2007 Annual Report

Wisconsin Birth Defects Registry

Maternal and Child Health Program
Family Health Section
Bureau of Community Health Promotion
Division of Public Health
Wisconsin Department of Health and Family Services

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January 2008

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INTRODUCTION

The Wisconsin Birth Defects Registry (WBDR) collects information on selected birth defects from physicians, pediatric specialty clinics and hospitals for children diagnosed from birth to age two. This report is intended to describe the burden of birth defects in Wisconsin, summarize the data that has been received, highlight gaps in the data, and suggest future improvements in reporting.

The WBDR was developed over four months, after the legislative rules were passed in April 2003. Four pilot sites were approached who agreed to test the WBDR website beginning in September. One of the sites, during the process, asked if it would be possible to report by direct upload from their electronic records system to the WBDR website. When the first phase of testing ended, the upload process was developed, other improvements were made, and a second pilot phase began in January of 2004.

The second pilot phase was completed in May and the WBDR began statewide rollout in June 2004. Training and information sessions were offered at eleven sites in June, July and August. As potential reporters completed training, they began to submit reports via the WBDR website.

Because the WBDR rollout occurred over several months, reports will not have been consistently received from every reporting source for the approximately three years of data included in this report. Not all organizations and physicians that should be reporting are doing so and some of those who have been reporting are not reporting regularly. The data show that some areas of the state are reporting more than others and some reportable conditions are generating more reports than others. Analyses are expected to spotlight where improvements can be made.

The data in this report were compiled by the Children and Youth with Special Health Care Needs (CYSHCN) Epidemiologist, Elizabeth Oftedahl, in the Division of Public Health (DPH), Bureau of Community Health Promotion (BCHP), Maternal and Child Health (MCH) Program. Professional consultation and written text was supplied by Dr. Richard Pauli, Geneticist, UW-Madison. The CYSHCN team provided ongoing consultation and review. Review and comments were provided by members of the Wisconsin Council on Birth Defect Prevention and Surveillance. The project was overseen by Terry Kruse, Unit Supervisor, Linda Hale, Section Chief, and Susan Uttech, Bureau Director. Special thanks for oversight and review to Dr. Sharon Fleischfresser, CYSHCN Medical Director and Dr. Murray Katcher, Chief Medical Officer.

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Wisconsin Birth Defect Prevention and Surveillance Council
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Thank you to the families who gave permission for their children's photographs to be included in this report.

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The Impact of Birth Defects

The term 'birth defect' refers to abnormal development of body structure, body function or body chemistry that is present at birth. Although birth defects are, by definition, congenital, they sometimes are recognized either during pregnancy or considerably after birth.

Birth defects are common. The best estimate is that they occur in about 1 in every 30 babies.[1] That means that in the United States every year approximately 120,000 babies are born with birth defects.[2] This suggests that in Wisconsin about 2,500 babies with congenital abnormalities are born each year. While collectively common, most specific congenital anomalies are individually rare, and among those thousands of different kinds of birth defects, there are many causes. Some result from genetic processes such as single gene changes or chromosomal abnormalities. Others may be caused by exposures (environmental pollutants, dietary factors, medications, etc.) during pregnancy. Some appear to be random events. Many arise from a poorly understood combination of factors.

Non-genetic causes such as environmental exposures, pharmacologic exposures and use of alcohol or illicit drugs probably account for less than 10% of all birth defects, and environmental exposures alone are likely to directly cause only 3%.[3] However, this is still not a trivial number. Furthermore, these are the ones most amenable to preventive intervention. Adding to these the approximately 25% of birth defects that appear to be multifactorial (genetic predisposition plus environmental 'triggers') demonstrates the potential consequences of our changing environment.[2]

The cause of only a minority of congenital anomalies is well understood. In fact the cause of at least two-thirds of all birth defects remains wholly or partly a mystery.[1] There is a real need for better understanding of the underlying cause of these processes, which may have devastating effects on the affected individuals and on their families.

The impact of birth defects is huge. They are the leading cause of all infant deaths and of intrauterine deaths (stillbirth), resulting in between 8-10,000 deaths each year in the United States [4-6] (and approximately 150-200 deaths in Wisconsin). About 20% of babies who die in the first year of life do

so because of congenital anomalies.[6] They are also the most common cause of childhood disability – both medical and developmental. Babies with birth defects are often born preterm which increases the risk of death.

The financial and societal consequences are also considerable. At least 1/3 of all pediatric hospital admissions are associated with birth defects or genetic conditions.[7,8] The estimated lifetime costs related to birth defects in the United States is at least $8 billion each year for the 18 'most significant' birth defects alone. Hundreds of thousands of dollars of additional expense arise, on average, for each baby with a birth defect. Birth defects are the fifth leading cause of years of potential life lost.[2]

Preventing birth defects can have major consequences for individuals, families, and for all of us. Yet most birth defects are not sufficiently understood to be preventable. The benefits of understanding an underlying cause are well demonstrated by the story of folic acid. Recognizing that folic acid deficiency is a major contributor to the development of neural tube defects (spina bifida and anencephaly) means that with appropriate intervention as many as 70% of these birth defects can be prevented.[9,10] The U.S. Public Health Service has recommended that all women of reproductive age take 0.4 milligrams of folic acid daily to reduce the risk of neural tube defects in their offspring.[11] Understanding the causes of other congenital anomalies might lead to equally effective ways to prevent them.

Purposes of Birth Defects Surveillance

The ultimate goal of birth defects surveillance is to decrease the frequency and consequences of birth defects. There are other, more limited, objectives. These include the following:

- *Epidemiologic data collection*. For many birth defects there are not enough data to know how often they occur and very limited Wisconsin-specific data for either frequency of occurrence or geographic distribution. There is no way of assessing whether prevalence is increasing, staying the same or decreasing. Surveillance should help to fill these gaps.

- *Identification of possible environmental causes or environmental triggers*. Finding disparate exposure histories (or placeholders for that) might suggest environmental changes that place infants at risk.
- *Formulating prevention strategies*. The example of folic acid is a good one to show the potential efficacy of identifying environmental triggers.
- *Tracking of apparent clusters*. Through surveillance, geographic or racial/ethnic clustering of specific birth defects may be identified. On the one hand, this may allow for identification of emerging environmental risks. On the other, good epidemiologic data can allow for recognition of false clusters and provide reassurance regarding perceived risks.
- *Contributing to generation of policy and programs*. Without accurate data on occurrence of birth defects, it is difficult to develop any rational assessment regarding current or future need for services.
- *Referral for services*. For individual families, birth defects surveillance can provide a linkage to services that might help their child and provide them with accurate information.

Surveillance is only one of a large number of needed activities related to birth defects and genetic disorders. Many others are discussed in the Genetic Services Plan for Wisconsin available at the Genetics in Wisconsin website.

Types of Birth Defects Surveillance

A variety of approaches can be used in trying to identify infants with birth defects. In designing a program, two fundamental questions need to be answered: Will ascertainment be *active* or *passive*? Will the program *stand alone* or be *linked*?

Active surveillance means that personnel of a surveillance system ascertain events through chart review, family interviews or other similar efforts, while *passive surveillance* depends upon reporting by others to some central facility. All agree that active surveillance is superior to passive systems. However, active surveillance is also far more expensive – prohibitively so for many states. In fact, only 12 states currently have active surveillance systems and only 9 of the 12 include the entire state in their surveillance system.[12]

Programs that are *stand alone systems* use a single source for data. So, for example, a system might depend entirely on physician reporting, or on birth certificates. In contrast, *linked systems* use many different sources to try to maximize identification and minimize reporting errors. In general, linked systems are superior to all stand alone formats, except for active surveillance ones.

History of Birth Defects Surveillance in Wisconsin

Beginning in the 1980s a small group of individuals from both state government and universities in Wisconsin initiated efforts to establish a reporting system for birth defects and related conditions. Those efforts culminated in legislation that created the Birth Defects Outcome and Monitoring Program (BDOMP). This was one of the earlier efforts by a state to create a meaningful surveillance program. Even then it was acknowledged that an active system was too costly to justify. So, a passive, physician reporting system linked to other sources of information (such as birth certificates, death certificates, hospital discharge summaries) was proposed. BDOMP began following legislative action in 1989. In retrospect, the program was overly ambitious in both what needed to be reported and how reporting was to be used, and was from the beginning underfunded. Although some valuable data accrued, it never became a fully active or successful program.

More recently both federal and state initiatives related to birth defects surveillance emerged. In particular, the Birth Defects Prevention Act of 1998 directed the National Centers for Disease Control and Prevention to work through the states to collect and analyze data on birth defects.

In Wisconsin a new Birth Defects Surveillance program was created through Wisconsin Statute 253.12 in May, 2000. This replaced the BDOMP program. The new program was a passive reporting system (requiring reporting by pediatric specialty clinics and physicians). It was intended to be a linked system, but limitations of the current statute have precluded such linkage thus far.

Not only did this legislation mandate reporting and development of a registry, but also required that a Birth Defects Surveillance Advisory Council be constituted. The Advisory Council includes representatives of relevant organizations (universities, physician providers, advocacy groups, community organizations, etc.) and parent/family representatives. It was charged with generating

administrative rules, developing a list of reportable conditions, helping with decisions regarding reporting details and so forth.

The administrative rules process was completed and the rules, Chapter HFS 116, took effect on April 1, 2003. After considerable testing at pilot sites, the Wisconsin Birth Defects Registry began to collect data in the summer of 2004.

The Wisconsin Birth Defects Registry (WBDR)

The current system has the following characteristics:

- It is a *passive* surveillance system, dependent upon reporting by physicians and specialty clinics.
- It is *selective* and *closed.* That is, reporting of only certain birth defects is mandated and the list of reportable conditions is explicit.
- It is *age delimited* – only children up to 2 years of age who have birth defects are reportable

A major challenge was the creation of a rational and consistent list of reportable diagnoses. The Advisory Council for WBDR created a Scientific Subcommittee charged with generating that list. An initial list was developed based on a set of primary criteria. The criteria said that the proposed birth defect should:

- Conform to the statutory definition of a birth defect – a structural deformation, disruption or dysplasia, or a genetic, inherited, or biochemical disease that occurs prior to or at birth.
- Usually be identifiable by 2 years of age (the limit of the statute).
- Be a major anomaly (having medical, surgical or developmental significance).
- Be of 'sufficient' frequency (birth prevalence) – an estimated prevalence of 1/30,000 births was selected; this would mean that 2 or more occurrences each year in Wisconsin would be expected.

The last of these was most challenging, primarily because for many birth defects no accurate prevalence estimates are available. The subcommittee also attempted to make the resulting list as consistent as possible with data being collected elsewhere in the country. In addition, the list does not

include most conditions identified by current newborn metabolic screening since ascertainment of these is virtually complete anyway.

The generated list of reportable conditions was reviewed and assessed by the Scientific Subcommittee. Relevant portions were then further reviewed by pediatric subspecialists. Nearly three dozen pediatric subspecialists (neurology, otolaryngology, hematology, urology, developmental medicine, neurosurgery, neonatology, orthopedic surgery, endocrinology and cardiology) participated. Their suggestions were then analyzed by the Scientific Subcommittee. Substantial revisions resulted: 9 conditions were added and 5 other reportable conditions were revised; none was deleted based on this review. This resulted in development of a 'final' list (p. 12-14) of reportable conditions. That list will undergo periodic review and conditions may be added or removed based on data that become available in the future.

WBDR Challenges / Next Steps

The WBDR must operate consistent with the provisions of the statute that created it. A primary challenge for complete birth defects reporting is that the current statute requires parental consent to report. The parental consent piece is a challenge for physicians, pediatric specialty clinics and hospitals since it is an administrative burden to request and document consent. Many of the reports come in without parent consent, not because a parent withheld consent, but because the reporting organization was unable to contact a parent and obtain consent before submitting a report.

The parent consent requirement also prevents the WBDR from being linked to other systems and does not allow WBDR data to be supplemented with information derived from other data collection systems because a parent did not specifically consent to information about their child being included in the WBDR. The WBDR as a result must operate as a stand-alone system. Work continues to correct this oversight and allow a linked system as was originally intended. Through such linkages a far better program can be anticipated. Linked systems mean multiple ascertainments. Multiple ascertainments will make missed cases less likely (more *complete* data) and will allow for correction of errors (more *accurate* data).

The Department of Health and Family Services currently collects information from several sources that would be helpful in supplementing and verifying data reported directly to the WBDR. These sources include birth records, death records, fetal death reports, hearing screening data, and data from the maternal and child health reporting system. Improving birth defects data will allow more accurate quantification of the burden of birth defects in Wisconsin, assure appropriate referral to services for families, and provide baseline data for use in prevention activities.

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WBDR 2004-2007 Data

From the summer of 2004, when the WBDR was rolled out statewide beginning in June, until August of 2007 (approximately three years), reports for 2,186 babies with 2,447 reportable conditions were submitted. Parent permission was obtained only about 25% of the time which limits the individually identifiable information included in the reports.

Gender

- 967 (44%) babies with reported birth defects are girls, 1,206 (55%) are boys and 3 (0.1%) are undesignated. 10 babies did not have gender reported.

Race / Ethnicity

- 1,272 (68%) of babies with reported birth defects are White; 149 (8%) are African American/Black; 42 (2.2%) are Asian; 8 (0.4%) are American Indian/Alaskan Native; 95 (5%) are other; 12 (0.8%) are Multiracial; and 289 (15%) are reported as race unknown. 319 babies did not have race reported.
- 53 (11.5%) babies with reported birth defects are Hispanic/Latino; 408 (88.5%) reported they are not Hispanic/Latino. 1,725 babies did not have ethnicity reported.

State of Residence

- 2,019 (93%) babies with reported birth defects are from Wisconsin; 139 (6.4%) are from the border states of Illinois, Iowa, Michigan and Minnesota; and 10 (0.5%) are from other states. 18 babies did not have state of residence reported.

Birth Type / Current Status

“Birth type” is defined as either a live birth or a stillbirth. “Current status” is whether the baby was alive at the time the birth defects report was submitted. This statistic allows differentiation between babies who died at birth and those who were born alive but died shortly after birth before a report was submitted.

- 1,015 (97.6%) babies with reported birth defects were born alive; 25 (2.4%) were stillborn. 1,146 babies did not have birth type reported.
- 945 (95.3%) babies with reported birth defects were alive at the time the report was submitted; 47 (4.7%) were deceased. 1,194 babies did not have current status reported.

Conditions Prenatally Diagnosed

- 333 (42.5%) babies with reported birth defects were diagnosed prior to birth; 451 (57.5%) were not. 1,402 babies did not have whether their conditions were prenatally diagnosed included as part of the report.

Gestational Age

- Gestational age was entered for 613 of the 2,186 babies with reported birth defects as follows:

<24 weeks	6	1.0%
24-27 weeks	14	2.3%
28-31 weeks	25	4.0%
32-35 weeks	57	9.3%
36-39 weeks	326	53.2%
40 weeks	138	22.5%
>40 weeks	47	7.7%

Birthweight

- Birthweight was entered for 720 of the 2,186 babies with reported birth defects as follows:

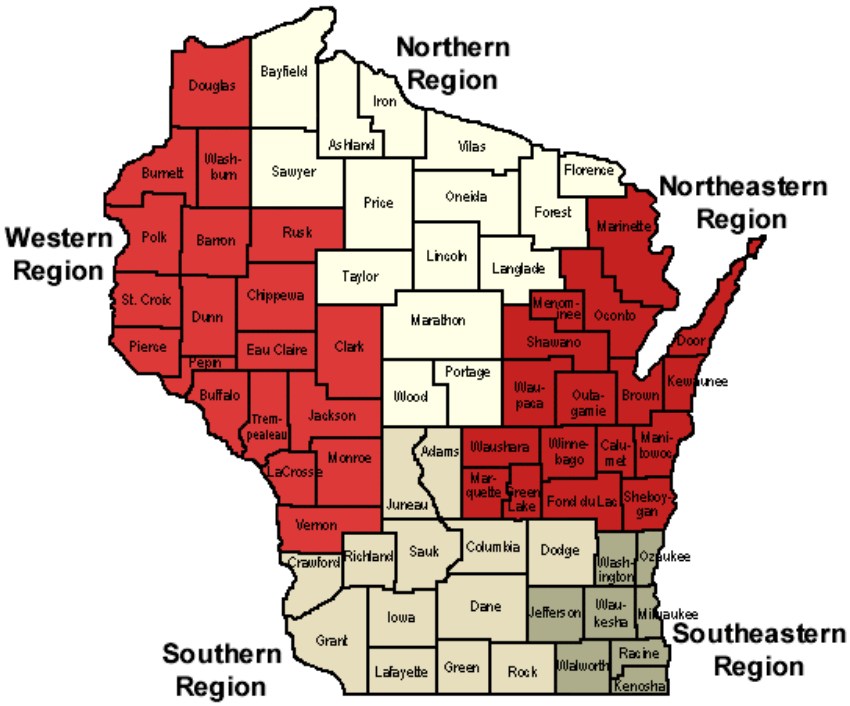
Extremely low birthweight (<1,000g)	34	4.7%
Very low birthweight (1,000-1,499g)	25	3.5%
Low birthweight (1,500-2,499g)	110	15.3%
Normal weight (2,500-3,999g)	498	69.2%
High birthweight (4,000+g)	53	7.4%

(1,000g ~ 2.2 lbs; 1,500g ~ 3.3 lbs.; 2,500g ~ 5.5 lbs.; 4,000g ~ 8.8 lbs.)

Reports by Department of Health and Family Services (DHFS) Regions

- There are five DHFS regions as shown on the map on the following page. The number and percent of the 2,019 Wisconsin resident babies with reported birth defects are shown by region and the number and percent of statewide births for 2006 supplied for comparison. For reports that did not include a city for the child or parents, the report was attributed to the region in which the reporting facility resides. This is intended to be a rough estimate of how complete birth defects reporting is by region based on the assumption that birth defects occur at the same rate in all regions.
- Based on the number and percent of births compared to the number and percent of birth defects reports received, it appears there were significantly fewer birth defects reported in the

Northeastern Region as would be expected. For the Southeastern Region, additional research is needed to assess whether a higher percent of birth defects is actually occurring in the region or if reporting is simply more complete than in the other regions.



DPH Region	WBDR reports	% reports
Northern	189	9.4%
Northeastern	257	12.7%
Southern	363	18.0%
Southeastern	980	48.5%
Western	230	11.4%
Wisconsin	2,019	100.0%

DPH Region	2006 births	% births
Northern	5,400	7.4%
Northeastern	14,844	20.7%
Southern	13,531	18.7%
Southeastern	28,887	40.0%
Western	9,640	13.2%
Wisconsin	72,302	100.0%

Reportable Conditions

Over the three years that data have been collected, it is estimated that more than a third (38%) of the reports expected have been received. The table that begins on page 12 includes:

- the expected frequency of each reportable condition
- the expected number of reports for Wisconsin based on an average birth count of 70,000 births per year
- the actual number of reports received
- the percent of reports received (actual reports divided by expected reports)

Caveats

There is a remarkable range in the number of reports for each condition compared with the population-based expected numbers. A number of factors may contribute to these differences:

1. For most conditions, there are no data regarding the distribution of ages of diagnosis since some are always recognized within the first two years and others much less frequently. So, it

would be expected that under-reporting of the latter (e.g., hemivertebrae, Marfan syndrome) would appear to be greater than in the first group (e.g., hypospadias, Down syndrome, achondroplasia). Analysis of reports received to date shows that about 70% of reports are received in the same calendar year that the babies are born. However, organizations that primarily report genetic birth defects submit only about 25% in the same calendar year that the babies are born. Not only may it take longer for a diagnosis to be made for some birth defects, but time from birth to report is probably lengthened by the necessary wait for appointments with one or more specialists.

2. At this stage, a disproportionate number of reports are being received from certain specialists (i.e., geneticists). Those conditions more likely to result in referral to a geneticist may be more often submitted than those that do not. In addition, a physician who refers a baby to a specialist may assume the specialist will submit a report and the specialist may assume the primary care physician will submit a report – so neither does.

3. For those with small expected numbers of reports, much of the apparent variability may be simply random. A larger number than expected to date could very well be offset by fewer cases in subsequent years and vice versa.

4. It appears that conditions that usually or always result in early neonatal death are currently disproportionately underreported. A number of reasons for this disparity can be postulated. For organizations that have electronic patient records, a baby who dies before hospital discharge may never have a separate patient record created so mining the records for birth defects information may miss these babies entirely.

5. Information is not submitted for pregnancy losses that occur prior to 20 weeks gestation and for elective terminations. Therefore any birth defects that occur in these groups are not included in the WBDR count of birth defects.

6. The expected frequencies for some conditions may be inaccurate – for example, many are based on limited data and should be viewed as rough estimates only. Virtually all are based on estimates not specific to Wisconsin. Many years of complete reporting will make it possible to provide more accurate, Wisconsin-specific frequencies.

WBDR Reportable Conditions

- Expected Incidence and Actual Number of Reports Received*

	CARDIOVASCULAR	**Frequency**	**Estimated**	**Actual**	**Percent**
100	Atrial Septal Defect	1 in 1500	128	137	107%
101	Atrioventricular Canal	1 in 6500	30	32	107%
102	Cardiac Arrhythmia	unknown		31	
103	Coarctation of the Aorta	1 in 2000	96	36	38%
104	Hypoplastic Left Heart	1 in 3000	64	31	48%
105	Tetralogy of Fallot	1 in 2000	96	45	47%
106	Total Anomalous PVR	1 in 30000	6	7	117%
107	Transposition of the Great Vessels	1 in 3500	55	22	40%
108	Truncus Arteriosus	1 in 30000	6	7	117%
109	Valvular Heart Disease	1 in 1000	192	76	40%
110	Ventricular Septal Defect	1 in 600	321	286	89%
	CHROMOSOMAL	**Frequency**	**Estimated**	**Actual**	**Percent**
150	Down Syndrome	1 in 700	275	168	61%
151	Klinefelter Syndrome	1 in 1600	120	2	2%
152	Trisomy 13	1 in 10000	19	1	5%
153	Trisomy 18	1 in 6000	32	13	41%
154	Turner Syndrome	1 in 6000	32	13	41%
155	Velocardiofacial Syndrome	1 in 2000	96	9	9%
156	Other Chromosomal Anomaly	1 in 500	385	58	15%
	ENDOCRINE	**Frequency**	**Estimated**	**Actual**	**Percent**
200	Hypothyroidism	1 in 3500	55	30	55%
	EYE	**Frequency**	**Estimated**	**Actual**	**Percent**
250	Cataract	1 in 1500	128	12	9%
251	Coloboma	1 in 4000	48	33	69%
252	Glaucoma	1 in 30000	6	6	100%
253	Microphthalmia/Anophthalmia	1 in 4500	43	10	23%
	GASTROINTESTINAL/ABDOMINAL	**Frequency**	**Estimated**	**Actual**	**Percent**
300	Biliary Atresia	1 in 8000	24	5	21%
301	Gastroschisis	1 in 5000	38	22	58%
302	Hirschsprung Disease	1 in 5000	38	15	39%
303	Omphalocele	1 in 4000	48	11	23%
304	Pyloric Stenosis	1 in 400	481	41	9%
305	Rectal/Colonic Atresia/Stenosis	1 in 5000	38	21	55%
306	Small Bowel Atresia/Stenosis	1 in 500	385	8	2%

* Expected reports are calculated using births per year times the frequency of the birth defect. Actual reports are the number of reports received on the WBDR between June of 2004 and August of 2007, approximately the first three years of mandated reporting.

Conditions, cont.

	GENITOURINARY	Frequency	Estimated	Actual	Percent
350	Ambiguous Genitalia	1 in 8000	24	9	38%
351	Epispadias	1 in 115000	2	7	350%
352	Exstrophy of the Bladder/Cloaca	1 in 45000	4	6	150%
353	Hypospadias	1 in 700	275	127	46%
354	Multicystic and/or Dysplastic Kidney	1 in 3200	60	18	30%
355	Obstructive Urinary Tract Defect	1 in 800	241	23	10%
356	Polycystic Kidney Disease, Dominant	1 in 4000	48	1	2%
357	Polycystic Kidney Disease, Recessive	1 in 9000	21	2	10%
358	Polycystic Kidney Disease, Uncertain	(in 357/358)		3	
359	Posterior Urethral Valves	1 in 13000	15	1	7%
360	Renal Agenesis/Hypoplasia	1 in 4000	48	10	21%
361	Urethral Stenosis/Atresia	1 in 20000	96	2	2%
	HEMATOLOGIC	**Frequency**	**Estimated**	**Actual**	**Percent**
400	Hemophilia	1 in 15000	13	14	108%
401	Hereditary Spherocytosis	1 in 5000	38	4	11%
402	Von Willebrand Disease	1 in 8000	24	6	25%
	MUSCULOSKELETAL	**Frequency**	**Estimated**	**Actual**	**Percent**
450	Achondroplasia	1 in 26000	7	26	371%
451	Amniotic Bands	1 in 10000	19	14	74%
452	Arthrogryposis Multiplex Congenita	1 in 7000	27	9	33%
453	Bone Dysplasia/Dwarfism, Other	1 in 6000	32	11	34%
454	Clubfoot (Congenital)	1 in 1000	192	102	53%
455	Hip Dislocation/Developmental Dysplasia of Hip	1 in 1000	192	129	67%
456	Hemivertebra	1 in 3500	55	4	7%
457	Osteogenesis Imperfecta	1 in 20000	10	10	100%
458	Scoliosis and/or Kyphosis			6	
459	Reduction Deformity, Arm or Hand	1 in 3000	64	32	50%
460	Reduction Deformity, Leg or Foot	1 in 6000	32	13	41%
	NEUROLOGIC	**Frequency**	**Estimated**	**Actual**	**Percent**
500	Anencephaly	1 in 2500	77	2	3%
501	Encephalocele	1 in 8000	24	3	13%
502	Holoprosencephaly	1 in 15000	13	8	62%
503	Hydranencephaly	1 in 7000	27	11	41%
504	Hydrocephalus	1 in 1500	128	23	18%
505	Microcephaly	1 in 600	321	49	15%
506	Porencephaly	1 in 9000	21	18	86%
507	Spina Bifida	1 in 1600	120	30	25%
508	Spinal Muscular Atrophy	1 in 10000	19	5	26%

Conditions, cont.

	OROFACIAL	Frequency	Estimated	Actual	Percent
550	Choanal Atresia	1 in 7000	27	7	26%
551	Cleft Lip with or without Cleft Palate	1 in 1000	192	118	61%
552	Cleft Palate	1 in 2000	96	110	115%
553	Craniosynostosis	1 in 1000	192	131	68%
554	Microtia/Anotia	1 in 3500	55	12	22%
	PULMONARY	**Frequency**	**Estimated**	**Actual**	**Percent**
600	Cystic Fibrosis	1 in 3200	60	37	62%
601	Diaphragmatic Hernia	1 in 2100	92	10	11%
	SYNDROMES/ASSOCIATIONS	**Frequency**	**Estimated**	**Actual**	**Percent**
650	Angelman Syndrome	1 in 16000	12	27	225%
651	Beckwith-Wiedemann Syndrome	1 in 14000	14	12	86%
652	CHARGE Association	1 in 10000	19	4	21%
653	De Lange Syndrome	1 in 30000	6	2	33%
654	Marfan Syndrome	1 in 10000	19	2	11%
665	Noonan Syndrome	1 in 2000	96	4	4%
656	Oculoauriculovertebral Association	1 in 6000	32	7	22%
657	Prader-Willi Syndrome	1 in 15000	13	7	54%
658	Robin Malformation Sequence	1 in 2000	96	12	13%
659	Smith-Lemli-Opitz Syndrome	1 in 29000	7	0	0%
660	Sotos Syndrome	1 in 20000	10	0	0%
661	Stickler Syndrome	1 in 10000	19	3	16%
662	VATER Association	1 in 6000	32	5	16%
663	Williams Syndrome	1 in 10000	19	1	5%
	TOTAL REPORTABLE CONDITIONS	**1 in ~30**	**6,417**	**2,447**	**38%**

Comparative Data – Birth Certificates

According to Wisconsin birth defects surveillance legislation, all reports must be submitted directly to the WBDR either via the secure website or on a paper form. Although Wisconsin birth defects legislation prohibits using alternative sources of data to supplement reports that come in directly to the WBDR, comparing reports that come in via the WBDR to data from other reporting sources to assess completeness of birth defects reporting is permitted.

Birth defects can be reported on birth certificates. Practically speaking, that limits reporting to conditions that are readily diagnosable immediately after birth. However, any anomaly can be reported rather than the prescribed list used for the WBDR. In 2006, there were 72,302 births to Wisconsin residents. 1,142 birth certificates (1.6%) were submitted with one or more anomalies reported.

More work on comparing birth records to WBDR reports will be completed over the next several years. The results will confirm whether some of the same children are being reported on birth certificates as are reported on the WBDR, and assess whether the same conditions are being reported in both places. Since the WBDR reportable conditions are not exactly the same as the anomalies that can be reported on a birth certificate, the analysis is not expected to result in perfect matches for children in both data sets. However, the results will help suggest areas in which outreach and education would be helpful in obtaining more complete WBDR data.

Comparative Data – Death Certificates

Some of the same issues that apply to data from Wisconsin birth certificates also apply to data from Wisconsin death certificates. Without a perfect match, it's difficult to certify if the birth defects reported on death certificates have also been reported on the WBDR.

Only 22 children who were born alive and died after birth were documented on the WBDR as having a reportable condition during the first three years of data collection. By comparison, in

2006 alone, data from death records indicated that 64 infant deaths out of 462 had at least one congenital anomaly present. It is likely that some children included in the WBDR died after the report was submitted. A follow-up report is not required if a child dies later. However, it is also likely that the WBDR is missing many of these children.

If the WBDR was automatically linked to birth and death records, there would be noticeable benefits. Diagnoses could be verified across data systems resulting in more accurate data and a report that only appears in one data system could be confirmed or refuted, and either added to the other data systems (if confirmed) or removed (if refuted) resulting in more complete data.

Currently, there is no accurate information on the number of Wisconsin residents living with birth defects making it difficult to plan for and provide appropriate services for all affected individuals. The ability to count new cases and make adjustments for deaths would be a beginning. Since relatively few birth defects are fatal, keeping accurate counts of children living with birth defects is critical in assuring that eligible children and their families receive the ongoing health and educational services to which they are entitled.

Comparative Data – Fetal Death Reports

Fetal death reports are submitted for miscarriages of at least twenty weeks gestation or stillbirths weighing at least 350 grams. By comparison, babies who are born alive and die, no matter how short a time they live, are documented with an official Wisconsin birth certificate and a death certificate.

There were 384 fetal death reports filed in 2006. 59 reports indicated at least one congenital anomaly was present. Only 25 fetal deaths were reported to the WBDR over the first three years of data collection which suggests that stillbirths with birth defects present are vastly underreported on the WBDR. The WBDR staff is currently working with stillbirth projects in two locations to improve reporting but more work is needed.

Improving fetal death reporting on the WBDR will require: (1) working with reporting facilities to improve compliance and/or, (2) allowing use of fetal death reports to supplement reports received directly via the WBDR.

Summary / Projection

WBDR reporting over the first three years was good in some areas, fair in other areas, and poor in still other areas. As compliance improves, completeness of reporting will also improve.

To improve compliance, WBDR staff will continue to publicize the directory through reports such as this one, and outreach activities and training opportunities, as well as working one-on-one with reporting facilities. WBDR staff is also working on initiatives to better serve children born with a birth defect, help prevent birth defects and assure that children with birth defects and their families are referred to appropriate services.

Appendix 1: On-Line Resources

Wisconsin Department of Health and Family Services: http://dhfs.wisconsin.gov/

Wisconsin Association for Perinatal Care (WAPC): http://www.perinatalweb.org/association/unhs.html

March of Dimes: http://www.marchofdimes.com/wisconsin/

Genetic Resources for Wisconsin Families and Professionals: http://www.slh.wisc.edu/genetics/

Wisconsin Council on Developmental Disabilities (WCDD): http://www.wcdd.org/

The Arc-Wisconsin Disability Association: http://www.arc-wisconsin.org/

Down Syndrome Association of Wisconsin: http://www.dsaw.org/

Family Voices of Wisconsin**:** http://www.wfv.org/fv/

Family Village: http://www.familyvillage.wisc.edu/

Waisman Center : http://www.waisman.wisc.edu/

Wisconsin Coalition for Advocacy: http://www.w-c-a.org

Wisconsin Family Assistance Center for Education, Training and Support (FACETS): http://www.wifacets.org/

Wisconsin Family Ties: www.wifamilyties.org

Wisconsin First Step: www.mch-hotlines.org

Wisconsin Stillbirth Service Program: http://www.wisc.edu/wissp/

National Information Center for Children and Youth with Disabilities: http://www.NICHCY.org/

Regional Children and Youth with Special Health Care Needs (CYSHCN) Centers

Northern Regional Center – Wausau: www.co.marathon.wi.us/cyshcn.asp

Northeast Regional Center – Neenah: www.northeastregionalcenter.org

Western Regional Center – Chippewa Falls: www.co.chippewa.wi.us/ccdph/cyshcn

Southern Regional Center – Madison: www.waisman.wisc.edu/cshcn

Southeast Regional Center – Milwaukee: http://southeastregionalcenter.org/

Appendix 2:
WBDR Reportable Conditions Definitions

Cardiovascular:

- **Atrial Septal Defect** – This is a hole that occurs when the septum separating the right and left atria doesn't close properly. This allows blood from the left atrium to flow into the right atrium, instead of into the left ventricle and on to the aorta and the rest of the body. The defect can cause several complications. Minor cases may cause no symptoms and may not require treatment. Larger defects may require surgical closure or cardiac catheterization.
- **Atrioventricular Canal** – A combination of defects, including a large hole in the center of the heart and a single common valve instead of the separate tricuspid and mitral valves. This is also called atrioventricular septal defect. The condition is often associated with Down syndrome. Infants may also have trouble breathing and not grow well. Surgery is often done in infancy to close the hole and reconstruct the valves.
- **Cardiac Arrhythmia** – Irregularity of the heartbeat caused by damage to or defect in the heart tissue and its electrical system.
- **Coarctation of the Aorta** – A narrowing (coarctation), or constriction, in a portion of the aorta. Coarctation forces the heart to pump harder to get blood through the aorta and on to the rest of the body. This defect can cause several life-threatening complications.
- **Hypoplastic Left Heart Syndrome** – In this condition, the left side of the heart is underdeveloped (hypoplastic), including the aorta, aortic valve, left ventricle and mitral valve. As a result, the body doesn't receive enough oxygenated blood. Treatment options for this life-threatening condition are a heart transplant or a multistage surgical procedure done during the first few years of life.
- **Tetralogy of Fallot** – This defect is a combination of four (tetralogy) congenital abnormalities. The four defects typically are ventricular septal defect (VSD), pulmonary stenosis, a misplaced (overriding) aorta and a thickened right ventricular wall (right ventricular hypertrophy). They usually result in an insufficient amount of oxygenated blood reaching the body. Surgical repair of the defects is required early in life.
- **Total Anomalous Pulmonary Venous Return** (TAPVR) – Total anomalous pulmonary venous return is a congenital heart disease in which none of the four veins that drain blood from the lungs to the heart is attached to the left atrium (upper chamber of the heart). In normal circulation, blood is oxygenated in the lungs and then returns to the left atrium, flowing from there to the left ventricle, through the aorta, and around the body. In TAPVR, oxygenated blood returns to the right atrium instead. From there it goes to the right ventricle, through the pulmonary artery and back to the lungs. This condition is fatal unless a large atrial septal defect (ASD) or patent foramen ovale (passage between the left and right atria) exists to allow oxygenated blood to flow to the left side of the heart and subsequently the body.
- **Transposition of the Great Vessels** – The positions of the aorta and the pulmonary artery (the great arteries) are reversed (transposed). The aorta arises from the right ventricle instead of the left and the pulmonary artery arises from the left ventricle instead of the right. This creates a circulatory pattern that prevents nourishing oxygenated blood from reaching the body. Surgical repair is usually necessary shortly after birth.
- **Truncus Arteriosus** – This is a defect in which the normally distinct pulmonary artery and aorta instead form one single great vessel (truncus) arising from the right and left ventricles. Surgery early in life, and sequential additional surgical procedures are usually needed.
- **Valvular Heart Disease** – Valvular heart disease refers to several disorders and diseases of the heart valves, which are the tissue flaps that regulate the flow of blood through the four chambers of the heart. Patients with valvular heart disease have a malfunction of one or more of these valves.
- **Ventricular Septal Defect** – Sometimes called a hole in the heart, this defect — the most common congenital heart defect — occurs when the septum, the wall separating the right and left ventricles, fails to fully form. Small holes may heal spontaneously or cause no symptoms. Larger holes may require surgical repair.

Chromosomal:

- **Down Syndrome (Trisomy 21)** – A congenital condition which is usually characterized by moderate mental retardation, unusual facial features, a broad short skull, broad hands and short fingers. Other congenital abnormalities may include heart defects, esophageal atresia and duodenal atresia. All of these findings are secondary to trisomy (an extra chromosome copy) of the 21st chromosome.
- **Klinefelter Syndrome** – A genetic syndrome caused by the presence of an extra X chromosome in a male. The normal male has one X and one Y chromosome, while in this disorder two X chromosomes and one Y chromosome are present. Infants usually appears normal at birth, Features often become apparent in puberty when secondary sex characteristics fail to fully develop. It is characterized by small testes, infertility and, in some instances, learning and behavioral problems.
- **Trisomy 13 (Patau Syndrome)** – A condition caused by the presence of three rather than the normal two copies of chromosome 13. Children born with this syndrome have multiple malformations, commonly including scalp defects, cleft lip and palate, malformations of the heart and abdominal organs, and extra digits. The majority of trisomy 13 babies die soon after birth or in infancy. In survivors mental retardation is profound.
- **Trisomy 18 (Edward's Syndrome)** – A condition causes by the presence of three instead of the normal two copies of chromosome 18. Children with this condition have multiple malformations, including low birth weight, small head (microcephaly), small jaw (micrognathia), malformations of the heart and kidneys, clenched fists with abnormal finger positioning, and malformed feet. Most affected infants die in the first year of life. In those who survive mental retardation is profound.
- **Turner Syndrome** – A genetic disorder in females that is caused by the absence of one X chromosome. Females typically have two X chromosomes, while those with Turner syndrome have only one. This disorder inhibits normal sexual development and causes infertility. Features often include webbing of the neck, short stature, coarctation of the aorta, low hairline, mildly unusual facial features, incomplete development of secondary sex characteristics, absence of menses and infertility.
- **Velocardiofacial Syndrome** – Also known as Shprintzen Syndrome, this disorder arises because of a microdeletion (a small piece of missing chromosomal material) of chromosome band 22q11.2. Features include cleft palate, heart defect, abnormal face, abnormal calcium metabolism, and learning problems. The same deletion in different individuals can cause velocardiofacial syndrome in some and DiGeorge sequence (associated with immune function problems) in others.

Endocrine:

- **Hypothyroidism** – Underactivity of the thyroid gland at birth, resulting in growth retardation, developmental delay and other abnormal features. The condition is detected by newborn screening.

Eye:

- **Cataract** (congenital) – A cataract, or clouding of the lens of the eye, sometimes begins in utero. There are many causes of congenital cataract. Treatment includes cataract removal and the insertion of an artificial lens.
- **Coloboma** – Congenital anomaly in which some of the structures of the eye are absent due to incomplete fusion of the fetal intraocular fissure during gestation. If of the iris, this is sometimes called a 'keyhole pupil.' If of the retina, it can result in vision deficits.
- **Glaucoma** (congenital) – This arises from increased intraocular pressure and can cause enlargement of the eyeball and damage to vision.
- **Microphthalmia** – An unnatural smallness of the eyes, occurring as the result of disease or of imperfect development.
- **Anophthalmia** – Congenital absence of all tissues of the eye.

Gastrointestinal / Abdominal:

- **Biliary Atresia** – This term refers to the abnormal development of the bile ducts inside or outside the liver. The obstruction of bile flow from the liver can lead to cirrhosis of the liver if not treated. Symptoms include persistent jaundice.
- **Gastroschisis** – A birth defect in which there is failure of development of a part of the abdominal wall. Abdominal contents may protrude through the resulting defect. The opening in the abdominal wall in gastroschisis is never at the site of the umbilicus (the navel or belly button) and is not covered by a membrane
- **Hirschsprung Disease** – A congenital condition which results in an enlarged and poorly functioning colon due to abnormal intestinal motility. It is caused by absence of certain nerve cells in the walls of the intestines. These patients are at risk for intestinal obstruction. Treatment is surgical.
- **Omphalocele** – An opening in the abdominal wall that is at the site of the umbilicus and directly connected to the umbilicus.. "Omphalo-" indicates a relationship to the umbilicus (the navel) and the suffix "-cele" refers to a hernia or rupture, so an omphalocele = a hernia or rupture at the umbilicus. In contrast to Gastroschisis, the protruding part of the intestine is covered by a membrane.
- **Pyloric Stenosis** – Muscular narrowing of the outlet of the stomach so that food cannot pass easily from it into the duodenum, resulting in feeding problems and projectile vomiting. The obstruction can be corrected by a relatively simple surgical procedure.
- **Rectal/Colonic Atresia/Stenosis** – Atresia is the absence or closure of the rectum and/or colon. Stenosis is a narrowing or stricture or the rectum and/or colon.
- **Small Bowel Atresia/Stenosis** – Absence, closure, narrowing or stricture of the small bowel.

Genitourinary:

- **Tracheoesophageal Fistula/Esophageal Atresia** – A congenital anomaly where the upper esophagus ends blindly (atresia) and does not connect with the stomach; often the lower esophagus connects to the trachea (tracheoesophageal fistula\ Treatment involves the surgical repair of the esophagus.
- **Ambiguous Genitalia** – External genitalia not clearly of either sex.
- **Epispadias** – A rare congenital defect in the location of the opening of the urethra. In boys with epispadias, the urethra generally opens on the top or side (rather than the tip) of the penis. In girls, the opening is usually between the clitoris and the labia, but may be in the abdomen.
- **Exstrophy of the Bladder** – A congenital birth defect that is the malformation of the bladder and urethra, in which the bladder is turned "inside out". The bladder does not form into its normal round shape but instead is flattened and exposed outside the body. The urethra and genitalia are not formed completely (epispadius) and the anus and vagina appear anteriorly displaced. Additionally, the pelvic bones are widely separated (diastasis).
- **Cloaca** – The congenital persistence of a common cavity into which the intestinal, urinary, and reproductive ducts open.
- **Hypospadias** – A condition in which the urethral opening does not form completely to the tip of the penis. Instead, the opening may be located anywhere along the underside of the penis. All except the mildest forms require surgical correction.
- **Multicystic and/or Dysplastic Kidney** – This refers to the presence of cysts and other abnormalities of the kidney. Often the affected kidney is markedly enlarged and may be detected either prenatally of by abdominal examination in infancy. Affected kidneys usually have little or no function. There are a number of causes of this process.
- **Obstructive Urinary Tract Defect** – Any blockage at any level that prevents urine flow from the kidneys to the tip of the urethra.

- **Polycystic Kidney Disease, Dominant** – A condition that results in an enlarged, cyst-filled kidney. Although usually discovered in adulthood, some infants with this disorder are symptomatic, including the presence of kidney failure and respiratory problems. Cysts may also be present in the liver.
- **Polycystic Kidney Disease, Recessive** – Unlike the dominant form, this is most commonly noted in infants (and is sometimes referred to as "infantile polycystic kidney disease"). ARPK in a fetus may be identified during a fetal sonogram. Early kidney failure is more common with this form than the autosomal dominant form of polycystic kidney disease.
- **Posterior Urethral Valves (PUV)** – An abnormality of the urethra (the tube that drains urine from the bladder to the outside). The abnormality occurs when the urethral valves, which are small leaflets of tissue, have a narrow, slit-like opening that partially impedes urine outflow. Reverse flow occurs and can affect all of the urinary tract organs. The degree of urinary outflow obstruction will determine the severity of the urinary tract problems.
- **Renal Agenesis** – Renal agenesis is the complete absence of one or both kidneys. If agenesis is unilateral (only one kidney is absent) the other can assume its function. If both are absent, this is a birth defect incompatible with life.
- **Renal Hypoplasia** – A condition in which there is reduction in the size of the functioning renal mass.
- **Urethral Stenosis/Atresia** – Urethral stenosis is a congenital narrowing of the urethral orifice. Urethral atresia means complete absence of this structure and is a rare and usually fatal cause of prenatal bladder outlet obstruction.

Hematologic:

- **Hemophilia** – A bleeding disorder caused by a deficiency in one of the blood clotting factors. Hemophilia A (often called classic hemophilia) is a hereditary disorder that accounts for about 80% of all hemophilia cases. It is a deficiency in clotting factor VIII.
- **Hereditary Spherocytosis** – A hereditary disorder that leads to chronic hemolytic anemia due to an abnormality in the red blood cell membrane.
- **Von Willebrand Disease** – A hereditary clotting disorder caused by defective or deficient Von Willebrand factor, a protein involved in normal blood clotting. It may result in bruising, nosebleeds, bleeding gums or heavy menstrual bleeding.

Musculoskeletal:

- **Achondroplasia** – A genetic abnormality of bone growth that results in dwarfism characterized by shortened arms and legs.
- **Amniotic Bands** – Rupture of the inner (amniotic) membrane around the developing fetus can result in floating strands of amniotic tissue that, in turn, may adhere to or wrap around parts of the fetus and result in constriction rings, limb amputations and other birth defects
- **Arthrogryposis Multiplex Congenita** – Limitation of range of joint motion in multiple sites of the body that are present at birth. There are many causes and many forms of arthrogryposis.
- **Bone Dysplasia/Dwarfism** – Dysplasia comes from Latin roots *dys* and *plasia*, meaning "bad growth". Bone dysplasias are inborn, heritable abnormalities of bone growth, many of which result in small stature (dwarfism).There are many types of bone dysplasias.
- **Clubfoot** – The most common congenital abnormality of the foot. Clubfoot may occur in several forms, but talipes equinovarus (turning of the foot downward and inward) is the most common.
- **Hip Dislocation** – The abnormal formation of the hip joint in which the ball at the top of the thighbone (the femoral head) is not stable within the socket (the acetabulum). Congenital hip dislocation, if not discovered and treated in infancy can lead to a number of serious orthopedic complications.

- **Developmental Hip Dysplasia** – Underdevelopment of the hip joint that is present at birth. Genetic factors likely play a role in this disorder. Hip dysplasia may result in hip dislocation.
- **Hemivertebra** – A congenital defect of the spine in which one side of a vertebra fails to develop completely.
- **Osteogenesis Imperfecta** – A group of genetic diseases of the bones. All result in brittle and increased risk for fractures.
- **Scoliosis** – A lateral curvature of the spine.
- **Kyphosis** – A posterior curvature of the spine.
- **Reduction deformity** – A congenital deformity in which a body part, especially a limb, is shorter than normal or missing certain of its segments.

Neurologic:

- **Anencephaly** – A congenital absence of the brain and cranial vault, with the cerebral hemispheres completely missing or greatly reduced in size. It is incompatible with extrauterine survival.
- **Encephalocele** – A congenital malformation characterized by herniation of the brain and/or meninges through a defect in the skull.
- **Holoprosencephaly** – A disorder caused by the failure of the *prosencephalon* (the embryonic forebrain) to sufficiently divide into the double lobes of the cerebral hemispheres. The result is a single-lobed brain structure and severe skull and facial defects. There is marked variability in the severity of the brain malformation and, in turn, marked variability in the severity of clinical consequences.
- **Hydranencephaly** – A rare condition in which the brain's cerebral hemispheres are absent and replaced by sacs filled with cerebrospinal fluid.
- **Hydrocephalus** – A condition in which the primary characteristic is excessive accumulation of cerebrospinal fluid in the spaces (ventricles) within the brain. Enlargement of the ventricles causes potentially harmful pressure on the tissues of the brain. Hydrocephalus is most often treated with surgical placement of a shunt system.
- **Microcephaly** – A medical condition in which the circumference of the head is smaller than normal. Microcephaly can be present at birth or it may develop in the first few years of life. It is most often caused by genetic abnormalities that interfere with the growth of the cerebral cortex.
- **Porencephaly** – A disorder of the central nervous system involving formation cysts or cavities in a cerebral hemisphere. The disorder can occur before or after birth.
- **Spina Bifida (SB)** – A neural tube defect (a disorder involving incomplete development of the brain, spinal cord, and/or their protective coverings) caused by the failure of the embryo's spine to close properly during the first month of pregnancy. Infants born with SB often have an open lesion on their spine where significant damage to the nerves and spinal cord has occurred. Although the spinal opening can be surgically repaired shortly after birth, the nerve damage may be permanent, resulting in varying degrees of paralysis of the lower limbs. SB may also cause bowel and bladder complications, and many children with SB have hydrocephalus.
- **Spinal Muscular Atrophy (SMA)** – A genetic, motor neuron disease caused by progressive degeneration of nerve cells in the spinal cord. The disorder causes weakness and wasting of the voluntary muscles. There are many types of SMA, including those known Werdnig-Hoffmann disease (SMA type I).

Orofacial:

- **Choanal Atresia** – A narrowing or blockage of the nasal airway by membranous or bony tissue. Choanal atresia blocking both sides (bilateral) of the nose causes acute and severe breathing problems. Blockage on only one side causes far less severe or acute problems.

- **Cleft Lip / Cleft Palate** – A cleft is a separation in a body structure. Clefts that occur in the facial region often involve the lip, the roof of the mouth (hard palate) or the soft tissue in the back of the mouth (soft palate). Two major types of facial clefts are cleft lip/palate and isolated cleft palate. Babies with cleft lip/palate have a cleft lip often accompanied by cleft palate. In isolated cleft palate, the lip is not involved.
- **Craniosynostosis** – The premature fusing of two or more of the bony plates that form an infant's skull. This can result in a misshapen skull and, if severe, can interfere with normal brain growth and development.
- **Anotia** – Congenital absence of an ear.
- **Microtia** – Abnormal smallness of the external ear.

Pulmonary:

- **Cystic Fibrosis** – An inherited disease that affects sodium channels in the body and causes respiratory and digestive problems.
- **Congenital Diaphragmatic Hernia** – The diaphragm is the breathing muscle that separates the chest cavity and the abdominal cavity. Congenital diaphragmatic hernia (CDH) is the absence of the diaphragm, or, more commonly, a hole in the diaphragm. The contents of the abdomen, including the stomach, intestines, liver and spleen may go through the hole (herniated) into the chest. The contents then prevents normal development of the lung (pulmonary hypoplasia) on the affected side

Syndromes / Associations:

- **Angelman Syndrome** – A genetic disease that causes neurological problems. The physician Harold Angelman first identified the syndrome in 1965, when he described several children in his practice as having "flat heads, jerky movements, protruding tongues, and bouts of laughter." Primary characteristics include mental retardation, seizures, little or no speech development, ataxia and 'inappropriate' laughter Angelman syndrome is the result of the deletion or inactivation of a particular series of genes that regulate a protein called ubiquitin (UBE3A) on chromosome 15q11-13.
- **Beckwith-Wiedemann Syndrome** – This syndrome is characterized by large size at birth, overgrowth of the tongue and internal organs (visceromegaly), umbilical hernia and risk for neonatal hypoglycemia.
- **CHARGE Association (CHARGE Syndrome)** – A constellation of congenital malformations (birth defects) that occur together more frequently than would be expected by chance. The name of the condition is an acronym of some of the most frequent features:
  C = Coloboma (cleft) of the eye
  H = Heart malformation,
  A = Choanal Atresia
  R = Retardation of growth after birth \ and Retardation of development
  G = Genital hypoplasia (underdevelopment) in males
  E = Ear malformations and/or deafness
- **DeLange Syndrome** – A congenital syndrome characterized by short stature, microcephaly, delayed development and mental retardation, distinct facial features, and variable upper limb anomalies; also referred to as deLange syndrome and Brachmann-deLange syndrome.
- **Marfan Syndrome** – A heritable condition that affects the connective tissues because of an abnormality of a protein called Fibrillin, type I. Marfan syndrome can affect many body systems resulting in tall stature, skeletal abnormalities, dislocated lenses of the eyes and risk for catastrophic blood vessel dissection.
- **Noonan Syndrome** – A multifaceted disorder with features including facial abnormalities, small stature, congenital valvular heart defects and other characteristics. It most often is caused by an abnormality of a gene called PTPN11.

- **Oculoauriculovertebral Association (Goldenhar Syndrome)** – This is a congenital disorder that varies markedly in severity. It results in asymmetric or unilateral abnormalities of the bones of the face and the external ears. It can cause marked asymmetry of facial form and may be associated with abnormalities of the eyes (epibulbar dermoids), the vertebral column (hemivertebrae) and other organs.
- **Prader-Willi Syndrome** – A complex genetic condition that affects many parts of the body. Affected infants have weak muscle tone (hypotonia), feeding difficulties, poor growth, and delayed development. Beginning in childhood, excessive eating (hyperphagia) develops and, without appropriate management will result in extreme obesity. This condition is caused by the loss of active genes in a specific region of chromosome 15.
- **Robin Malformation Sequence** – The primary abnormality in this condition is a small, underdeveloped lower jaw. The small jaw affects the position of the tongue (retroglossia), which, in turn interferes with the normal closure of the palate (cleft palate) and may cause breathing problems at birth. It may be seen alone or as part of one of several genetic syndromes. It is called a sequence because it results from a series of events that originate in a single birth defect (small lower jaw) with a secondary cascade of additional features.
- **Smith-Lemli-Opitz Syndrome** – A multiple anomalies syndrome caused by an abnormality in cholesterol metabolism resulting from deficiency of the enzyme 7-dehydrocholesterol reductase. It is characterized by prenatal and postnatal growth retardation, microcephaly, moderate to severe mental retardation, and multiple major and minor malformations.
- **Sotos Syndrome** – A genetic condition, also known as cerebral gigantism because of the combination of a large and unusually shaped head and physical overgrowth during the first years of life. It also may result in hypotonia and developmental delays.
- **Stickler Syndrome** – An autosomal dominant condition arising because of abnormality of certain types of collagen. It may result in Robin sequence, high myopia (nearsightedness) and risk for retinal detachment, hearing loss and premature development of osteoarthritis.
- **VATER Association** – VATER is an acronym used to describe a set of features and birth defects that tend to occur together more often than would be expected by chance. They include:
  V = Vertebral anomalies
  A = Anal Anomalies (e.g., imperforate anus).
  TE = Tracheo-esophageal fistula
  R = Radius (lower arm bone) and/or Renal (kidney) problem(s).
  Sometimes other letters are used and the possible acronyms are VAHTER, VACTERL, VACTERLS, and VATERS with H = hydrocephalus C = cardiac, L = limb and S = single umbilical artery.
- **Williams Syndrome** – A congenital (present at birth) disorder in which there are characteristic “elfin” facial characteristics, developmental disability with a specific pattern of strengths and weaknesses (strong verbal skills, unusual verbal utterances, but significant abnormality in other learning spheres), characteristic personality (approaching and social), heart abnormalities (often supravalvular aortic stenosis) and risk for elevated calcium levels.

Appendix 3: Technical Notes

The Wisconsin Birth Defects Registry (WBDR) is a secure web-based system which exists as a program area module (PAM) on the Health Alert Network (HAN). A handful of reports have been submitted on a paper form. Paper reports are entered into the WBDR website reporting system. The WBDR is a real-time database. Reports become part of the ongoing database as soon as they are submitted.

Reporters must register on the HAN and then obtain permission from the WBDR state administrator to become approved reporters. The state administrator grants approved reporters access to the WBDR website. One or more reporters at each reporting site becomes the local administrator, the official contact for WBDR reporting.

Reports may be submitted on paper or on the secure website one report at a time, or may be uploaded as a file containing more than one report directly from a reporting entity's electronic records system to the secure website.

Reporters may view all reports submitted for their reporting entity. Only the state administrator may view all reports submitted by all reporting entities.

The WBDR went statewide in 2004. For purposes of this report, since the WBDR was rolled out over several months, the WBDR was considered fully operational as of October 1, 2004.

Birth, death, and fetal death information was provided by the Bureau of Health Information and Policy (BHIP), Division of Public Health, Department of Health and Family Services. BHIP analysts provided counts from Wisconsin Vital Records data for this report.

Birth defect incidence numbers are estimates; actual occurrence will vary from year to year. Variability is especially noticeable for birth defects where the estimated incidence is less than ten occurrences in a birth cohort of approximately 70,000 births in a year.

More than one reportable birth defect may exist in a single infant. Therefore, the number of babies with birth defects will be fewer than the number of reported birth defects.