

# Texas Birth Defects Monitor

Texas Birth Defects Monitoring Division

Texas Department of Health

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## *From the Director*



I am pleased to announce three significant developments in the past six months. The first is the completion of the pilot phase of the Texas Birth Defects Registry. The second is the hiring of the management teams for regions recently added to the Texas Birth Defects Registry. Lastly, the Texas Birth Defects Monitoring Division has been awarded a grant by the Centers for Disease Control and Prevention to establish the Texas Birth Defects Research Center (see below).

Staff in Regions 6 (Houston) and 11 (South Texas) have completed all case ascertainment and abstraction activities for the pilot phase of the Registry. The pilot consisted of selected major structural malformations occurring among hospital births in 1994 and 1995 to residents of Public Health Regions 6 and 11. Staff are currently analyzing data and compiling prevalence figures from the Registry. In addition, pilot phase referral data are becoming available (see page 2), and the pilot as a whole is being evaluated. Both regions have begun 1996 case ascertainment and abstractions. The case definition has expanded considerably in 1996 to include a more complete list of structural malformations. Other changes include additional data sources, including clinics and birthing centers.

The Arlington, San Antonio, and El Paso regional offices have been opened in expansion regions 2/3, 8, and 9/10, respectively. Management and support staff are in place, and field staff will come on board by mid-February. The new regions will begin surveillance activities with 1997 deliveries.

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## *Texas Birth Defects Research Center Established*

Thanks in part to a strong and growing birth defects registry, the Centers for Disease Control and Prevention have awarded Texas a grant to establish the Texas Birth Defects Research Center. This is one of six centers in the U.S. to conduct collaborative national and local studies. The overall goal is to find the causes of birth defects.

We will contribute to a national research study (the Birth Defects Risk Factor Surveillance Program, or BDRFSP) by conducting interviews with parents of Texas children with selected birth defects, as well as with parents of unaffected children. By examining cases from six states, we will be able to complete studies on rare conditions much more quickly and with more subjects than was ever possible before.

We will also be conducting several studies of local interest. One such study will closely examine genetic and environmental factors involved in neural tube defects and other birth defects observed to be higher in Hispanics. A second study will examine obesity as a risk factor for birth defects. These first two case-control studies will utilize telephone interviews and the collection of biological and environmental samples from selected subjects. A third local effort will provide funding for a research initiative identified by the ongoing Texas Neural Tube Defect Project, which works along the US/Mexico border. It will examine pollutant exposure as estimated by geographic information systems. The final local study will be a telephone survey to examine the knowledge, attitudes, and behavior of Texas women of childbearing age about factors related to birth defects (see page 3). Local study collaborators will include epidemiologist Kim Waller, Ph.D., at the University of Texas School of Public Health and geneticist Rick Finnell, Ph.D., at the Texas A&M University College of Veterinary Medicine.

### ***International Conference on Fetal Alcohol Syndrome***

At a recent workshop on fetal alcohol syndrome (FAS) in Seattle, an important report by Ann Streissguth, Ph.D., and colleagues at the University of Washington School of Medicine, entitled "Understanding the Occurrence of Secondary Disabilities in Clients with Fetal Alcohol Syndrome (FAS) and Fetal Alcohol Effects (FAE)" was presented. This study examined 415 persons with FAS/FAE between the ages of 6 and 51, focusing on risk and protective factors for secondary disabilities (mental illness, disrupted school experience, trouble with the law, confinement, inappropriate sexual behavior, alcohol and drug problems). Two findings were highlighted. Children with FAS/FAE who live in a more stable environment, as well as those who receive a diagnosis before 6 years of age, are *less*

likely to have secondary disabilities. In addition, children with FAE or higher IQs are *more* likely than those with FAS or lower IQs to have several of these secondary disabilities, presumably because the FAS group experiences more *primary* disabilities. A limited number of copies of this report are available by contacting Mary Ethen, M.P.H., FAS Coordinator, Texas Birth Defects Monitoring Division: (512) 458-7232.

Susan Astley, Ph.D., described her recently published study in which computer analyses of facial photographs accurately distinguished between individuals with and without FAS. This study demonstrated the potential of this approach for developing screening, diagnostic, and surveillance tools, even for adults. For more information, refer to the following: *Astley SJ and Clarren SK. A Case Definition and Photographic Screening Tool for the Facial Phenotype of Fetal Alcohol Syndrome. Journal of Pediatrics 1996; 129:33-41.*

### ***Referral to Services***

As required by the Birth Defects Act, the Texas Birth Defects Monitoring Division provides referral services and information to the children and families identified through its surveillance system. The law states that "a child who meets the medical criteria prescribed by board rule, and the child's family, shall be referred to the department's case management program for guidance in applying for financial and/or medical assistance."

Children identified through the surveillance system are referred to the Texas Department of Health's Social Services Regional Director. Regional social work staff then check to see if the child is already enrolled in services such as Medicaid or the Chronically Ill and Disabled

Children (CIDC) program, a program which provides assistance to children of limited financial resources for a variety of medical procedures and/or medical devices as needed by the child.

Children who are eligible for services and not already enrolled receive assistance in applying for services. If the child is not eligible for services, the Texas Birth Defects Monitoring Division mails a letter to the parent or guardian of the child including a list of general resources and literature describing additional programs.

There are only a few situations in which a child or family may not be considered for services by the regional Social Work Services Division. These include a deceased child or a child with a lethal condition whose prognosis is less than one-month survival, a pregnancy termination, or a stillbirth or late fetal death (20 weeks gestation or greater). In these situations, the assistance of hospital social workers has been requested to provide the family with referrals to services such as support groups or genetic counseling.

This referral process was recently pilot tested in Region 11 (South Texas) using cases identified among children born during 1995. The results of this pilot indicate that many children are being referred to services for the first time as a result of this process.

### *Folic Acid Developments*

**Grain Fortification:** The U.S. Food and Drug Administration has announced that grains will be fortified nationally with folic acid, for the purpose of preventing neural tube defects (NTDs). This will be implemented no later than January 1, 1998. The fortification level, 140 micrograms/100 grams of grain, is less than what had been recommended by the Texas Department of Health and the Centers for Disease Control and Prevention for optimal NTD prevention. At this level, it is estimated that only 4 to 20% of the folic

acid-preventable NTD cases can be prevented. Therefore, we need to move ahead with strategies to increase folic acid consumption statewide.

**Multivitamin Supplements:** In March 1996, the Division of Women's Health at the Texas Department of Health sent out a survey to all of their family planning and prenatal care providers. These providers are agencies contracted with the State to provide services funded by Title V, Title XIX, Title X, and Title XX. The survey included information on the benefits of folic acid and the opportunity to receive multivitamins with 0.4 mg of folic acid free of charge. After receiving responses from most of the providers, the Texas Department of Health distributed 222,220 bottles of multivitamins (100 vitamins per bottle) to 65 agencies across the state. This quantity is sufficient to cover 55,555 women of child bearing age for a complete year. The agencies were also given sample brochures with information about folic acid, as well as instructions for reordering.

**Women's Health Survey:** The Texas Birth Defects Research Center at the Texas Birth Defects Monitoring Division is currently designing a telephone survey to determine the knowledge, attitudes, and behaviors of Texas women about factors related to birth defects. Examples include taking vitamin supplements containing adequate folic acid, avoiding alcohol during pregnancy, using prenatal diagnostic services, and pre-pregnancy planning. The anonymous survey will be conducted statewide among women of childbearing age. It will be repeated periodically (currently scheduled to be every three years) to assess changes in knowledge, attitudes, and behaviors over time. These surveys will provide particularly useful information for folic acid education efforts.

## *Birth Defects in Texas*

**The Problem:** Birth defects are abnormalities of structure, function, or body metabolism which are present at birth, and which often result in physical or mental handicap, or death. It is estimated that serious birth defects occur in 1 out of every 33 live births. Some of the most common birth defects are heart defects (which occur in 1 out of every 200 live births), cleft lip/palate (1 out of every 550 live births), Down syndrome (1 out of every 900 live births), neural tube defects (1 out of every 1,400 live births), and limb defects (1 out of every 1,750 live births).

It is estimated that over 120,000 children are born annually with serious birth defects in the United States. Birth defects are the leading cause of infant mortality in the United States, directly causing over 20% of all infant deaths. In addition, they are the fifth leading cause of premature death among all persons less than 65 years of age. In 1988, over 670,000 years of potential life were lost in the United States due to birth defects. Birth defects are also a leading cause of lifetime disability.

Known causes of birth defects include both genetic factors and environmental factors. Genetic factors include autosomal and sex-linked genetic diseases, new mutations, and chromosomal abnormalities. Environmental factors include maternal conditions (such as diabetes), infectious agents (such as rubella), and mechanical problems (such as amniotic band constrictions). Other environmental factors include chemicals, drugs, radiation, and hyperthermia. Alcohol use during pregnancy is the most common known cause of birth defects. The cause of 65-75% of birth defects is still unknown. Many of the birth defects for which a cause is not known are thought to be multi-factorial, that is, they involve both genetic and environmental factors.

**Texas Statistics:** It is estimated that nearly 10,000 children are born with serious birth defects in Texas every year. Birth defects are the leading cause of infant mortality in Texas, directly causing 25% of all infant deaths. Birth defects are the third leading cause of death among 1-14 year olds in Texas, and are the seventh leading cause of premature death among all Texans.

Since Texas does not currently have a statewide birth defects registry, there are little reliable data available on birth defects in Texas. In Cameron county in 1991, an unexplained cluster of anencephaly, a fatal birth defect characterized by partial or total absence of the brain, highlighted the need for accurate and timely information on the scope and magnitude of birth defects in Texas. Without a statewide registry, Texas must rely on other less reliable sources for birth defects data. One example is vital statistics data. While information on birth defects has been collected on live birth, fetal death, and death certificates for several years, these data have generally proven to be a poor primary source of information for most birth defects.

**The Cost of Birth Defects:** Birth defects morbidity costs billions of dollars per year for health care and special education in the United States. Children with birth defects account for 25-30% of all pediatric hospital admissions. While cost data are not available for all birth defects, estimates have been made for certain defects. For example, it is estimated that the lifetime per-patient cost for spina bifida is approximately \$290,000. The cost of medical and surgical treatment alone for those with spina bifida and similar conditions is estimated at \$200 million per year in the United States. In Texas, the estimated 161 children born with spina bifida every year result in a total lifetime cost of over \$46 million. The lifetime per-patient cost for Down Syndrome is estimated

to be over \$450,000. In Texas, the estimated 353 children born with Down Syndrome every year result in a total lifetime cost of \$158 million. The lifetime per-patient cost for Fetal Alcohol Syndrome (FAS) is estimated to be \$240,000. In Texas, the estimated 600 children born with FAS every year result in a total lifetime cost of \$144 million.

**Prevention & Intervention:** Current birth defect prevention measures include vaccines which can prevent infectious diseases that cause birth defects. An example of such a disease is rubella. Congenital rubella syndrome can cause central nervous system defects, deafness, heart defects, thyroid disorders, and even death. Recently, evidence has emerged that folic acid supplementation can prevent some neural tube defects. The Centers for Disease Control and Prevention (CDC) now recommends that all women of childbearing age consume 0.4 mg of folic acid daily. CDC estimates that following this recommendation may reduce the occurrence of neural tube defects by as much as 50%. Other prevention measures include the use of immune globulin to prevent birth defects caused by Rh disease. The use of immune globulin prevents problems in 95-99% of cases, thereby reducing the need for high cost transfusions and neonatal intensive care.

Birth defects monitoring and research can identify clusters, study patterns to determine causes, and develop prevention strategies. For example, it was a study of birth defect clusters that led to the discovery of FAS. Having now determined the cause of FAS, efforts can be taken to prevent it by discouraging maternal alcohol use during pregnancy. Birth defects monitoring also allows affected children to be identified and referred to services. For many defects, this early intervention is critical to the long term prognosis of the child.

In 1993, the 73rd Texas Legislature passed

Senate Bill 89, which directed the Texas Department of Health to develop and maintain a statewide birth defects registry. The Texas Birth Defects Monitoring Division was created to set up and maintain the registry and to investigate clusters of birth defects throughout the state. At that time, the Legislature provided funding for a pilot project which included counties in the lower Rio Grande Valley and most of the Texas Gulf Coast area. In 1995, the Texas Legislature appropriated funds for partial expansion of the registry. However, until additional funds are available to expand the registry statewide, responding to birth defects clusters in non-registry areas will continue to be problematic. Additionally, the Texas Birth Defects Monitoring Division cannot identify and refer to services those children born with birth defects in non-registry areas.

**Cost Savings:** Rubella immunizations save \$354,000 for each case of congenital rubella syndrome prevented. A 50% reduction in the number of children born with spina bifida in Texas every year would result in a savings of \$20 million over the lifetime of these individuals. The prevention of FAS, a completely preventable birth defect, could save Texans an estimated \$144 million per year.

Preventive measures, proper diagnosis, appropriate referral and treatment, and early intervention can, in many cases, improve long-term outcomes and reduce premature mortality. This allows affected children to lead a more productive life, thereby lowering the costs associated with these defects.

Note: References for this article are available upon request.



## *Case-Control Study Update from the Texas Neural Tube Defect Project*

The primary goal of the Texas Neural Tube Defect Project (TNTDP) is the prevention of the recurrence of neural tube defects (NTDs) among women of the Texas-Mexico border area. The case-control study is one of the three components of the TNTDP. (The other two are surveillance and intervention.) The purpose of the case-control study is to find risk factors for NTDs by comparing the environmental, biological, social, and dietary aspects of the women with NTD-affected pregnancies to those of women who have had a healthy child.

For the case-control study, case-women are those identified through surveillance who resided in the 14-county study (Texas-Mexico border) area at the time of delivery or termination of their NTD-affected pregnancy, and who continue to reside in the area at the time of the case-control study. Two population-based, nonmatched control-women are selected for each case-woman; control-women are those with non-NTD-affected pregnancies. The study protocol includes a Mother Questionnaire; a Food Frequency Questionnaire; a Biomarker Questionnaire; and laboratory tests.

The Mother Questionnaire was designed specifically for the TNTDP, but is based largely upon the Birth Defects Mother Questionnaire used by the Centers for Disease Control and Prevention. This questionnaire is 165 pages long and covers such items as maternal health, reproductive history, family demographics, and such sensitive issues as drug use (including alcohol, street drugs, and inhalants) and sexually transmitted diseases. The database for the Mother Questionnaire contains 6,000 variables.

The Food Frequency Questionnaire was

developed by the University of Texas School of Public Health and is based upon a detailed analysis of one hundred 24-hour dietary recalls obtained from border residents. This questionnaire considers all of the basic food groups and nutrients, but is weighted with regard to its emphasis on folic acid intake. Also a part of the dietary analysis is a test of tortilla samples for fumonisins, which are toxins produced by certain molds and commonly found in corn.

The Biomarker Questionnaire addresses environmental and occupational exposures. Also a part of the environmental analysis are screening tests for total nitrate/nitrite level in drinking water and Global Positioning System coordinates identifying possible residences of exposure.

Laboratory tests are performed on 1) maternal, paternal, and infant blood specimens, 2) maternal stool specimens (to check for ova and parasites), 3) maternal biomarkers (urine is the main specimen), 4) and infant tissue specimens (placenta, cerebrum, cerebellum, spinal cord, kidney, and liver).

The case-control study began in late June 1995. The study involves 21 different hospitals and hundreds of midwives along a 1,000-mile border. Through the end of September 1996, there have been 78 eligible case-women and 106 eligible control-women. "Eligible" means that these women completely met the case or control definition. Fifty-six of the 78 (72%) eligible case-women and 84 of the 106 (79%) eligible controls have been enrolled and have provided at least one of the case-control items (cord blood, placental specimen, Mother Questionnaire, or biomarker specimens). Those not enrolled represent eligible participants whose information is pending or who have moved, are lost, or have refused enrollment.

The prevalence of various exposures, intakes, and conditions will be compared between case- and control-women and odds ratios and 95% confidence limits will be calculated for all risk estimates. Mean levels and percent above or below normal levels of the various biological analytes and markers will be compared between case- and control-women.

For a copy of the most recent report from the Texas Neural Tube Defect Project, please call (512) 458-7328.

### *Announcements*

**Fetal Alcohol Syndrome and Neural Tube Defects Prevention Seminar:** A full day seminar entitled "Fetal Alcohol Syndrome and Neural Tube Defects: Prevention is the Answer" will be held April 9, 1997, in Corpus Christi. Sponsored by the University of Texas Health Science Center at San Antonio and the Texas Department of Health, the seminar is targeted toward nurses, educators, social workers, counselors and other health care related staff. Participants will learn about fetal alcohol syndrome/fetal alcohol effect and neural tube defects, and how to assess, educate, and refer clients. Continuing Nursing Education (CNE) type I credits are being applied for. There are no registration fees for this seminar. To register, call Adele Fuentes at (512) 883-9288, ext. 2256. Deadline for registration is March 19, 1997.

**Forum on Neural Tube Defects:** A scientific and community forum on Neural Tube Defects in South Texas is being planned for Brownsville, Texas, in April 1997. For more information, contact Sandy Wicker in Austin at (512) 458-7232 or Jorge Trevino in Harlingen at (210) 444-3204.

### *Useful Phone Numbers*

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Note: References available upon request.

**Recent Articles:** If you have not already seen them, you may be interested in two recent journal articles on folic acid:

- *Cuskelly GJ, McNulty H, Scott JM. Effect of Increasing Dietary Folate on Red-cell Folate: Implications for Prevention of Neural Tube Defects. Lancet 1996;347:657-659.* In an intervention study in the U.K., this group of researchers found that supplements and fortified food were effective at increasing the blood folate level of study participants, but consumption of extra folate through diet (as natural food folate) was not.

- *Daly LE, Peadar N.K., Molloy A, Weir DG, Scott JM. Folate Levels and Neural Tube Defects: Implications for Prevention. JAMA 1995;274(21):1698-1702.* From a case-control study in Ireland, these researchers found that a woman's risk for having an NTD-affected pregnancy was associated with red blood cell folate levels in early pregnancy in a continuous inverse dose-response relationship. They concluded that supplementation with vitamins should be targeted to high-risk women who are identified through blood folate testing. On the other hand, food fortification is preferable for an overall population-based NTD prevention approach.