

# Wisconsin Birth Defects Registry collecting data

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## Abstract

Wisconsin Statute 253.12 was enacted in May 2000 to create the Wisconsin Birth Defects Registry (WBDR), replacing the Birth and Developmental Outcome Monitoring Program, a previous birth defects and developmental disabilities reporting system initiated in 1989. In the summer of 2004, the new registry began collecting demographic, diagnostic, and identifying information for children from birth to 2 years of age who are born with reportable birth defects and/or are receiving health care services for them in Wisconsin. This article describes the development of the registry and outlines expectations for reporting of birth defects.

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## Introduction

In Wisconsin, approximately 2000 infants are born with a birth defect each year, affecting approximately 3% of all births.<sup>1</sup> More than 4500 different birth defects have been identified, and together they cause more than 20% of all infant deaths. Birth defects are a substantial cause of childhood morbidity and long-term disability and are the fifth leading cause of years of potential life lost. Birth defects are also expensive. In Wisconsin, the estimated lifetime cost of only 12 selected birth defects among infants born in a given year is \$141 million.<sup>2</sup>

As many as two-thirds of all birth defects have no identifiable cause. In order to prevent birth defects, we must know more about their causes. Fundamental questions such as how often each type of birth defect occurs and whether they are clustered in certain parts of the state or among certain racial/ethnic groups must be examined. Surveillance is a means to begin identifying, counting, and providing services to babies with birth defects and their families.

## Birth Defects Reporting Legislation

The Wisconsin Legislature supported passage of Wisconsin Act 114 in May 2000 in an effort to address this substantial public health problem. The goal of this law is to improve birth defects reporting in Wisconsin so that this information

can be used to identify birth defect risk factors, conduct epidemiologic studies, decrease the incidence of birth defects, and facilitate service provision to the families of children with birth defects. The law mandates reporting of birth defects by physicians and pediatric specialty clinics. Any hospital in the state may also voluntarily report the occurrence of birth defects in children diagnosed or treated within a given facility.

The law also requires the Department of Health and Family Services (DHFS) to maintain an up-to-date registry of the diagnosed birth defect of any Wisconsin child age birth to 2 years and to develop rules regarding which birth defects must be reported. DHFS must specify how the reporting will be accomplished and must notify the persons required to report. The administrative rules were passed by the Legislature and took effect April 1, 2003.

Statute 253.12 defines a birth defect as a structural deformation, disruption or dysplasia, or a genetic, inherited, or biochemical disease that occurs prior to or at birth and that requires medical or surgical intervention or interferes with normal growth and development. The administrative rules define a list of reportable birth defects (Table 1). This list was developed by the Scientific Advisory Subcommittee of the Council on Birth Defect Prevention and Surveillance. The subcommittee selected reportable

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birth defects based on the following criteria:

- conforms to the statutory definition of a birth defect
- usually identifiable by age 2 years
- has medical, surgical, or developmental significance
- occurs at an estimated birth prevalence of 1 in 30,000 or more
- is likely to be ascertained through assessment in 1 or more specialty clinics

The resulting list was reviewed by approximately 30 pediatric subspecialists for accuracy and relevancy, and then was finalized.

### Registry Development

As the list of reportable birth defects was being developed, the Council also discussed exactly how a birth defects registry would work. Registries in other states were examined, and discussions ensued regarding what information would be most helpful for surveillance in Wisconsin. An electronic registry available on the Internet was deemed most efficient, most accessible, and more confidential than sending paper reports via mail or fax. However, it was also acknowledged that some reporters would be unable or unwilling to use the Internet, so it was decided to develop an electronic registry as well as a paper reporting option.

In August 2003, the registry was ready to be tested by DHFS staff; it was revised following the test period. Four specialty clinics were then asked to test the registry—the University of Wisconsin Waisman Center (Genetics); Children's Hospital of Wisconsin-Milwaukee (Genetics); Gunderson Lutheran Medical Center, Inc. (Pediatric Neurodevelopment Center); and Marshfield Clinic.

Preliminary testing at these 4 sites revealed many issues. A major issue was access to and stability of the test site. To access the test site, each re-

porter had to be an approved user of the Health Alert Network (HAN) and also an approved user of the WBDR. During the development process, Wisconsin implemented an additional layer of security called the Web Access Management System (WAMS), which meant that each new user had to register 3 times and be approved 3 times to gain access to the WBDR. In addition, because changes were frequent, the site was not always available. For busy clinicians, it was frustrating to clear so many hurdles just to be approved to report and still not be able to count on getting into the Web site or completing a report without the site going down.

Another substantial issue was parental consent. Current legislation requires parent approval to submit a complete report. If parent permission is not obtained, the report is still made, but identifying information such as names and addresses of child and parents are not included.

A third issue arose at Marshfield Clinic, which has a state-of-the-art electronic patient records system. They were reluctant to individually key in birth defects reports when the information was already in their electronic patient records system. They requested we investigate the possibility of directly uploading information to the WBDR.

These issues and others made it clear that a short pilot testing phase was insufficient. The pilot sites were asked to stop testing while improvements were made. These improvements included streamlining the 3-stage registration process, stabilizing the Web site, working out the site's navigation issues, narrowing the list of required variables, and making it possible to upload data directly from an electronic patient records system to the WBDR.

The pilot sites resumed testing in January 2004. A recent survey indicated a positive response to the

improvements to the Web site. Three of the 4 sites have successfully reported babies with birth defects via the secure Web site. Marshfield Clinic uploaded its records for the first time at the end of June 2004 and is set up to submit any new reports monthly. The WBDR will go statewide by the end of 2004; training and information sessions were offered at 11 sites statewide.

Some fundamental issues still need to be resolved. For example, the current statute does not allow linkage of WBDR to other databases that may contain important information about infants and children with birth defects, such as the newborn metabolic screening and birth certificate data. However, as legislative work continues to address this issue, WBDR is collecting vital epidemiologic data in Wisconsin.

### Summary

Although not without hurdles, the WBDR development has proceeded from legislation through pilot testing. When the WBDR goes statewide in late 2004 it will be the culmination of several years of intensive work. Beginning in 2005 it will be possible to describe the number and type of birth defects reported annually in Wisconsin and to begin to use these data to provide appropriate services to affected children and their families.

### References

1. Wisconsin Vital Statistics, Bureau of Health Information, Division of Public Health, Wisconsin Department of Health and Family Services.
2. National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Atlanta, GA.

**Table 1.** Wisconsin Birth Defects Registry Reportable Conditions

**CARDIOVASCULAR**

- 100 Atrial Septal Defect
- 101 Atrioventricular Canal/Endocardial Cushion Defect
- 102 Cardiac Arrhythmia (Congenital)
- 103 Coarctation of the Aorta
- 104 Hypoplastic Left Heart
- 105 Tetralogy of Fallot
- 106 Total Anomalous Pulmonary Venous Return
- 107 Transposition of the Great Vessels
- 108 Truncus Arteriosus
- 109 Valvular Heart Disease (Congenital)
- 110 Ventricular Septal Defect

**CHROMOSOMAL**

- 150 Down Syndrome
- 151 Klinefelter Syndrome
- 152 Trisomy 13
- 153 Trisomy 18
- 154 Turner Syndrome
- 155 Velocardiofacial Syndrome (22q Deletion Syndrome)
- 156 Other Chromosomal Anomaly (not Down Syndrome, Klinefelter Syndrome, Trisomy 13, Trisomy 18, Turner Syndrome or Velocardiofacial Syndrome)

**ENDOCRINE**

- 200 Hypothyroidism (Congenital)

**EYE**

- 250 Cataract (Congenital or Early)
- 251 Coloboma
- 252 Glaucoma (Congenital)
- 253 Microphthalmia/Anophthalmia

**GASTROINTESTINAL/ABDOMINAL**

- 300 Biliary Atresia
- 301 Gastroschisis
- 302 Hirschsprung Disease
- 303 Omphalocele
- 304 Pyloric Stenosis
- 305 Rectal/Colonic Atresia/Stenosis
- 306 Small Bowel Atresia/Stenosis
- 307 Tracheo-Esophageal Fistula/Esophageal Atresia

**GENITOURINARY**

- 350 Ambiguous Genitalia
- 351 Epispadias
- 352 Exstrophy of the Bladder/Cloaca
- 353 Hypospadias
- 354 Multicystic and/or Dysplastic Kidney
- 355 Obstructive Urinary Tract Defect (not Posterior Valves; not Urethral Stenosis/Atresia)
- 356 Polycystic Kidney Disease, Autosomal Dominant Form
- 357 Polycystic Kidney Disease, Autosomal Recessive Form
- 358 Polycystic Kidney Disease, Uncertain Form
- 359 Posterior Urethral Valves
- 360 Renal Agenesis/Hypoplasia
- 361 Urethral Stenosis/Atresia

**HEMATOLOGIC**

- 400 Hemophilia
- 401 Hereditary Spherocytosis
- 402 Von Willebrand Disease

**MUSCULOSKELETAL**

- 450 Achondroplasia
- 451 Amniotic Bands
- 452 Arthrogryposis Multiplex Congenita
- 453 Bone Dysplasia/Dwarfism, Other (not Achondroplasia)
- 454 Clubfoot (Congenital)
- 455 Hip Dislocation (Congenital)/Developmental Dysplasia of Hip (Congenital)
- 456 Hemivertebra
- 457 Osteogenesis Imperfecta
- 458 Scoliosis (Infantile) and/or Kyphosis
- 459 Reduction Deformity, Arm or Hand
- 460 Reduction Deformity, Leg or Foot

**NEUROLOGIC**

- 500 Anencephaly
- 501 Encephalocele
- 502 Holoprosencephaly
- 503 Hydranencephaly
- 504 Hydrocephalus (Congenital or Early)
- 505 Microcephaly (Congenital or Early)
- 506 Porencephaly
- 507 Spina Bifida
- 508 Spinal Muscular Atrophy (Infantile)

**OROFACIAL**

- 550 Choanal Atresia
- 551 Cleft Lip with or without Cleft Palate
- 552 Cleft Palate
- 553 Craniosynostosis
- 554 Microtia/Anotia

**PULMONARY**

- 600 Cystic Fibrosis
- 601 Diaphragmatic Hernia

**SYNDROMES/ASSOCIATIONS**

- 650 Angelman Syndrome
- 651 Beckwith-Wiedemann Syndrome
- 652 CHARGE Association
- 653 De Lange Syndrome (Cornelia De Lange Syndrome)
- 654 Marfan Syndrome
- 655 Noonan Syndrome
- 656 Oculoauriculovertebral Association (including Goldenhar Association and Hemifacial Microsomia)
- 657 Prader-Willi Syndrome
- 658 Robin Malformation Sequence (Pierre Robin Sequence)
- 659 Smith-Lemli-Opitz Syndrome
- 660 Sotos Syndrome
- 661 Stickler Syndrome
- 662 VATER Association
- 663 Williams Syndrome