

NATIONAL PREVALENCE ESTIMATES

In the past, efforts to investigate higher rates of birth defects in particular areas or among specific populations, or to evaluate the effectiveness of population-based prevention efforts, have been stymied by a lack of reliable baseline birth defect rates. However, over the past 10 years 18 state birth defects registries have either been established or significantly enhanced operations (many funded by the Centers for Disease Control and Prevention). Furthermore, the establishment of the National Birth Defects Prevention Network (NBDPN) has encouraged and supported collaborative projects.

One such project has produced a breakthrough in the form of national prevalence estimates for 18 categories of birth defects. This study, originally published in the CDC's Morbidity and Mortality Weekly Report (MMWR 2006;54:1301-1305) and reprinted in the Journal of the American Medical Association (JAMA, February 8, 2006—Vol 295, No. 6), calculated pooled prevalence estimates using data from 11 states with active birth defects registries. The 11 states pooled for

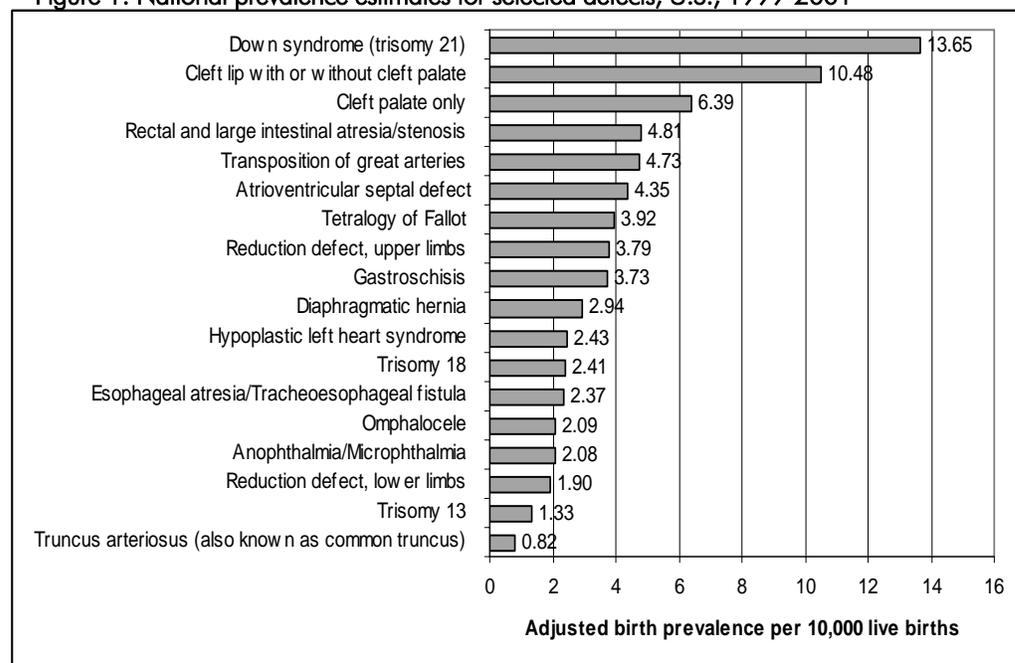
these estimates represent approximately 22% of all U.S. live births, with a similar racial/ethnic distribution. To obtain the national prevalence estimates, race/ethnicity-specific prevalence rates were calculated for all defects, and maternal age-specific prevalence estimates were calculated for Down syndrome (trisomy 21), trisomy 13, and trisomy 18. The authors then applied these adjusted prevalence estimates to racial/ethnic and mater-

(Continued on page 7)

Inside this issue:

Severity of Birth Defects and Cluster Investigations	2
Recent Publications	4
Paxil Warning	4
Preventive Health Services Report	4
Spina Bifida Research Resource	5
FAS Recommendations	6
Symposium Highlights	7

Figure 1: National prevalence estimates for selected defects, U.S., 1999-2001



FROM THE REGISTRY

1999-2002 DATA

Severity of Birth Defects as a Tool for Dealing with Detection Bias in Cluster Investigations

Peter Langlois, Ph.D., Angela Scheuerle, M.D., and Allison Winter, B.S.

Background

Birth defects registries are often requested to examine whether birth defect rates are higher near sites of environmental concern such as hazardous waste sites. Since different birth defects seem to be caused by different factors, it may be more meaningful to evaluate the occurrence of individual defects than to lump them all together. But which defects should be examined?

One problem is that several birth defects vary in severity. While severe cases would be diagnosed and recorded universally, mild cases may only be picked up by clinicians, health care facilities, or practices that employ new or more sensitive equipment or particular practice styles. Detection of those mild cases can lead to artifactual "clusters" of high rates of defects in certain areas or to "epidemics" with apparently increasing rates over time as new technology is gradually adopted. The impact of such clinical practice variation on prevalence has been documented for cardiac defects (Martin et al, 1989; Khoury and Erickson; 1992; Wilson et al, 1993) and craniosynostosis (French et al, 1990; Alderman et al, 1997).

One way to deal with that diagnostic bias is to evaluate occurrence using birth defects that tend to be severe. This report tries to identify such defects.

Method

Cases in the Texas Birth Defects Registry were arbitrarily designated as "severe" if they had any of the following characteristics:

- were reported as having had surgery, repair, or autopsy;
- had a pregnancy outcome of spontaneous fetal death or pregnancy termination;
- if a live birth, the child died within the first year of life.

This was admittedly crude, but it was a definition that could be uniformly applied to all birth defects.

Cases delivered in 1999-2002 were examined. They had to have "isolated" defects, defined as:

- only one major birth defect; or
- one major birth defect and one or more minor defects (Rasmussen et al, 2003).

This was to ensure that a case was not designated as severe by simply having more than one significant structural anomaly. A case with both a chromosomal abnormality and, for example, a heart defect, was considered to have two major defects and was not included in this analysis.

For all cases with a particular birth defect, the proportion that met the criteria for being "severe" cases was tabulated.

Results and Discussion

Overall, 23.7% of cases in the Texas Birth Defects Registry were considered to be "severe" using these criteria. There was a great range of severity between defects. Defects at the upper (severe) end of Table 1 such as trisomy 18 (98.4% severe) and anencephaly (98.1%) may be less susceptible to clinical variation in diagnosis and recording than defects near the lower (less severe) end such as aniridia and Ebstein anomaly (both 0.0%), ventricular septal defects (3.1%), or anotia/microtia (3.4%).

This approach was admittedly crude. For example, most people would consider reduction defects of the upper or lower limbs to be severe defects, but they may not threaten life and thus would not rate highly on this table. Some reduction defects may not be as amenable to surgical intervention; without having surgery, repair, or autopsy they would not be labeled 'severe'. Further, limb reduction defects are obvious and unlikely to be missed, and thus may not suffer from diagnostic variation as much as the table might indicate.

On the other hand, the defect ranking in this paper agreed roughly with the prioritization based on a subjective evaluation of diagnostic accuracy by two dysmorphologists and one pediatrician (Environmental Public Health Tracking Workshop, 2005). Therefore this approach may be a start toward identifying birth defects that are less susceptible to artifactual clusters or temporal increases, and thus more reliable in cluster investigations. Alternatively, using this computer algorithm may allow registries to crudely but easily identify "severe" cases of any birth defect in areas of environmental concern, and compare their occurrence to occurrence in unaffected areas.

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Milestone

As of June 13, 2006, the Texas Birth Defects Registry added its 100,000th case. The program began collecting data in 1994.

Table: Birth defects sorted in descending order by severity.

Birth Defect	# Cases	% Severe
Trisomy 18 (Edwards syndrome)	64	98.4
Anencephaly	314	98.1
Pyloric stenosis	2490	90.1
Hypoplastic left heart syndrome	31	87.1
Trisomy 13 (Patau syndrome)	15	86.7
Diaphragmatic hernia	162	79.6
Encephalocele	43	79.1
Omphalocele	73	75.3
Holoprosencephaly	12	75.0
Stenosis or atresia of large intestine, rectum, or anal canal	200	75.0
Agenesis, aplasia, or hypoplasia of the lung	124	73.4
Tracheoesophageal fistula/esophageal atresia	79	73.4
Gastroschisis	402	69.4
Stenosis or atresia of small intestine	139	68.4
Biliary atresia	50	64.0
Transposition of the great vessels	60	63.3
Hirschsprung disease	124	62.9
Spina bifida without anencephaly	160	60.6
Coarctation of the aorta	58	60.3
Craniosynostosis	403	54.1
Bladder exstrophy	6	50.0
Cataract	112	42.0
Common truncus	10	40.0
Endocardial cushion defect	25	40.0
Tetralogy of Fallot	121	38.8
Cleft lip with or without cleft palate	1047	34.6
Trisomy 21 (Down syndrome)	545	28.1
Hydrocephaly	301	27.2
Renal agenesis or dysgenesis	149	25.5
Anophthalmia	8	25.0
Cleft palate alone (without cleft lip)	399	22.8
Choanal atresia or stenosis	80	20.0
Hypospadias or epispadias	3184	16.5
Obstructive genitourinary defect	1509	15.0

Birth Defect	# Cases	% Severe
Patent ductus arteriosus	2080	13.7
Aortic valve stenosis	59	11.9
Congenital hip dislocation	394	8.1
Reduction defects of the upper limbs	180	7.2
Anomalies of the tricuspid valve	15	6.7
Microcephaly	278	6.5
Reduction defects of the lower limbs	46	6.5
Atrial septal defect	1682	6.1
Microphthalmia	18	5.6
Pulmonary valve atresia or stenosis	218	4.6
Anotia or microtia	176	3.4
Ventricular septal defect	3235	3.1
Aniridia	4	0.0
Ebstein anomaly	16	0.0
Fetal alcohol syndrome or other alcohol related birth defects	8	0.0
Infants and fetuses with any monitored birth defect	32773	23.7%

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PREVENTION

FDA STRENGTHENS ITS WARNING ON PAXIL

WASHINGTON (AP) - The Food and Drug Administration is strengthening its warning that the antidepressant Paxil may be associated with birth defects, citing a new study that found increased risk of fetuses developing heart defects.

The FDA asked manufacturer GlaxoSmithKline PLC to reclassify the drug, which goes by the generic name paroxetine, as a "Category D" drug for pregnant women. The classification means that studies in pregnant women have shown a risk to the fetus. However, the FDA said, the benefits of the drug may outweigh the risk to the fetus.

Two studies of pregnant women taking Paxil during their first trimester have shown their babies have heart defects one and a half to two times a greater rate than the norm, the FDA said. The agency announced the strengthened warning Thursday. It issued a previous warning in September.

The FDA is advising doctors not to prescribe Paxil to women in their first three months of pregnancy or people who are planning to become pregnant, unless there are no other options.

REPORT RANKS AMERICA'S PREVENTIVE HEALTH SERVICES

A new Partnership for Prevention report that ranks America's highest impact and most valuable preventive health services is now available at <http://www.prevent.org/ncpp>. *Priorities for America's Health: Capitalizing on Life-Saving, Cost-Effective Preventive Services.* This document identifies those preventive services that are most beneficial and cost-effective for the U.S. population yet are being utilized by less than half of Americans.

"Currently about 95 percent of health care dollars in the United States is spent on treating diseases, with relatively little attention paid to preventing disease," said David Satcher, M.D., former U.S. Surgeon General and chair of the

National Commission on Prevention Priorities, which was convened by Partnership for Prevention to help guide the report. "This landmark study highlights the importance of shifting focus to preventive care, which can provide an enormous positive impact on health and well-being, while also more effectively allocating our precious health care dollars. Basically, these are the preventive health services that offer the biggest bang for the buck."

Relying on scientific research and clinical preventive recommendations issued by the US Preventive Services Task Force and Advisory Committee on Immunization Practices, the authors developed a system for ranking services based on both health benefit (Clinically Preventive Burden Ranking) and economic value (Cost Effectiveness). Data on utilization rates were then captured for those services ranking highest.

The following preventive health services are of particular interest for birth defects prevention:

Service	Clinically Preventive Burden Ranking (1-Lowest, 5-Highest)	Cost Effectiveness Ranking (1-Lowest, 5-Highest)
Problem Drinking Screening and Brief Counseling	4	4
Folic Acid Chemoprophylaxis	2	3
Obesity Screening	3	2
Diabetes Screening	1	1

Additional information about each of these services as well as a complete list of the rankings can be found at www.prevent.org/ncpp. Detailed report findings will be published in the July 2006 edition of the *American Journal of Preventive Medicine*.

LIVING WITH BIRTH DEFECTS

The Spina Bifida Research Resource (SBRR) has received an additional five years of funding from the NIH. The SBRR was initiated in 1997 to study genetic and environmental factors that contribute to spina bifida, and has received NIH funding since 2000. The Principal Investigator of the SBRR, Laura Mitchell, Ph.D., is an Associate Professor at the Institute of Biosciences and Technology, Texas A&M University Health Science Center in Houston.

Over 650 families have already joined the SBRR, but information from many more families is needed to ensure that the factors associated with spina bifida will be identified. Over the next four years, the SBRR will recruit an additional 400 families. Although spina bifida is one of the most common birth defects, it is rare compared to diseases like breast cancer and cardiovascular disease. Consequently, every family that participates in the SBRR is very important.

Individuals with spina bifida (myelomeningocele) and their families, and families that have received a prenatal diagnosis of spina bifida, are eligible to participate in the SBRR. Par-

ticipation includes an interview, which can be completed in English or Spanish, to gather information on family and pregnancy history, and the collection of samples (saliva, cheek cells or blood) for DNA analysis. There are no costs or travel associated with participation. All aspects of the study can be accomplished by telephone and mail. For further information about the SBRR, please contact Barbara Weyland, project coordinator, toll-free at 1-866-521-7289 or at bweyland@ibt.tamhsc.edu.

ANNOUNCEMENTS

YOUR FEEDBACK REQUESTED

Please take a few moments to go online to express your views on this publication at www.surveymonkey.com/s.asp?u=866951111809.

TEXAS 2002 PRAMS DATABOOK AVAILABLE

The Pregnancy Risk Assessment Monitoring System (PRAMS) is a Centers for Disease Control and Prevention (CDC)-sponsored initiative to reduce infant mortality and low birth weight births. PRAMS is an ongoing state specific population-based surveillance system designed to identify and monitor selected maternal experiences before, during and after pregnancy.

Texas is one of twenty-nine states participating in PRAMS. Many states have used PRAMS findings to increase understanding of maternal behaviors and experiences and their relationship with adverse pregnancy outcomes. These data can be used to develop and assess programs and policies designed to reduce adverse pregnancy outcomes and improve the health of babies and mothers. Texas PRAMS conducts surveys by mail and telephone of mothers who are residents of Texas who have recently given birth. The Texas PRAMS 2002 Databook presents findings on key survey questions on the following topics: Insurance (prenatal care and delivery), nutrition and folic acid awareness, prenatal care, communication with health care providers, smoking, alcohol use, abuse before and during pregnancy, infant health, infant sleeping position, postpartum depression, pregnancy intention, and contraceptive use.

A summary of findings from the PRAMS 2002 Databook is on the web and can be downloaded at www.dshs.state.tx.us/chscontracts/newsletter/Spring2006/PRAMS.shtm.

TEXAS OBESITY STRATEGIC PLAN

Obesity has been the focus of several recent birth defect studies (see *Monitor* Vol. 10-2). Now, the Nutrition, Physical Activity and Obesity Prevention Program at the Texas Department of State Health Services has released the Strategic Plan for the Prevention of Obesity in Texas 2005-2010. The goals of the plan are to:

- Increase awareness of obesity as a public health issue that impacts the quality of life of families.

(Continued on page 6)

Little People, Big World, Rare Defect

Little People, Big World is a documentary series aired on The Learning Channel (TLC) that looks at the day-to-day life of the Roloff family. This family is composed of parents who are each little people, and their four offspring, including twin boys. Three of the children are normal sized, and one of the twins is also a little person.

According to the Gallup organization, the series is viewed by 1.3 million adults weekly. The popularity of the show may have greatly increased awareness of the successes and challenges of an otherwise average family dealing with dwarfism.

The dad, Matt Roloff, is affected by diastrophic dysplasia. According to TLC, this condition is the third-most-common cause of short stature (one per 110,000 births), and negatively affects bone and joint structure and leads to broad, short fingers. His wife Amy and son Zach, however, have achondroplasia, which is much more common (about one per 26,000 to 40,000 births) and results in disproportionately short arms and legs.

Among 1999-2002 deliveries in the Texas Birth Defect Registry there were 46 cases of achondroplasia, and 3 babies affected by diastrophic dysplasia (out of 1,449,943 live births).

More information about this show can be found at <http://tlc.discovery.com/fansites/lpb>.

(Continued from page 5)

- Mobilize families, schools, and communities to create opportunities to choose lifestyles that promote healthy weight.
- Promote policies and environmental changes that support healthful eating habits and physical activity.
- Monitor obesity rates and related behaviors and health conditions for planning, evaluation and dissemination activities.

A free copy can be printed from the website at www.dshs.state.tx.us/phn/obesity.shtm. For more information, call 512-458-7200.

CORNELIA DE LANGE SYNDROME VIDEO

A free video about Cornelia de Lange Syndrome (CdLS), "Find One Child", can be ordered from the CdLS-USA Foundation by calling 1-800-753-2357 or email at awareness@cdlsusa.org. The web site is www.cdlsusa.org.

Note: The birth prevalence of CdLS in Texas is about 0.06 per 10,000 live births, with 9 cases identified among deliveries during 1999-2002.

CDC ISSUES PRECONCEPTION HEALTH CARE RECOMMENDATIONS

The Centers for Disease Control and Prevention (CDC) has released national recommendations encouraging women to take steps toward good health before becoming pregnant. The recommendations on preconception health and health care identify more than a dozen risk factors and conditions that require interventions before pregnancy to be effective. Among the topics addressed are folic acid supplementation; detecting and treating existing health conditions; reviewing medications; stopping smoking and eliminating alcohol; family planning counseling to avoid unplanned pregnancies; and counseling on behaviors related to weight, nutrition, exercise and oral health. These recommendations were published in the Morbidity and Mortality Weekly Report (MMWR) and may be accessed at www.cdc.gov/mmwr/preview/mmwrhtml/rr5506a1.htm.

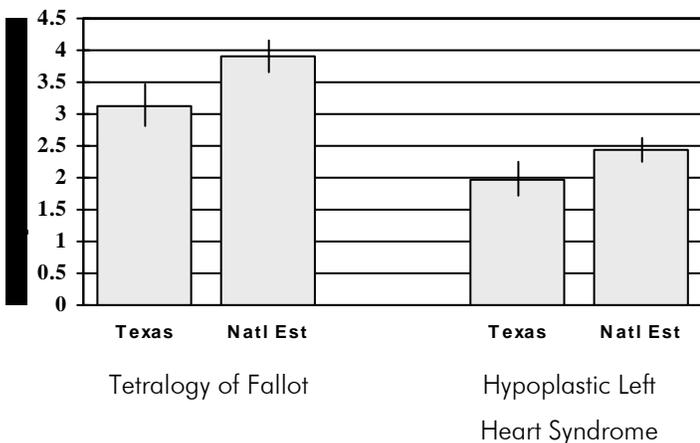
FAS CORNER

REPORT SUMMARIZES RECOMMENDATIONS CONCERNING CHILDREN AND DIAGNOSIS OF FETAL ALCOHOL SYNDROME

Guidelines for Identifying and Referring Persons with Fetal Alcohol Syndrome updates and refines diagnostic and referral criteria for fetal alcohol syndrome (FAS), incorporating recent scientific and clinical evidence. The report, published in the October 28, 2005, issue of Morbidity and Mortality Weekly Report Recommendations, summarizes diagnostic guidelines formulated by a scientific working group convened by the Centers for Disease Control and Prevention in coordination with the National Task Force on Fetal Alcohol Syndrome and Fetal Alcohol Effect, and other organizations concerned with FAS. The report is intended for use by health professionals, policy-makers, and others in facilitating early identification of individuals affected by prenatal alcohol exposure so that they and their families can receive services. The report is available at www.cdc.gov/mmwr/preview/mmwrhtml/rr5411a1.htm.



Figure 2: National prevalence estimates compared to Texas rates for two defects, 1999-2001



(Continued from page 1)

nal age distribution of all live births in the United States during 1999-2001 to obtain national prevalence estimates (See Figure 1).

The average prevalence ranged from 0.82 per 10,000 live births for truncus arteriosus to 12.94 for Down syndrome. Most estimates clustered near the 11-state average estimate; however, variation was observed between states for each defect. For example, hypoplastic left heart syndrome ranged from 1.16 per 10,000 live births in the state with the lowest prevalence to 3.75 in the state with the highest prevalence; cleft palate ranged from 3.89 per 10,000 live births in the state with the lowest prevalence to 9.65 in the state with the highest prevalence. Variation might have occurred for several reasons, including 1) differences in surveillance ascertainment methods, 2) differences in maternal risk factors, such as smoking or nutrition during pregnancy, 3) differences in the racial/ethnic composition of the population for defects that vary by race/ethnicity, 4) differences between urban and rural settings.

Nearly one baby in every 10 born in the United States is born in Texas, so naturally Texas contributed a relatively large number of cases to this study. Thus, it is not surprising to see that national rates in this study are very close to those in Texas. In fact, the national estimates varied significantly for only two of the defects in Texas during the same time period (See Figure 2.)

An additional manuscript is underway using these same data to analyze rates of various defects by racial and ethnic categories.

RESEARCH CENTER

2006 SYMPOSIUM HIGHLIGHTS

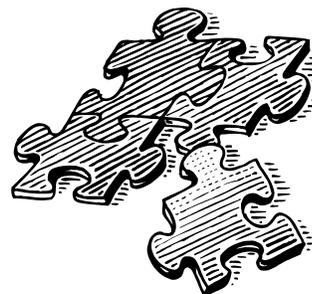
On April 19, 2006, the Texas Birth Defects Research Symposium was held in the Austin Hilton in Austin, TX. Over 130 participants including public health professionals, nurses, and those in the fields of academics and genetic counseling gathered to attend lectures given by 13 of their

colleagues. Speakers came from across the state of Texas, as well as from the University of Alabama at Birmingham, the University of Oklahoma Health Sciences Center and the Centers for Disease Control and Prevention, to present varied topics relating to birth defects and prevention. Topics addressed include:

- *Mosquito Control Pesticides and Genitourinary Malformations* (Jennifer Peck, Ph.D., University of Oklahoma Health Sciences Center Department of Biostatistics and Epidemiology)
- *Prepregnant Obesity and Risk for Structural Birth Defects* (Kim Waller, Ph.D., University of Texas Health Science Center at Houston School of Public Health)
- *Maternal Residential Proximity to Industrial Facilities and Hazardous Waste Sites and Selected Birth Defects in Texas* (Jean Brender, Ph.D., Texas A&M School of Rural Public Health Department of Epidemiology and Biostatistics)
- *Assisted Reproductive Technologies and the Risk for Adverse Pregnancy Outcomes* (Sonja Rasmussen, M.S., M.D., Division of Birth Defects and Developmental Disabilities at the Centers for Disease Control and Prevention)
- *Exposure to Fumonisin and the Occurrence of Neural Tube Defects along the Texas-Mexico Border* (Lucina Suarez, Ph.D., Texas Department of State Health Services, Epidemiology & Disease Surveillance Unit)
- *Are Birth Defects in Texas Higher Along the Mexican Border?* (Peter Langlois, Ph.D., Birth Defects Epidemiology & Surveillance, Texas Department of State Health Services)
- *Changes in the Birth Prevalence of Selected Birth Defects after Grain Fortification with Folic Acid in the U.S.* (Mark Canfield, Ph.D., Birth Defects Epidemiology & Surveillance, Texas Department of State Health Services)

If you would like more information on topics covered at the symposium or would like to contact any of the speakers, please email amy.case@dshs.state.tx.us or call 512-458-7232.

-- Allison Winter, Intern, Texas A&M University.



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2006

August

4-5: 2nd Annual Texas Parent to Parent Conference, Austin. <http://txp2p.org/conference.htm>

15-19: Texas Rural Health Summit, Austin. Contact: 512-472-8921, contact@trha.org, www.trha.org/conferences.htm

September

12-14: CDC's 2006 National Health Promotion Conference, Atlanta, Georgia. Contact: Claudia Brogan Phone: 770-488-6509, CBrogan@cdc.gov

16: Scientific Symposium on Children's Health as Impacted by Environmental Contaminants, Austin. Contact: Phone: 512-657-7405. www.cehi.org/ Sarah.Jones@cehi.org.

23: Hispanic Women's Health Symposium, Amarillo. Contact: 806-356-4617.

25-27: 4th Annual Public Health Information Network Conference, Atlanta, GA. Contact: Barb Nichols, Phone: 404-639-7600, phin2006@cdc.gov. www.cdc.gov/phin

National Fruit & Vegetable Month

October

National Down Syndrome Awareness Month

National Spina Bifida Awareness Month

2007

January

National Birth Defects Prevention Month

8-14 National Folic Acid Awareness Week

The Annual Meeting of the National Birth Defects Prevention Network will be held in Texas in February 2007! Check www.nbdpn.org for details.