisomy 18	61.0	9.0	10.0	18.0	2.0
TOTAL BIRTH DEFECTS	94.4	1.9	1.3	2.1	0.3

Texas active surveillance system, 1996-1997 cases.
GA=Gestational Age Elective **Term**=Elective Termination **Unspecified**=unspecified abortion (termination vs. fetal death).
Source: Texas Birth Defects Monitoring Division (2002)

Tables G and H summarize some basic questions that EPHT programs will need to address to understand the limitations and applicability of the available birth defects data.

Table G

Summary of Potential Questions on Data Content and Quality Influencing the Application of Birth Defects Surveillance to EPHT

1) **DEFINITION, SELECTION AND VARIATION IN CATEGORIZATION OF BIRTH DEFECTS**
Terminology, standardization, classification and coding.
Changes in definitions and completeness of diagnosis (prenatal to infant) over time or area.
Multiple abnormalities or syndromes.
Separate syndromic versus non-syndromic cases for specific type of defects.

2) **DEFINITION OF TYPE OF FETUS/BIRTH OUTCOME**
Spontaneous abortion, terminated pregnancies, stillbirths, live births.
Gestational age descriptors and method of assignment.
Definition of what is or is not included.

3) **DEFINITION OF REGISTRATION PERIODS**
Early prenatal period, prenatal, neonatal, post neonatal, infant, or child.
Ability to link with other data sources (vital stats, hospital discharge).

4) **DEFINITION OF REGISTRATION BASE**
Hospital versus population.
Temporal & geographic boundaries.
Changes in boundaries, population or surveillance methods in time.

5) **ASCERTAINMENT METHODS**
Active case finding and/or ascertainment.
Passive reporting with or without follow up, audits or active component.
Legislative authority/mandate for reporting requirements including presence/absence or changes over time.
Multiple sources.

6) **VARIABLES AVAILABLE**
Demographic and geographic.
Unique identifiers for linking (maternal, infant, child).
Other risk factors (reproductive history, family history, occupations, etc).

Table H

Summary of Potential Questions on Utilizing Birth Defects Surveillance in Descriptive Temporal, Spatial or Environmental Analyses Relevant for EPHT

1) **SPECIFICITY OF DIAGNOSIS**
Case ascertainment, methods or diagnostic criteria vary over time influence trends and determination 'baseline' prevalence.

2) **STANDARDIZATION OF DIAGNOSTIC DETAIL OVER TIME**
Consistent and comparable for trends.
Consistent and comparable for between area comparisons.

3) **SMALL NUMBERS OVER TIME & PLACE.**
Affect stability of annual rates (grouped year rates) for area.
Relative power for geographic comparisons (e.g. observed to expected).

4) **COMPLETENESS**
Relative completeness vary with changes in surveillance methods influence ability to identify actual trends, excess, or associations.

5) **TIMELINESS**
Diagnosis to ascertainment/registration
Relevance to 'cluster' analysis or other immediate concerns

6) **SPATIAL REFERENCE**
Residence at diagnosis, residence at birth, residence at conception

7) **COMMON RISK FACTORS / DISPARITY MEASURES**
Smoking, alcohol, occupation, maternal age
SES, education, race/ethnicity

8) **AVAILABILITY OF LINKAGE VARIABLES**
Unique personal identifiers or geographic identifiers
Link on infant, child or mother.

9) **DEMOGRAPHIC DETAIL**
Available for geographic or social units of interest
Live births, or live births and fetal deaths, by geographic entity (census tract, zip code, county) with age, sex, race/ethnicity detail.

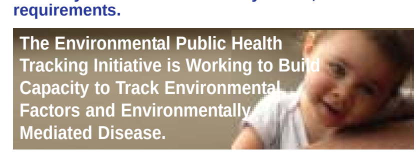
RECOMMENDATIONS/NEXT STEPS

Although the number of specific environmental toxins causally associated with human birth defects may be currently limited, other factors also indicate the prudence of selecting at least a subset of birth defects for environmental public-health tracking including:

(1) The toxicological precedent of a large number of identified animal teratogens;

(2) The potential for population exposure to known animal teratogens with existing exposure data (such as pesticides and heavy metals); and,

(3) The relative availability, and related public expectation for the use, of various levels of birth defect surveillance data.

Unfortunately to date, the selection of any such subset of birth defects may be based more on the technical and financial surveillance considerations, than on biological plausibility or environmental appropriateness.

State programs considering birth defects as a tracking outcome must understand the characteristics of their current data that will influence its uses (i.e. data completeness by defect type, baseline rates, time trends, county or smaller geographic area level assessments, cluster investigations, population-based research), as well as, the additional technical, funding, and legislative requirements to expand identified data elements for utility in future specific EPHT goals. Potential next steps should include a candid discussion between the data providers, including formal birth defect surveillance programs, subject matter experts, and tracking programs to identify/prioritize the specific birth defects most applicable to EPHT with both the currently available data and any future, ideal data requirements.