National Center on Birth Defects and Developmental Disabilities

Strategic Plan 2011-2015

February 2011

National Center on Birth Defects and Developmental Disabilities
Office of the Director

CDC's National Center on Birth Defects and Developmental Disabilities
Strategic Plan
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Our Vision and Priorities

The National Center on Birth Defects and Developmental Disabilities (NCBDDD) works to advance the health and well-being of our nation's most vulnerable populations. Our focus on women, children, people with a range of disabilities and complex disabling conditions positions us as a resource within public health that is unique and vital.

As we approach our ten year anniversary, a reflection on what we have accomplished and what we plan to achieve over the next five years is necessary. This plan describes our vision for the upcoming years and our commitment to achieve important health outcomes during that period.

This plan is intended to help guide our own work, but we have also used its creation as a tool to engage our partners and to solicit their input, expertise and commitment to move our collective efforts forward in meaningful and measurable ways.

As we have created this plan, we have made the following assumptions:

- Funding for current programs and activities will not increase substantially in the next three years given the current economic environment
- Staffing levels within the center will remain relatively stable
- Evolving trends within government and the private sector have important implications for our work
- Opportunities to retool and reframe current programs are based on evidence-based information

In a few instances, we have proposed activities and outcomes that will require resources beyond what is currently available. However, we have weighed these circumstances in light of disease burden, clear opportunities to intervene, and health impact.

Note: Throughout this plan, various acronyms and abbreviations are used. Please refer to Appendix I for a complete list of acronyms.

Our Background and History

Established by the Children's Health Act of 2000, CDC's National Center on Birth Defects and Developmental Disabilities (NCBDDD) is currently organized into three divisions: Birth Defects and Developmental Disabilities (DBDDD), Blood Disorders (DBD), and Human Development and Disability (DHDD). Congress mandated the creation of the Center in response to strong advocacy efforts by external organizations whose view was that children's health issues were not being adequately addressed by CDC. Prior to formation of NCBDDD, CDC conducted an extensive internal assessment. This assessment focused on existing maternal-child health programs and it made recommendations about critical functions, programs and structure. The initial structure selected was the least disruptive to the Agency while also fulfilling the congressional mandate. It included the activities of two of the current divisions: Human Development and Disability and Birth Defects and Developmental Disabilities. It was not until 2004 that the Division of Blood Disorders was transferred to the Center from the National Center for Infectious Diseases. The rationale for moving Blood Disorders to NCBDDD was based upon the fact that the Center was already working on a number of chronic disability conditions with a strong genetic component.

Clearly, the Center's name does not accurately capture the breadth of our work. For example, though each division has a focus on children, as we have gained a deeper understanding of our populations and as programs have matured, we come to realize that our perspective must be one of a life course approach. In addition, as a Center, we consider health in the context in which people live and value health equity and inclusion for all groups and individuals.

Similarly, our work with individual disorders is leading us to adopt a common approach in which we are building a public health framework that promotes quality of life for all rather than approaching health issues disorder by disorder. In this way, we are working to build a model for a whole array of disabling conditions. Nevertheless, we recognize that many of the conditions that we address do not easily lend themselves to a "one size fits all" approach. Both advocates and experts alike push for programs tailored to address the special needs of individuals with relatively rare conditions. Our challenge is to balance this desire with economic realities. The economic and long-term societal costs of rare disorders are substantial. For example, in the United States, 20,000 people have hemophilia, yet the annual healthcare cost for them is $3 billion. Sickle Cell Disease affects 100,000 people in the U.S. at an annual cost of $1 billion.

Our rationale for focusing on a life course approach is illustrated by the dilemma encountered by many people with disabilities (PWD). As they lead ever more robust and longer lives through the advent of health and environmental advances, transition issues from one life stage to another become increasingly more important. For example, children with chronic conditions/disabilities like Sickle Cell Disease, congenital heart defects or spina bifida have historically been treated by pediatricians, but with ever-increasing life spans, adolescents and young adults are transitioning from the pediatric to the adult healthcare system. Adult healthcare providers may not have the training, specialty knowledge or equipment to adequately address the needs of these individuals. Therefore, the issue of transition from one provider type, practice setting or specialty facility is important in terms of long-term health outcomes for these populations. Similar circumstances occur for individuals with fetal alcohol spectrum disorders and autism spectrum disorders. In order to maximize their long-term health, we must develop and implement strategies to fundamentally improve these transitions through education, policy and systems change.

In addition to this example, a few examples of each division's activities by life course are also provided:

Examples of NCBDDD Activities by Life Course

	Pre- & peri-conception	**Infancy & early childhood**	**Youth & young adulthood**	**Adulthood**
DBDDD	Research on the fetal effects of medications used during pregnancy	Surveillance on the prevalence of birth defects and developmental disabilities	Long-term follow up of children with birth defects including spina bifida, congenital heart disease and Down Syndrome	
DBD	Women's health - awareness and education on bleeding and clotting disorders	Early identification and referral for hemoglobinopathies	Treatment centers focusing on healthcare quality issues of people with hemophilia	Chronic disease risk factor reduction efforts for persons with blood disorders
DHDD		Early identification and referral for congenital hearing loss	Longitudinal studies and follow-up of adolescents with spina bifida, muscular dystrophy, ADHD and other disabling conditions	Assess health status and risks of persons with disabilities; promote health care access and healthy lifestyles

The Division of Birth Defects and Developmental Disabilities focuses on surveillance, public health research and primary prevention of birth defects and developmental disabilities. The Division of Human Development and Disability's work focuses on secondary prevention with a strong emphasis on quality of life issues for persons with disabilities. The Division of Blood Disorders' focus is on primary and secondary prevention with a strong educational component for adults of reproductive age, providers and patients with blood disorders.

Our Partners and Constituents

NCBDDD works collaboratively across federal agencies and other partner organizations to leverage work and services being carried out by the Center and our grantees. For example, we work closely with HRSA's Title V MCH block grant recipients, organizations funded through Department of Education programs, developmental disabilities councils, pediatricians, hematologists, and a variety of disability-oriented organizations. Each of these groups amplify the work of NCBDDD by using our science and programs to inform and guide their work.

A strong relationship with other federal agencies, including NIH, HRSA, and SAMHSA has evolved over the past 40 years. NCBDDD works closely with several of NIH's Institutes, including NICHD, NIMH, NINDS, NIAAA and NHLBI. In addition, we work with state health departments, non-governmental organizations and specialty organizations. Our unique contribution to these relationships is the public health perspective – including the provision of data from population-based surveillance, the focus of our research on major preventable risk factors, and the rapid translation of research findings to prevention programs.

Though some of our activities are designed to address the *needs of vulnerable **individuals***, we clearly recognize our mandate to address the *needs of vulnerable **populations***. For example, in the area of emergency preparedness, our extensive knowledge of these vulnerable groups and their specific needs in an

emergency such as the influenza pandemic is a high priority area for our work. We focus on policies that address those needs and work to ensure that existing programs consider the unique requirements of these populations. As a Center, our philosophy is consistent with Hubert Humphrey's famous quotation:

"It was once said that the moral test of the government
is how that government treats those who are in the dawn of life, the children;
those who are in the twilight of life, the elderly;
and those who are in the shadows of life, the sick, the needy and the handicapped."

Our Mandates and Authorities

In addition to operating under various types of authorities (general, specific and shared) NCBDDD receives Congressional committee language annually that specifies Congress' intent regarding how funds are to be expended. For example, some of the specific authorities for the center direct the Center to work on particular programmatic activities, including Fetal Alcohol Syndrome, folic acid, and Tourette's Syndrome to name a few. In some cases, authorities for a particular area of work may reside in specific and shared authorities. This is the case with autism.

In general, annually each CDC Center receives language after the passage of appropriations law, and NCBDDD leaders work closely with staff in CDC's OD and in the Financial Management Office to ensure that the Center is responsive to Congress and compliant with these mandates and authorities. A list of the Center's general, specific and shared authorities are provided in Appendix II.

Our Public Health Approach, Responsibility to Our Constituents

As the Center begins its second decade, we thought an in-depth reflection of what we have accomplished, how we have operated, and what we want to accomplish in the next few years was in order. This strategic planning process has allowed us to accomplish this work, and most importantly, to identify promising new areas ripe for active engagement. In creating this plan, we identified our priorities by focusing on issues that share these characteristics:

1. The magnitude of the problem is known and significant.
2. Opportunities for prevention, intervention and improvement are present.
3. Several conditions can be improved based on evidence-based interventions and strategies.
4. Disparity reduction/amelioration is a central aspect of what we work to accomplish.
5. The priorities are reinforced or supported by CDC-wide priorities, and clearly fit within our Center's mandate.
6. The priorities build on previous work or known scientific evidence.

Center leaders led the strategic planning process, and at key stages of the work subject matter experts from throughout the Center were brought into discussions. Once the plan was drafted, a thorough vetting process with internal staff and key partners was undertaken. Input into the draft plan was actively sought and each recommendation was tracked, considered, and in almost every case included in this version of the plan.

Subsequent reviews of the plan will be conducted within (and throughout) CDC, and implementation plans for our cross-cutting priorities will be developed within the next year.

Specifically, our strategic planning discussions confirmed that, in carrying out our work, **NCBDDD strives to:**

- Characterize the problem, incidence, prevalence, and distribution of our Center's priority health conditions to inform public health research, priority setting and program monitoring;
- Conduct epidemiological research to understand the major modifiable risk factors in order to develop intervention/prevention programs and policies; and
- Develop, evaluate and disseminate effective programs and policies for adoption by global, national, state and local organizations.

Success in these population-based activities will result in:

- Prevention of birth defects and developmental disabilities;
- Health promotion and inclusion of people with disabilities;
- Prevention of chronic disease and subsequent mortality in people with our targeted conditions;
- Effective population level management of a number of existing conditions including blood disorders; and,
- Preparedness for disasters and other potential catastrophic health events.

With these perspectives in mind, the Center's three priorities are to:

1. Prevent major birth defects attributable to maternal risk factors.

2. Prevent death and disability due to deep vein thrombosis (DVT) and pulmonary embolus (PE).

3. Reduce disparities in obesity and other health indicators in children, youth and adults with disabilities.

Relationship between Center Priorities and Relevant Healthy People 2020 Goals

Center Priority 1: Prevent major birth defects attributable to maternal risk factors encompasses several Healthy People 2020 goals. These include:

MICH-1.6 Infant deaths related to all birth defects

Baseline:	1.4 infant deaths per 1,000 live births were attributed to birth defects (all birth defects) in 2006
Target:	1.3 infant deaths per 1,000 live births
Target-Setting Method:	10 percent improvement
Data Source:	National Vital Statistics System (NVSS), CDC, NCHS

MICH-1.7 Infant deaths related to birth defects (congenital heart defects)

Baseline:	0.38 infant deaths per 1,000 live births were attributed to congenital heart and vascular defects in 2006
Target:	0.34 infant deaths per 1,000 live births

Target-Setting Method:	10 percent improvement
Data Source:	National Vital Statistics System (NVSS), CDC, NCHS

MICH-11.1 Alcohol

Baseline:	89.4 percent of pregnant females aged 15 to 44 years reported abstaining from alcohol in the past 30 days in 2007–08
Target:	98.3 percent
Target-Setting Method:	10 percent improvement
Data Source:	National Survey on Drug Use and Health (NSDUH), SAMHSA
Baseline:	95.0 percent of pregnant females aged 15 to 44 years reported abstaining from binge drinking during the past 30 days in 2007–08
Target:	100 percent
Target-Setting Method:	Total coverage
Data Source:	National Survey on Drug Use and Health (NSDUH), SAMHSA

MICH-15 Reduce the proportion of women of childbearing potential who have low red blood cell folate concentrations

Baseline:	24.5 percent of non-pregnant females aged 15 to 44 years did not have low red blood cell folate concentrations in 2003–06
Target:	22.1 percent
Target-Setting Method:	10 percent improvement
Data Source:	National Health and Nutrition Examination Survey (NHANES) CDC, NCHS

Center Priority 2: Prevent death and disability due to deep vein thrombosis (DVT) and pulmonary embolus (PE) relates to the following Healthy People 2020 goals:

BDBS-12 Reduce the number of persons who develop venous thromboembolism (VTE)

Baseline:	54.3 persons per 10,000 population aged 18 years and older developed venous thromboembolism (VTE) in 2007 (age adjusted to the year 2000 standard population)

Target:	48.9 per 10,000 population
Target-Setting Method:	10 percent improvement
Data Source:	National Ambulatory Medical Care Survey (NAMCS), CDC, NCHS; National Hospital Ambulatory Medical Care Survey (NHAMCS), CDC, NCHS

BDBS-13 (Developmental) Reduce the number of adults who develop venous thromboembolism (VTE) during hospitalization

BDBS-13.1 (Developmental) VTE among adult medical inpatients.

Potential Data Source:	National Hospital Discharge Survey (NHDS), CDC, NCHS

BDBS-13.2 (Developmental) VTE among adult surgical patients.

Potential Data Source:	National Hospital Discharge Survey (NHDS), CDC, NCHS; Joint Commission on Accreditation of Health Care Organizations (JCAHO) survey

Center Priority 3: Reduce disparities in obesity and other health indicators in children, youth and adults with disabilities relates to:

DH2.1 Increase the number of State and the District of Columbia health departments that have at least one health promotion program aimed at improving the health and well-being of people with disabilities.

Baseline:	16 States and the District of Columbia had health promotion programs for people with disabilities in 2010
Target:	18 States and the District of Columbia
Target-Setting Method:	10 percent improvement
Data Source:	Periodic Assessment of State Health Promotion Programs, CDC, NCBDDD

DH-8 (Developmental) Reduce the proportion of people with disabilities who report physical or program barriers to local health and wellness programs

Potential Data Source:	National Health Interview Survey Supplement (NHIS), CDC, NCHS

Weight Status

NWS-9 Reduce the proportion of adults who are obese

Baseline:	34.0 percent of persons aged 20 years and over were obese in 2005–08 (age adjusted to the year 2000 standard population)
Target:	30.6 percent
Target-Setting Method:	10 percent improvement
Data Source:	National Health and Nutrition Examination Survey (NHANES), CDC, NCHS

NWS-10.4 Children and adolescents aged 2 to 19 years

Baseline:	16.2 percent of children and adolescents aged 2 to 19 years were considered obese in 2005–08
Target:	14.6 percent
Target-Setting Method:	10 percent improvement
Data Source:	National Health and Nutrition Examination Survey (NHANES), CDC, NCHS

PA-1 Reduce the proportion of adults who engage in no leisure-time physical activity.

Baseline:	36.2 percent of adults engaged in no leisure-time physical activity in 2008
Target:	32.6 percent
Target-Setting Method:	10 percent improvement
Data Source:	National Health Interview Survey (NHIS), CDC, NCHS

Specific approaches to address these Center priorities are detailed in the strategic plan, and division-by-division descriptions of how these outcomes will be achieved are provided.

Division of Birth Defects and Developmental Disabilities

Our Vision, Approach and Priorities

The Division of Birth Defects and Developmental Disabilities works to identify causes and prevention strategies for birth defects and developmental disabilities by applying a public health approach that incorporates three essential elements—surveillance or disease tracking, public health research to identify modifiable risk factors, and prevention research and programs. The division's unique expertise in surveillance and epidemiologic research on birth defects and developmental disabilities has yielded and continues to yield opportunities for prevention that promote healthy births and optimal development for all children. Public health research is critical to identify risk factors for future prevention efforts which will allow us to decrease the prevalence of these very serious disorders.

Building upon this history and approach, we have identified an ambitious, yet realistic set of goals, priorities and outcomes for the near term. By leading in these efforts, we will move the entire field of birth defects surveillance, prevention and care forward in a substantial and tangible manner.

Our Goals, Priorities and Outcomes for the Future

Over the next four years, the goals of DBDDD are to:

- Expand and enhance surveillance and tracking systems for birth defects and developmental disabilities, including follow up for longer term outcomes (e.g., survival, use of special education services, health care utilization).
- Identify and understand the preventable risk factors (e.g., use of valproic acid during pregnancy) for birth defects and developmental disabilities.
- Develop, evaluate and disseminate effective prevention strategies aimed at preventing the occurrence of birth defects and developmental disabilities.
- Develop, evaluate and disseminate programs and strategies aimed at maximizing the quality of life for individuals with birth defects and developmental disabilities.

To support achievement of these goals, DBDDD has identified the following priority objectives:

Priority 1: Enhance surveillance and research for autism and other developmental disabilities to monitor changes in prevalence and contributing risk factors, and better inform prevention policy and programs.

Priority 2: Prevent congenital heart defects and other major birth defects associated with obesity, diabetes, and medications.

Priority 3: Prevent alcohol-exposed pregnancy among reproductive age women to prevent fetal alcohol syndrome and other adverse effects of prenatal alcohol exposure.

Priority 4: Eliminate folic acid-preventable neural tube defects in the United States and globally.

Priority 5: Enhance the quality and usefulness of newborn screening data and programs.

By focusing on our goals and priorities, we intend to achieve the following outcomes:

- Decrease or eliminate birth defects and developmental disabilities occurring due to known causes (e.g., isotretinoin exposure, insufficient folic acid).
- Improve longer term outcomes of children with birth defects and autism and other developmental disabilities, and eliminate racial/ethnic disparities in these outcomes.
- Identify preventable risk factors of birth defects and developmental disabilities and develop appropriate interventions to reduce these risks.

How We Developed Our Priorities: Building on Successes

For over forty years, CDC has led the nation and world in birth defects surveillance. This surveillance platform has enabled us to understand the magnitude of the problem, to pursue meaningful research into etiology and prevention, and to develop programs to address the underlying issues. A few examples of the division's past work in surveillance, public health research and prevention are briefly described.

Characterizing the Problem through Surveillance

Surveillance of birth defects and developmental disabilities has been carried out for years using several complementary surveillance systems. Through these systems, DBDDD investigators and funded partners have pioneered techniques for population-based surveillance of birth defects and developmental disabilities. These include the Metropolitan Atlanta Developmental Disabilities Surveillance Program (MADDSP), the Autism and Developmental Disabilities Monitoring Network (ADDM), the Metropolitan Atlanta Congenital Defects Program (MACDP), and fourteen funded state-based birth defects surveillance programs, surveillance of Fetal Alcohol Syndrome, and stillbirth surveillance pilots.

Through this important, fundamental public health investment in surveillance, NCBDDD has established the critical infrastructure needed to determine that on an annual basis in the U.S. approximately 130,000 babies have a major birth defect, and over 500,000 children are diagnosed with a developmental disability. And from surveillance data, we have determined that congenital heart defects (CHDs) represent the most common birth defects, affecting nearly 1% of infants in the U.S. Population-based surveillance data has also allowed DBDDD investigators to assess survival of affected infants, and identify significant disparities in one-year survival by race/ethnicity for infants with certain birth defects such as congenital heart defects, Down Syndrome and spina bifida.

In addition, over the past ten years, we have made great improvements in our understanding of autism spectrum disorder (ASD). We have built a robust scientific infrastructure which has helped us to understand key features of ASD. For example, the ADDM Network has established that 1 in 110 eight-year-old children has an ASD and that the condition occurs in all racial, ethnic, and socioeconomic groups, but is four times more likely to occur in boys than in girls. We know that both advanced maternal and paternal age are risk factors for ASD, disparities exist in early identification and access to services for some children with ASD, and certain hazardous air pollutants such as methylene chloride, quinoline, and styrene warrant further investigation for a possible role in autism etiology.

Conducting Research into Risk and Protective Factors

Population-based surveillance data identifies cases for NCBDDD's public health research to identify preventable exposures associated with birth defects and developmental disabilities. Epidemiological research has helped us to identify pre-pregnancy maternal obesity and diabetes as important risk factors for congenital heart defects, neural tube defects, and other major birth defects. It has also helped identify the risk of birth defects associated with the use of specific medications during pregnancy such as SSRIs and certain antibiotics, thereby allowing for more informed risk versus benefit evaluation by women and their health care providers.

Some relatively common exposures play an important role in the etiology of birth defects. DBDDD's population-based public health research has helped us to identify the following key risk factors for birth defects which are modifiable and therefore provide opportunities for interventions that can have a major public health impact:

- 1 in 8 women drink during pregnancy; 1 in 50 binge drink
- Approximately 20% of women smoke in the month before pregnancy or in the first trimester of pregnancy
- Approximately 10% of babies are born to women who have diagnosed diabetes before pregnancy or develop diabetes in pregnancy and 20 – 30% are born to women who are obese at the beginning of pregnancy
- Approximately 50% of women use at least one prescription medication in pregnancy

In the area of autism, our work in characterizing the magnitude of the condition has fueled the search for risk factors and causes of ASD and has informed key policy decisions to assist families in need of access to services and treatments. However, we are at a critical juncture in our understanding of ASD. We still do not know what is causing this condition with often debilitating symptoms. We still do not know how much of the increase in prevalence is due to a true increase in risk and how much is due to extraneous factors. Over the next ten years, we are likely to make unprecedented advances in our understanding, building upon the strong infrastructure we now have in place. Recognizing that we are dealing with a serious developmental disability with unstable rates and no known cause, it is of critical importance that we amplify efforts and use all tools available to study this condition. To do this, CDC is working in concert with other Federal and private agencies through the Federal Interagency Autism Coordinating Committee (IACC). NCBDDD ASD activities provide an epidemiologic perspective to the 2010 IACC Strategic Plan addressing objectives such as:

- 7I- Supplement existing ADDM Network sites to use population-based surveillance data to conduct at least 5 hypothesis-driven analyses evaluating factors that may contribute to changes in ASD prevalence by 2012.
- 7L- Expand the number of ADDM sites in order to conduct ASD surveillance in younger and older age groups; conduct complementary direct screening to inform completeness of ongoing surveillance; and expand efforts to include autism subtypes by 2015.
- 3F- Initiate studies on at least 10 environmental factors identified in the recommendations from the 2007 IOM report "Autism and the Environment: Challenges and Opportunities for Research" as potential causes of ASD by 2012.

Developing, Implementing and Translating Feasible and Effective Interventions

DBDDD's efforts in prevention have focused on prevention of fetal alcohol syndrome and folic-acid preventable neural tube defects (NTDs). Our studies reveal that there are 0.2 to 2.0 cases of FAS for every 1,000 live births in certain areas of the U.S. Fetal Alcohol Spectrum Disorders (FASDs), including FAS, refers to a range of effects that can result in prenatal alcohol exposure. Researchers estimate that there are at least three times as many cases of FASDs as FAS alone. FASDs are 100% preventable and may cause lifelong physical, behavioral, and cognitive disorders, ranging from mild to severe. The lifetime cost for one individual with FAS in 2002 was estimated to be $2 million. NTDs remain a significant cause of infant mortality and childhood morbidity globally. There are 3,000 NTD-affected pregnancies in the U.S. each year, and lifetime medical costs for a child with spina bifida are estimated to be $560,000. Globally more than 300,000 infants are born with NTDs each year. Some successes from these two programs include:

Project CHOICES – An evidence-based screening and brief intervention program that counsels and educates women of reproductive age about the impact of drinking during pregnancy. We are currently evaluating the effectiveness of bundling a tobacco cessation intervention with Project CHOICES, and are working with SAMHSA, HRSA, IHS and the CDC STD program to implement the Project CHOICES screening and brief intervention model in the context of these various delivery systems.

Folic Acid Fortification – Surveillance and research conducted by NCBDDD and others throughout the world demonstrated the important role of periconceptional folic acid in preventing NTDs. These findings resulted in a major population-level policy intervention: mandatory folic acid fortification of enriched cereal grain products in the United States. This intervention resulted in the prevention of an estimated 1,000 NTDs per year since its full implementation in 1998. NCBDDD has also developed a global plan for elimination of folic acid-preventable NTDs, and is working closely with external partners and the FDA on a food additive petition to allow folic acid fortification of corn masa flour.

Currently global micronutrient fortification programs that include folic acid are preventing only about 9% of total annual cases of folic-acid preventable NTDs. Expanding the number of developed and developing countries with mandatory folic acid fortification of high consumption staples can potentially safely eliminate the majority of NTDs. Current research and increasing fortification efforts have demonstrated the ability to eliminate those NTDs that are sensitive to folic acid. Assuming 50%-70% of NTDs are folic acid-preventable, and assuming an annual prevalence of 300,000 NTDs, worldwide folic acid fortification could lead to the prevention of 150,000-210,000 NTDs per year.

In the area of autism and developmental disabilities prevention the Division has focused on the opportunity to improve the outcomes of children with autism through an extensive health communication campaign to identify children with developmental delays as early as possible. The "Learn the Signs. Act Early." (LTSAE) an education program for providers, parents and early educators, aims to improve awareness of early, healthy child development milestones. The intent of this program is to help parents, caregivers and others interacting with a child to quickly identify physical or developmental delays, especially focused on cognitive, emotional and social delays, link the child and family to early intervention services, thereby achieving the best possible outcomes.

After three years of program implementation, analyses of data show that pediatricians who were aware of LTSAE indicated that they were significantly more confident discussing cognitive development with parents, that they were more likely to be aware of resources available for referral and treatment, and that they were likely to have resources to educate parents. Preliminary results of a HealthStyles survey show that parental awareness of the campaign correlated with greater knowledge about childrens' developmental milestones,

increased confidence in talking with teachers and doctors about child development issues, and an increased ability to find appropriate services for children with delays.

To maximize the potential public health impact of future work, DBDDD has prioritized work on diabetes, obesity, and medications used during pregnancy. These risk factors were selected based on their prevalence and the epidemiological evidence showing the strength of their association with major birth defects. (See Appendix IV for a brief overview of major birth defects.) Successful efforts to modify these preventable exposures should prevent major birth defects and the lifelong consequences that accompany these serious disorders.

Yet another fundamental component of the Center's work involves newborn screening. An effective screening program is an important precursor to intervention and treatment programs. Newborn screening identifies conditions that can affect a child's long-term health or survival. Early detection, diagnosis, and intervention can prevent death or disability and enable children to reach their full potential. Each year, millions of babies in the U.S. are routinely screened, using a few drops of blood from the newborn's heel, for certain genetic, endocrine, and metabolic disorders. DBDDD works closely with other agencies and CDC programs to implement, evaluate, and enhance screening programs to optimize outcomes for affected children identified through newborn screening.

DBDDD's Strategic Plan

Priority 1: Enhance surveillance and research for autism and other developmental disabilities to monitor changes in prevalence and contributing factors, and better inform policy and program activities.

Characterize the Problem: Prevalence, Incidence and Distribution

Four Year Outcome: Data on the prevalence of Autism Spectrum Disorders (ASDs) among 8-year-old children will be reported biennially by the ADDM Network to monitor the changing prevalence of these serious conditions. These data will be used for national/state/local policy and planning to better meet the needs of children with ASDs and their families.

Progress Indicators:

1. Publish updated ASD prevalence estimates among 8-year-old children every two years, within 6 months of final submission of data to CDC, and assess factors potentially contributing to the rising prevalence.
2. Complete the first "Early ADDM" study to estimate the prevalence of ASDs among 4-year-old children; initiate prevalence study to follow up this initial cohort at age 8. (IACC Objective 7L)
3. Evaluate the presence of racial and ethnic disparities in the proportion of children with ASDs having a first evaluation by 36 months of age.
4. Implement a data dissemination and communication plan to increase the utility of ADDM data and improve policy and practice.

Four Year Outcome: We will have a better understanding of how changes in diagnosis and other system issues influence trends in ASD prevalence and ensure that knowledge is disseminated to policy makers and program leaders at the national, state, and local levels.

Progress Indicators:

1. Evaluate the impact of changes in awareness and diagnostic practices on ASD prevalence trends through data and modeling approaches. (IACC Objective 7I)
2. Complete analyses to identify factors that may explain: a) why certain groups of children are more likely to be identified by community providers as having ASD; b) how often our surveillance case definition matches the providers' impressions; c) whether these factors are changing over time; d) the impact of these factors on ASD prevalence trends; and e) what recommendations should be made to address the factors identified (e.g., increased ASD screening for children in certain groups, promoting the use of robust diagnostic instruments.) (IACC Objective 7I)

Conduct Research into Risk and Protective Factors

Four Year Outcome: Based on data from the Study to Explore Early Development (SEED) Phase I, a population-based study of epidemiologic risk factors for ASD, we will have a better understanding of the strength of the association of ASD with exogenous hormones, such as infertility treatments, and maternal infections with ASD that will inform potential action for prevention or intervention.

Progress indicators:

1. Complete enrollment and data collection for 6 SEED, Phase I sites.
2. Initiate a second round of SEED data collection (SEED Phase II) to increase the total SEED pooled sample size for investigation of SEED's high priority risk factors, thereby enhancing the potential for identifying important ASD risk factors that will inform action for prevention or intervention. (IACC Objective 3F)
3. With SEED Phase I data, investigate the role of exogenous hormones, such as infertility treatments, and maternal infections as high priority risk factors for ASD. (IACC Objective 3F)

Develop, Implement and Translate Feasible and Effective Interventions

Four Year Outcome: Learn the Signs. Act Early. campaign materials are relevant, effective and are used by public, professional and private organizations that serve parents and caregivers of young children.

Progress Indicators:

1. Reassess the appropriateness and adequacy of the current LTSAE materials to meet the current information needs of parents of young children and other caregivers.
2. Refine campaign materials and implementation strategies to improve their reach and effectiveness in meeting the needs of the public, professional and private organizations who serve parents and caregivers of young children.

3. Work with the Health Resources and Services Administration (HRSA) to explore campaign implementation strategies and the effectiveness of the campaign in reaching and educating specific population subgroups.
4. Broadly disseminate LTSAE materials through organizations, various systems and professional and lay efforts serving young children and their families (e.g., Nurse Family Partnerships, WIC, etc.)

Four Year Outcome: Effective strategies exist and are used in professional practice to reduce racial/ethnic disparities in the identification of ASD and other developmental disabilities.

Progress Indicators:

1. Explore strategies to reduce racial/ethnic disparities in the identification of ASD and other developmental disabilities by enhancing the Learn the Signs. Act Early. health communications campaign efforts among special populations such as low-income groups.
2. Continue collaborative work with HRSA to carry out two projects addressing early identification among lower socioeconomic status groups and Hispanic populations.
3. Identify and evaluate strategies to reduce racial/ethnic disparities in the identification of ASD and other developmental disabilities.
4. Develop partnerships to disseminate proven strategies for improving provider and parental knowledge of cognitive development milestones to targeted audiences.

Priority 2: Prevent congenital heart defects and other major birth defects associated with obesity, diabetes and medications.

Characterize the Problem: Prevalence and Distribution

Four Year Outcome: Improved birth prevalence and survival estimates for individual birth defects by specific subpopulations are used for national/state/local policy and planning to better meet the needs of families affected by major birth defects.

Progress indicators:

1. Develop standards for data acquisition and clinical review in birth defects surveillance to improve program assessment and evaluation and guide ongoing program development.
2. Develop strategies to maximize the potential of electronic medical records to improve the accuracy and efficiency of birth defects surveillance.
3. Pool surveillance data from multiple state-based surveillance programs to evaluate the survival of infants with specific birth defects in the United States and to identify social determinants of health measured at the community level that influence survival.
4. Make data more readily available to programs and policy makers by launching an interactive, web-based version of the annual birth defects surveillance data reported to the National Birth Defects Prevention Network.

Four Year Outcome: Understanding the contribution of birth defects to longer term outcomes (i.e., beyond infancy) and the economic impact of specific birth defects will inform medical and public health practices, provide key updated information on prognosis to parents and health care providers, and improve health and support services. Linking data from birth defects surveillance programs to data sets of other health outcomes and administrative data will allow investigators to more fully characterize the public health impact of birth defects and allow for the examination of research questions of greater policy and clinical relevance.

Progress indicators:

1. Link data from birth defects surveillance systems with pediatric cancer registries, special education data, educational achievement data, and other available outcome data to identify opportunities to improve the longer term outcomes of children with birth defects.
2. Link data from birth defects surveillance systems with health care cost data (e.g., Medicaid claims) to examine the economic burden associated with birth defects.

Conduct Research into Modifiable Risk and Protective Factors for Major Birth Defects

Four Year Outcome: Safety and risk information on medications frequently used by reproductive-aged women and used during pregnancy is disseminated and used to assist women and their health care providers in making informed treatment decisions.

Progress indicators:

1. Identify medications that are most frequently used immediately before and during pregnancy to assess their safety or risk to the fetus and allow an appropriate evaluation of risk versus benefit. In collaboration with AHRQ, FDA, and NIH, develop a framework for an ongoing, systematic, evidence-based review to evaluate the safety or risk of specific medications during pregnancy.
2. Assess the percentage of all medications (prescription and over-the-counter) with sufficient data to determine the safety or risk when used during pregnancy.
3. Follow up on key findings from the National Birth Defects Prevention Study showing associations between specific medications (e.g., specific SSRIs, antibiotics) and major birth defects. Additional data will inform guidance to clinicians on the safest choices for managing maternal illness during pregnancy and will prevent the occurrence of major birth defects.

Four Year Outcome: Collaborate with NCCDPHP to evaluate potential effect modifiers for diabetes and obesity to develop appropriate clinical or policy-based interventions for women at high risk for diabetes and obesity.

Progress Indicators:

1. Evaluate diet quality and other nutritional factors as potential effect modifiers of the association between diabetes/obesity and major birth defects.
2. Assess physical activity and type of obesity as potential independent risk factors and as potential effect modifiers of the risk of major birth defects.
3. Identify and evaluate potential policy changes that could be implemented related to diabetes screening strategies, nutrition, and physical activity among women with pre-existing and gestational diabetes.

Develop, Implement and Translate Feasible and Effective Interventions

Four Year Outcome: Programs to reduce birth defects and other adverse pregnancy outcomes are evaluated, disseminated, and implemented.

Progress Indicators:

1. In coordination with the Division of Diabetes Translation (DDT), the Division of Nutrition, Physical Activity, and Obesity (DNPAO), and the Division of Reproductive Health (DRH), and with input from other experts and stakeholders, develop a public health framework for diabetes and pregnancy that will: a) identify and prioritize data collection; and b) recommend prevention and health promotion strategies to reduce adverse pregnancy outcomes associated with pregestational diabetes.
2. Develop and implement effective policy approaches and other interventions to improve reproductive outcomes among women at risk of diabetes and obesity.

Priority 3: Prevent alcohol-exposed pregnancy among reproductive age women to prevent fetal alcohol syndrome and other adverse effects of prenatal exposure.

Develop, Implement and Translate Feasible and Effective Interventions

Four Year Outcome: The percentage of primary health care providers who always screen for alcohol use and conduct brief interventions with women of reproductive age who screen positive for risky drinking will increase by 10%.

Progress Indicators:

1. Promote alcohol screening and brief intervention in primary care settings serving women of reproductive age.
2. Determine the potential impact of "bundling" a number of prevention strategies, e.g., – brief alcohol counseling with smoking cessation strategies.
3. Develop, disseminate and implement recommendations based on research conducted regarding the effectiveness of alcohol screening and brief intervention in specific settings and when bundled with other prevention programs.
4. Work with alcohol programs in NCIPC and NCCDPHP to identify primary care collaborators to support, monitor and evaluate implementation of alcohol screening and intervention guidelines for women of reproductive age.

Four Year Outcome: Increased awareness of the dangers of alcohol use during pregnancy among women and their health care providers.

Progress Indicators:

1. Assess knowledge of alcohol risk during pregnancy among women of reproductive age.
2. Initiate activities to assess knowledge of alcohol risk during pregnancy among health care providers.

3. Clarify messaging around alcohol use and pregnancy for women of reproductive age and their health care providers.
4. Target awareness and educational activities to women of reproductive age and their healthcare providers.

Four Year Outcome: Increase use of Project CHOICES among a wide range of service providers serving women at risk of an alcohol exposed pregnancy.

Progress Indicators:

1. Implement activities to assess the feasibility and impact of Project CHOICES in settings serving women at risk of alcohol-exposed pregnancy.
2. Establish the training and technical assistance infrastructure to support Project CHOICES.
3. Modify and evaluate Project CHOICES based on feasibility studies to meet the needs of service providers of women at risk of an alcohol-exposed pregnancy.
4. Disseminate Project CHOICES and provide technical assistance to a wide range of service providers serving women at risk of an alcohol-exposed pregnancy.

Four Year Outcome: National data on populations at risk for an alcohol-exposed pregnancy are available and disseminated to states and national organizations serving women of reproductive age to inform FASD prevention messages and influence clinical and public health policy on alcohol screening and brief intervention.

Progress Indicators:

1. Work with National Center for Health Statistics (NCHS)/National Survey of Family Growth (NSFG) to better define the at-risk population for alcohol-exposed pregnancy.
2. Incorporate Behavioral Risk Factor Surveillance System (BRFSS) alcohol questions into NSFG.

Priority 4: Eliminate folic acid-preventable neural tube defects in the United States and globally.

Characterize the Problem: Prevalence, Incidence and Distribution

Four Year Outcome: Optimal blood folate concentration is used as a biomarker to evaluate the impact of fortification efforts.

Progress Indicators:

1. Work with the World Health Organization and others to determine optimal blood folate concentrations for NTD prevention, by examining available scientific data and convening an expert panel.
2. Disseminate recommendation on optimal blood folate concentrations and develop guidance on appropriate folic acid fortification strategies for low- to middle-income countries. (Note: high income countries have effective fortification strategies in place. Many of these strategies focus on grain fortification, but in most cases this excludes corn masa fortification.)

Domestic

Four Year Outcome: Prevalence of NTDs among Hispanic women is not more than 10% higher than the prevalence in non-Hispanic whites.

Progress Indicators:

1. Work with FDA, GRUMA (a major manufacturer of corn masa flour products), American Academy of Pediatrics, Spina Bifida Association, March of Dimes and other partners to develop a food additive petition to allow folic acid fortification of corn masa flour.
2. Conduct formative research to gauge the level of consumer acceptance of fortified corn masa flour and related products.
3. Contribute safety data to be used in the development of a food additive petition to allow fortification of corn masa flour.

Global

Four Year Outcome: Number of countries fortifying highly consumed staples with folic acid increases by 20% (from 53 to 64).

Progress Indicators:

1. NCBDDD supports the work of the International Micronutrient Malnutrition Prevention and Control Program (IMMPCt) and others in the prevention of NTDs globally (e.g., supporting existing program efforts, initiating new program efforts, providing scientific and technical support for monitoring and surveillance).
2. CDC provides technical assistance to inform policy recommendations and NTD surveillance activities in countries engaged in folic acid fortification efforts.
3. Broad consensus on optimal blood folate level for NTD prevention is established and used to develop guidance and monitor the impact of fortification efforts globally.
4. CDC supports development of birth defects surveillance for NTDs in at least two low- to middle-income countries to evaluate prevention efforts.

Priority 5: Enhance the quality and usefulness of newborn screening data and programs.

Four Year Outcome: New functional screening programs are implemented as recommended by the Health and Human Services Secretary, and existing screening programs are enhanced and evaluated, to assess and optimize outcomes for affected children identified by newborn screening.

Progress Indicators:

1. Co-sponsor a critical cyanotic congenital heart disease (CCCHD) workgroup meeting and assist with dissemination of the outcomes of that meeting to develop protocols for CCCHD screening.
2. Develop and implement a strategy to evaluate CCCHD screening programs.
3. Publish methodologies and outcome data on long-term follow-up of children with inborn errors of metabolism.

4. Develop a newborn screening follow-up quality improvement educational module for primary care providers.
5. Finalize a national contingency plan for newborn screening, assist state programs with developing standard operating procedures to carry out the plan in the event of emergencies, and maintain and update the plan as needed.

Division of Blood Disorders

Our Vision, Approach and Priorities

Millions of Americans have inherited or acquired conditions of the blood that result in adverse health outcomes. Initially focused on coagulation disorders and conducting research to determine the hematological mechanisms for clotting, the Division's activities have expanded over the years to strengthen our hemophilia and blood safety programs and establish new programs focused on preventing and reducing complications and death caused by nonmalignant bleeding, clotting and other blood disorders such as Sickle Cell Disease, thalassemia, and Diamond Blackfan Anemia.

Data collection and analysis has always been a central feature of DBD's work, though from a public health standpoint, these activities have been carried out from a laboratory or clinical perspective. In recent years DBD's focus has shifted to a population-based public health model that addresses the critical public health challenges associated with bleeding, clotting and other blood disorders and related secondary conditions. This comprehensive public health approach includes, in addition to data collection and monitoring, research to identify preventable or modifiable risk factors, developing and testing prevention strategies and assuring widespread adoption of effective prevention strategies.

Working with our many public health partners – other federal and state agencies, academia, and professional and community-based organizations – DBD strives to improve the lives of people at risk or affected by blood disorders.

Our Goals, Priorities and Outcomes for the Future

The goals of DBD are to:

- Establish blood disorders as a public health priority
- Understand the causes of and risk factors for blood disorders
- Understand and minimize occurrence and complications of blood disorders
- Develop and evaluate evidence-based interventions for blood disorders
- Ensure that people with or at risk for blood disorders have access to credible health information

The Division has prioritized its work with an immediate focus on nonmalignant blood disorders with the greatest burden and unmet need.

Priority 1: Prevent clotting disorders – deep vein thrombosis and pulmonary embolism.

Priority 2: Prevent and control complications resulting from hemoglobinopathies, with a focus on Sickle Cell Disease and thalassemia.

Priority 3: Prevent and control complications resulting from bleeding disorders such as hemophilia and von Willebrand Disease.

In meeting our goals and priorities, we intend to contribute to achieving the following outcomes:

- Reduce the incidence of deep vein thrombosis/pulmonary embolism, and prevent related mortality and serious morbidity.
- Improve the life expectancy of people with Sickle Cell Disease.
- Reduce the morbidity and mortality related to bleeding disorders in women, particularly during pregnancy.
- Prevent emerging morbidities of people with bleeding disorders.

How We Developed Our Priorities

Non-malignant blood disorders such as deep vein thrombosis/pulmonary embolism (DVT/PE), Sickle Cell Disease (SCD), thalassemia, hemophilia and von Willebrand disease (VWD) affect millions of people in the United States. They cut across the boundaries of age, race, sex and socioeconomic status and result in significant healthcare costs, complications, co-morbidities and sometimes death.

Deep Vein Thrombosis/Pulmonary Embolism (DVT/PE)

DVT/PE are under-diagnosed, serious, potentially preventable medical conditions associated with an estimated 100,000 deaths each year. The precise number of people affected by DVT/PE is unknown, however estimates range from 350,000-600,000 annually with associated healthcare costs up to $10 billion. Sudden death is the first symptom in about a quarter of the people who have a PE. DVT/PE also poses important problems for women; there is a five times greater risk for developing DVT/PE during pregnancy, the post partum period, and while taking hormonal contraceptives. Other major risk factors include advance age, immobility, surgery, and obesity. While several genetic risk factors have also been identified, an estimated one-third to one-half of DVT/PE cases have unknown risk factors.

Because DVT/PE occurs in diverse healthcare settings as well as in the community, it is diagnosed and managed by myriad providers. While evidence-based guidelines on DVT/PE prophylaxis and management for hospitalized patients exist, about two-thirds of cases occur after patient discharge. Therefore, a need exists to develop evidence-based standards for outpatient DVT/PE prophylaxis and management. In addition, there is a need to increase the public's awareness of the risk factors and symptoms of DVT/PE, and provider knowledge of, and adherence to, guidelines for the management and treatment of these conditions.

Hemoglobinopathies – Sickle Cell Disease (SCD) and thalassemia

Although hemoglobinopathies can affect people of all ethnicities, they are most common among ethnic minorities and represent a major health disparity. SCD primarily affects people of African descent and is becoming increasingly prevalent among Hispanics. Thalassemia is most common among people whose ancestors come from the Mediterranean region, the Middle East, India, Southeast Asia, Southern China and Africa. In the U.S, there are approximately 100,000 people with SCD and an estimated 3 million with Sickle Cell trait. Up to $1.1B in total annual healthcare costs are associated with SCD. Thalassemia is thought to affect approximately 1,000 persons, but the number of carriers of the trait is unknown as is the amount of healthcare expenditure. While many advances have been made, persons with SCD have a three decades shorter life expectancy than other Americans and can experience repeated episodes of severe pain, organ

damage, serious infections or anemia. People with the severe anemia form of thalassemia – Cooley's Anemia- usually do not live into adulthood without treatment often requiring monthly blood transfusions and medication to remove iron overall.

There is a lack of understanding of the overall population impact of hemoglobinopathies. Data is needed to identify risk factors, barriers to care, and effective inventions and prevention measures for those affected by these disorders. In addition, public and healthcare provider awareness and education is needed on how to treat and manage these rare disorders.

Bleeding Disorders

There are an estimated 20,000 people (mostly males) with hemophilia in the United States with healthcare costs approaching $3 billion a year. People with hemophilia (PWH) have spontaneous internal bleeding and prolonged bleeding after an injury or surgery that can lead to long-term damage and chronic conditions such as a disabling joint disease. Approximately 15-20% of hemophilia patients develop an inhibitor where the individuals' immune system destroys the clotting factor replacement products, making the treatments ineffective, increasing hospitalization rates and co-morbidities. A single patient with complications and an inhibitor can have over $1M in treatment costs annually. Research on how and why a PWH may develop an inhibitor is needed to identify effective prevention/intervention strategies.

VWD is a bleeding disorder that affects 1% of all women in the U.S. (approximately 1 in 100) and is associated with an increased risk for miscarriage, bleeding during pregnancy, and postpartum hemorrhage. Women with VWD have a maternal mortality rate 10 times higher than that of women without VWD. Many women learn to live with the problems their bleeding causes, such as heavy menstrual periods, and do not realize that they may have a blood disorder. VWD is often not diagnosed and can result in costly and inappropriate treatment such as unnecessary hysterectomies. With early identification, women with bleeding disorders can avoid many of the associated complications and improve their quality of life. Increasing the awareness of women about the symptoms of a bleeding disorder, and education and guidelines for healthcare providers in early recognition and appropriate referral for treatment is needed.

DBD's Strategic Plan

Priority 1: Prevent clotting disorders – deep vein thrombosis and pulmonary embolism.

Characterize the problem: Prevalence, Incidence and Distribution

Four Year Outcome: Surveillance system tracks the occurrence and recurrence of DVT/PE and effectiveness of prevention efforts.

Progress Indicators:

1. Work with CDC colleagues (DHQP, DHDSP, NCHS) and external partners (such as AHRQ, ASH, CMS, NHLBI) to assess the strengths and limitations of the use of existing datasets as sources of information for DVT/PE surveillance.
2. Support investigator-initiated population-based surveillance pilot projects to allow for the evaluation of the effectiveness of multiple approaches for DVT/PE surveillance, and improve our understanding of opportunities for, and impact of, prevention efforts over the long term.

3. Develop and implement a viable surveillance system for DVT/PE based on our evaluation of existing datasets and the surveillance pilot projects.

Develop, Implement and Translate Feasible and Effective Interventions

Four Year Outcome: An increase in knowledge and awareness of the general public and healthcare providers about the prevention of DVT/PE.

Progress Indicators:

1. Assess the awareness of risk factors, signs, symptoms, and prevention of DVT/PE, among the general public and healthcare providers and identify knowledge gaps.
2. Collaborate with professional and community-based organizations to develop and implement a public awareness campaign to educate the public and health care providers on the risk factors, signs, and symptoms associated with DVT/PE.
3. Collaborate with professional organizations to develop health care provider education and training programs that promote evidence-based practice for the prevention, diagnosis and treatment of DVT/PE.

Four Year Outcome: An increase in the use of evidence-based prevention and management guidelines for DVT/PE in the hospital and outpatient settings.

Progress Indicators:

1. Work with appropriate professional organizations (such as AAP, ACOG, AMA, ASH and the Joint Commission) and federal partners to assess existing private sector, national and international guidelines for the prevention and management of DVT/PE.
2. Assess healthcare provider knowledge and use of DVT/PE guidelines.
3. Develop and disseminate a uniform set of evidence-based prevention and management guidelines for DVT/PE in the hospital and outpatient settings, to promote interventions for high-risk inpatients and risk reduction for the general population.
4. Collaborate with appropriate professional organizations (such as AAP, ACOG, AMA, ASH and the Joint Commission) and federal partners to implement guidelines.
5. Evaluate barriers to implementation of, and compliance with, DVT/PE prevention and management guidelines.

Priority 2: Prevent and control complications resulting from hemoglobinopathies, with a focus on Sickle Cell Disease and thalassemia.

Characterize the problem: Prevalence, Incidence and Distribution

Four Year Outcome: Hemoglobinopathies surveillance system (developed as part of the Registry and Surveillance of Hemoglobinopathies–RuSH-project) tracks key health indicators for people with hemoglobinopathies in order to provide the research, public health, clinical and consumer communities data to inform research, services, and policy agenda.

Progress Indicators:

1. Establish pilot surveillance projects for SCD and thalassemia in six states (RuSH).
2. Fund 1-2 additional states to cover 50% of African Americans and 50% of the Asian Population in the U.S.
3. Develop case definition, common data sets, and indicators for longitudinal monitoring.
4. Conduct validation studies to evaluate the utility of administrative and other existing data sources for hemoglobinopathy surveillance and epidemiologic research.
5. Publish surveillance reports and conduct analysis projects using year-one data.
6. Generate baseline indicators to measure 4-6 Healthy People 2020 objectives that have been determined measurable using RuSH surveillance.
7. Characterize the population of people with SCD and thalassemia, (e.g., accurate prevalence estimates, healthcare utilization, treatment adherence, co-morbidities).

Conduct Research into Risk and Protective Factors

Four Year Outcome: Identification of factors (genetic, environmental, behavioral, and health services) that lead to improved health outcomes of people with SCD and thalassemia to inform public health, clinical and consumer communities and policymakers.

Progress Indicators:

1. Evaluate models of health services delivery for people with SCD and thalassemia to assess impact on health outcomes.
2. Identify and evaluate the impact of genetic, environmental, behavioral and other risk factors such as micronutrient deficiencies, co-morbidities and co-occurring conditions, and access to health services on health outcomes of persons with SCD and thalassemia.
3. Examine the economic burden of SCD and thalassemia to identify areas where there are opportunities to reduce economic burden while simultaneously improving health outcomes of persons with SCD and thalassemia.

Develop, Implement and Translate Feasible and Effective Interventions

Four Year Outcome: An increase in knowledge and awareness of the public and healthcare providers about treatment for, and management of, people with SCD and thalassemia.

Progress Indicators:

1. Collaborate with professional and community-based organizations to develop and implement a public awareness campaign to educate the public and healthcare providers on SCD and thalassemia.
2. Collaborate with professional organizations to develop health care provider education and training programs on the treatment and management of people with SCD and thalassemia.
3. Provide community-based organizations and state and local health departments' support, coordination, and technical assistance in conducting SCD education and training activities for parents, communities, and providers.

Priority 3: Prevent and control complications resulting from bleeding disorders such as hemophilia and von Willebrand Disease.

Characterize the Problem: Prevalence, Incidence and Distribution

Four Year Outcome: Improved representativeness and expanded utility of surveillance data on bleeding disorders.

Progress Indicators:

1. Evaluate the current HTC-based bleeding disorders surveillance system (Universal Data Collection program - UDC) in terms of current representativeness and use.
2. Engage relevant healthcare provider groups and policy makers to make recommendations for potential expanded uses and required data element modifications.
3. Provide recommendations for deleting/adding information to be collected, as the basis for improved research and prevention efforts.
4. Modify UDC data elements as appropriate for research and prevention efforts.

Conduct Research into Risk and Protective Factors

Four Year Outcome: Improved capacity for epidemiologic research studies of populations with inherited bleeding disorders such as hemophilia and VWD.

Progress Indicators:

1. Evaluate the quality and utility of existing datasets to describe the epidemiology of blood disorders (e.g. burden of disease, risk factors, co-morbidities, mortality, and disability).
2. Summarize and publish existing datasets as resources to study the epidemiology of blood disorders.
3. Address information gaps by collecting information not previously available
4. Disseminate and implement recommendations to enhance existing datasets.
5. Conduct research to be used as the basis for developing intervention strategies.

Develop, Implement and Translate Feasible and Effective Interventions

Four Year Outcome: Increased use of intervention strategies by hemophilia treatment providers, to reduce the development of inhibitors (antibodies to products used to treat bleeding episodes) in persons with hemophilia.

Progress Indicators:

1. Evaluate the cost effectiveness of genotyping hemophilia patients to identify those at high risk for inhibitor development and, as appropriate, the need for alternative treatment strategies.
2. Establish a baseline rate of patients receiving routine inhibitor screening.
3. Assess inhibitor screening practices of hemophilia treatment providers.
4. Develop and disseminate national guidelines for inhibitor screening.

Four Year Outcome: An increase in the knowledge and awareness of women about the symptoms (menorrhagia, nosebleeds, postpartum hemorrhage) of a bleeding disorder such as VWD.

Progress Indicators:

1. Conduct a survey to assess women's (ages 18-25) overall awareness and knowledge of bleeding disorders and communication channels they would use to learn more about bleeding disorders.
2. Collaborate with professional organizations and community-based organizations to develop and implement an education campaign for women about the signs and symptoms of bleeding disorders.

Four Year Outcome: Increased proportion of OB/GYNs engaged in early identification and appropriate referral of women with bleeding disorders.

Progress Indicators:

1. Conduct a survey of OB/GYNs' awareness, knowledge and practices relating to the identification of women's bleeding disorders.
2. Develop and disseminate recommendations to OB/GYNs.
3. Collaborate with professional organizations, such as ACOG and ASH to implement identification and referral guidelines.

Four Year Outcome: Increased use of effective transition plans by young people with blood disorders to obtain the care needed to maintain health and prevent secondary complications in adulthood.

Progress Indicators:

1. Identify and evaluate existing materials and communication modalities for transition planning as it pertains to young people with blood disorders.
2. Collaborate with professional and consumer organizations to address information gaps and develop curriculum on self-management skills needed by young people with blood disorders for successful transition into adulthood.

Collaborate with professional and consumer organizations to disseminate, promote and evaluate the adoption of transition planning for young people with blood disorders.

Division of Human Development and Disability

Our Vision, Approach and Priorities

The Division of Human Development and Disability was created to prevent secondary conditions and enhance the quality of life among persons who already have a disability. DHDD combines multiple funding lines to promote healthy development and reduce health disparities across the life course for persons with or at risk of disability. Our approach is evolving through three Institute of Medicine (IOM) reports (1991, 1997, 2007), two Surgeons General reports, development of 20 HP2020 objectives, and the Affordable Care Act (ACA) to include disability populations within general public health activities. The DHDD programs have contributed to the maturation that the field of disability and public health has experienced through surveillance, epidemiological research, and implementation of effective policies, programs and practices.

DHDD's diverse programs and range of activities are unified by its life course mission. Healthy development across the life course is a major focus of the division. Beginning prior to birth, a program of early parenting intervention has been demonstrated to optimize child development and prevent mild developmental delays and disorders to children born into poverty. Its roll-out for large-scale implementation through the Early Head Start network is underway. Working with HRSA, the Early Hearing Detection and Intervention (EHDI) program monitors identification and intervention for newborns with hearing loss. During childhood, the health and developmental needs of children with complex disabling conditions, such as spina bifida and muscular dystrophy, and their families are characterized to determine public health interventions needed to promote optimal outcomes. DHDD collaborates to articulate and implement a public health approach to addressing successful transition of youth with disabilities into adulthood. Finally, DHDD has developed a network of state Disability and Health programs that monitor and intervene to meet the public health needs of adolescents, adults and elders with disabilities.

Our Goals, Priorities and Outcomes for the Future

Building upon our history and public health knowledge, DHDD has identified the following goals for the future:

- Improved health and developmental outcomes for children.
- Improved quality of life and life expectancy of people with disabilities.
- Eliminated health disparities of children, youth and adults with disabilities.

Identified Public Health Needs for our Populations

- Newborns are at risk of developmental delays if hearing loss is not detected and interventions received. Effective newborn hearing screening can be assured by understanding the *prevalence* of hearing loss, audiological exams and referrals for intervention after screening newborns, *incidence* of screening, referrals and interventions, and *distribution* of each regionally and by state.

- Children living in poverty are at higher risk for developmental delays than children not living in poverty. One way to address this is to consider parenting interventions as public health practices to mitigate the influence of poverty on children's risk of developmental delays.

- Disparities are evident across multiple health indicators for persons with disabilities. For example, obesity rates in children/youth with disabilities exceed those without disabilities (22.5% versus 15.74%) and this disparity is even greater in adults (37.6% versus 23.7%). Persons with mobility and intellectual limitations are at highest risk of obesity. Reducing disparities in obesity rates requires expanding research to increase our understanding on how to best *develop*, *implement* and then *translate* knowledge gained into *feasible* and *effective* public health *interventions*. By focusing on obesity and other health indicators, we are aligning our work with a framework developed by IOM in 2008, and creating the potential to work on broader, related issues such as barriers to health promotion and social participation.

- Unmet health care needs of persons with disabilities are more than double that of the general population. Even with insurance, 16% of persons with disabilities report skipping needed health care in the previous year because of cost compared with 5.8% of the rest of the population. Adults with disabilities are more likely to rely on public insurance such as Medicaid or Medicare, and health care providers may be less likely to accept these forms of insurance. For those without insurance, these numbers reach 61% for persons with disabilities and 31% for others. Identifying *prevalence* of selected health care services, the *incidence* in which they occur, and how these services vary in their *distribution* between regions and states is required to address the problem. Model programs of conducting accessibility assessments and providing training to providers highlight potential public health interventions.

- Disability status is frequently overlooked as a demographic characteristic in surveying the health of populations. Of approximately 44 currently inventoried CDC surveillance systems, only 3 include disability status identifiers. Disability indicators are required by ACA for use in public health surveys to allow researchers and others to more accurately identify prevalence of disability and disability type, incidence of disability in our aging population, and distribution of disability and disability type regionally and between states.

Priorities for the Division of Human Development and Disability (ordered chronologically):

Priority 1: Reduce disparity in obesity and other health indicators in children, youth and adults with disabilities.

Priority 2: Improve developmental outcomes of all children.

Priority 3: Ensure that all newborns are screened and assessed for hearing loss and receive appropriate intervention according to established guidelines.

Priority 4: Identify and reduce disparities in health care for persons with disabilities.

Priority 5: Incorporate disability status as a demographic variable into all relevant CDC surveys, policies and practices.

By focusing on our goals and priorities, we intend to achieve the following outcomes:

- Change personal health behaviors and risk factors (e.g., obesity, smoking) to improve health in children, youth and adults with disabilities.
- Mediate the impact of poverty on developmental outcomes for young children.
- Increase early identification and intervention for infants and young children with disabling conditions.
- Improve health care access for children, youth and adults with disabilities.
- Improve public health surveillance systems to track the health, development and participation of persons with disabilities across the life course.

How We Developed Our Priorities

DHDD's reach is broad. Its networks of EHDI programs in 53 states and territories ensures infant hearing screening for virtually all children born in the United States. Our 16 Disability and Health state programs and 6 NACCHO community sites provide public health outreach and access to persons with disabilities in many areas around the country. Through implementation, evaluation and dissemination of evidence-based preventive health activities targeting people with disabilities, DHDD seeks to prevent the development of secondary conditions, address health disparities, and develop a stronger evidence base of effective preventive programming. Our desire is to expand this network as the programs demonstrate their merit in coordinating essential public health services such as emergency preparedness and health care access. DHDD also supports eight condition-specific public health practice and resource centers, such as the Christopher and Dana Reeve Foundation and Special Olympics International. These resource centers have large national and global constituencies. In our new DHDD Partnership initiative, they are working together on DHDD-specific goals such as obesity, as well as developing collaborations for shared public health concerns. DHDD also works closely with national and international cross-disability and academic organizations to extend its reach and translate its research into programs.

Surveillance and Research

A particular focus of DHDD and the CDC Disability and Health Work Group is to address inclusion of disability in surveillance, programs and policies. In order to ensure understanding of disability populations, disability status needs to be incorporated into surveillance tools in ways that it currently is not. The ACA, in its language in Section 4302, confirms the need to address this deficit. Through its surveillance and public health research, DHDD has illustrated that people with disabilities incur higher rates of obesity, diabetes, preventable infections, sexual violence and mental health conditions such as depression than do the general population. Further, people with disabilities are over-represented in populations experiencing obesity, smoking and other chronic conditions, requiring us to translate best practices in ways that combat these behaviors by working closely with our counterparts in chronic disease and injury prevention programs. In the next four years, DHDD is specifically addressing obesity in persons with disabilities as an exemplar of inclusion.

Developing and Implementing Effective Interventions

Working with the newly established CDC Disability and Health Work Group and with partners in states, DHDD is actively pursuing the inclusion of people with disabilities in public health programs and policies. For example, obesity prevention and intervention programs need to plan for purposeful inclusion of persons in wheelchairs, with visual limitations, and intellectual disabilities. In cases where inclusion of disability

populations are not effective, research is needed to provide policies, programs, and practices that are effective for our populations.

Effectively communicating health information to our different audiences is critical to our ability to translate research to action. For example, public awareness campaigns on health concerns, like the importance of mammograms for women with mobility limitations, have contributed to changing health behaviors among these women. Similarly, conferences that bring together the perspectives of persons with disabilities, researchers, and professionals are important venues to ensure that proposed strategies and approaches are appropriate, credible and feasible. Our website provides valuable information to multiple audiences and will soon be supplemented with information for each state on the health disparities of adults with disabilities.

Ongoing activities in surveillance and research, program implementation, and communication and dissemination are the tools employed by our Division to effect change. The overlap between these activities requires evaluating performance, investing in social capital and public health infrastructure, and translating knowledge gained into effective public health practices. In addition to contributing to change through expanded diffusion of disability awareness into mainstream public health activities, inclusion is also a product of change, inextricably linked to successful outcomes DHDD seeks to effect change in areas such as public policy, employment, health care finance, technology, research, communication and education, and changes to the built environment. Examples of outcomes supported by DHDD in which inclusion plays a key role:

- Public policy: providing evidence contributing to legislation that can improve the health and well-being of people with disabilities, such as the Americans with Disabilities Act (ADA);

- Employment: providing knowledge of the impact of employment and disability on health, contributing to creative programs, such as the Ticket to Work and Work Incentives Act (TWWIA), that can improve health and maintain employment for people with disabilities;

- Health care finance: providing cost estimates for disability populations, such as persons with spina bifida or those with mobility limitations, to help plan for inclusion of disability populations into accountable care organizations (ACOs), a new health care delivery system in the ACA;

- Technology: providing evidence that supports the expanded use of electronic medical records and meaningful use in ways that assure improved outcomes, for example, among newborns and children screened for hearing loss;

- Research: inclusion of disability into developing specialized disability-specific measurement approaches such as disability adjusted quality of life indicators, use of waist circumferences as more accurate proxies for obesity, or positive parenting skills that contribute to improved developmental milestones in children as means to assure that outcomes can be more accurately measured;

- Communication and education: inclusion of approaches that incorporate Section 508 (a 1998 amendment to the U.S. Rehabilitation Act of 1973), which assures that electronic and information technologies be accessible to people with disabilities; and

- Changes to the built environment: providing evidence of successful implementation of elements of universal design that allow access to health care for people with disabilities, reflected in Title III, Section 4203 of the ACA.

DHDD's Strategic Plan

Priority 1: Reduce disparity in obesity and other health indicators in children, youth and adults with disabilities.

Characterize the problem: Prevalence, Incidence and Distribution

Four Year Outcome: Determine surveillance systems and monitor progress on specific health indicators of children, youth and adults with disabilities.

Progress Indicators:

1. Publish surveillance data that identifies prevalence of people with disabilities who are at greatest risk of obesity.
2. Monitor status of HP2020 Disability and Health objectives and promote achievement of select objectives.
3. Prioritize health indicators for intervention.

Develop, Implement and Translate Feasible and Effective Interventions

Four Year Outcome: Reduce by 10% the disparity in obesity between children, youth and adults with mobility and intellectual limitations relative to persons without disabilities.

Progress Indicators:

1. Summarize and publish research on effectiveness of current generic obesity prevention/ intervention programs (including communication campaigns) to determine how program access and delivery may need to be modified for disability inclusion.
2. Develop guidance for practitioners that focuses on methods for including people with disabilities in obesity prevention and intervention programs.
3. In collaboration with the Division of Nutrition, Physical Activity and Obesity, Division of Adolescent and School Health and others, increase targeting of current population-based obesity prevention efforts to accommodate persons with disabilities.
4. Expand capacity to implement state-based interventions related to health indicators of persons with disabilities.
5. Engage in strategic collaboration with NCBDDD Public Health Resource and Information Centers to enhance educational materials for consumers and their families, and for health and public health personnel.

6. Support demonstration projects and translation initiatives of effective interventions targeted at highest risk groups and those most likely to succeed.
7. Implement effective targeted evidence-based interventions for obesity prevention/ intervention for children and adults with disabilities.
8. Increase the number and quality of obesity prevention/intervention programs that are appropriate for people with disabilities.

Priority 2: Improve developmental outcomes of all children.

Characterize the Problem: Prevalence, Incidence and Distribution

Four Year Outcome: Develop, expand, and disseminate population-based estimates of prevalence, developmental outcomes and factors associated with developmental outcomes among children at developmental risk (e.g., children with known conditions and those living in poverty.)

Progress Indicators:

1. Publish updated prevalence estimates, patterns of service utilization, and developmental outcomes among children at developmental risk.
2. Conduct follow-up studies with children who have key conditions and identify factors associated with their developmental outcomes.
3. Evaluate the utility of disability indicators in the National Survey of Children's Health and the National Survey of Children with Special Health Care Needs relative to prevalence of disability, service utilization, and health and developmental outcomes.

Four Year Outcome: The health needs, service utilization, environments, and outcomes for children and youth with complex disabling conditions are characterized to facilitate transition and optimize development.

Progress Indicators:

1. The DHDD life course conceptual model (based on the International Classification of Functioning) is recognized and applied in research and intervention for children and youth with complex disabling conditions to better facilitate transition from childhood to adulthood.
2. Retrospective studies identify service utilization, outcomes and information gaps for persons with select conditions (e.g., muscular dystrophy).
3. Collect and analyze standardized, longitudinal data on needs, service utilization, environmental factors and health and developmental outcomes for persons with select conditions (e.g., spina bifida, Fragile X).
4. Release care guidelines for select complex disabling conditions and document that knowledge of care providers and service delivery has improved.
5. NCBDDD Public Health Resource and Information Centers demonstrate the impact of educational materials for consumers and their families, and for health and public health personnel.

Develop, Implement and Translate Feasible and Effective Interventions

Four Year Outcome: With the Administration for Children and Families (ACF) and other organizations, implement the Legacy for Children (LFC) parenting program in multiple sites to decrease the prevalence of intervention children falling below average on measures of intellectual functioning or demonstrating behavioral problems.

Progress Indicators:

1. Collaborate with ACF on a feasibility study to develop, plan and adapt LFC for community implementation in selected ACF Early Head Start sites.
2. Evaluate the feasibility of implementing LFC in selected ACF Early Head Start sites, evaluating the community context, participant-program match, program perceptions, participation patterns, goals attainment, and lessons learned (consistent with Early Head Start program demonstration evaluations).
3. Leverage the Early Head Start system to ensure early identification and continuity of care for children enrolled in the intervention with suspected developmental delays.
4. Adapt LFC curriculum by incorporating findings from the ACF Early Head Start feasibility study.
5. Design a demonstration study to evaluate the effectiveness of the LFC program within Early Head Start-Legacy sites.
6. Fund demonstration sites to assess the impacts of LFC on enrolled children relative to comparison children at 2 and 3 years of age.

Priority 3: Ensure that all newborns are screened and assessed for hearing loss and receive appropriate intervention according to established guidelines.

Characterize the problem: Prevalence, Incidence and Distribution

Four Year Outcome: Improve the quality and use of data to assure the following newborn hearing program objectives have been achieved in the reporting jurisdictions: a) screening rate of 95% or more; b) audiology testing of 70% of infants failing hearing screening; and c) referral to early intervention services of 90% of infants with hearing loss.

Progress Indicators:

1. Enhance state-based electronic case management for follow-up of infants.
2. Expand capacity to validate and analyze data related to hearing screenings and audiology tests.
3. Increase efforts to collect referral data to early intervention and other services.
4. Adopt health information technologies common to multiple child health information systems to assist in follow-up activities and collection of data.

Four Year Outcome: Validated metrics are established to measure appropriateness for referral and intervention success for infants with hearing loss.

Progress Indicators:

1. Support a feasibility study in a limited number of states to develop data management systems capable of collecting and reporting developmental outcomes of infants/children identified through the hearing screening programs.
2. Identify predictors of appropriateness for referral to intervention based on success of intervention.

Priority 4: Identify and reduce disparities in health care for persons with disabilities.

Characterize the problem: Prevalence, Incidence and Distribution

Four Year Outcome: Unmet health care need of persons with disabilities is monitored and reported regularly to advance health care access.

1. Develop and publish regular reports on unmet health care needs generally and on targeted topic reports through Vital Signs, MMWR or other avenues.
2. In collaboration with partners and others, disseminate information on health care access.

Four Year Outcome: Develop a national plan for sustainable health surveillance of adults with intellectual disabilities.

Progress Indicators:

1. Determine a feasible definition of intellectual disability for surveillance purposes.
2. Collaborate with international initiatives to refine definitions and approaches to assess health status and health needs of persons with intellectual disabilities.
3. In collaboration with other federal agencies and national organizations, identify available data and data needs on populations of adults with intellectual disabilities.
4. Support and evaluate pilot studies on catchment-area surveillance of health status and health needs of persons with intellectual disabilities.

Develop, Implement and Translate Feasible and Effective Interventions

Four Year Outcome: States use disability disparity data to prioritize their public health policies and programs and to improve health care access for people with disabilities who are at greatest risk of not receiving quality health care.

Progress Indicators:

1. Analyze state-level BRFSS data to compare people with disabilities to those without disabilities for approximately 65 health care and health behavior indicators.
2. Create an interactive, web-based platform to display results; provide technical assistance to states using the reporting system; work with state partners, researchers, and others to develop and identify user-based needs and preferences.
3. Enhance BRFSS data with NHIS and NHANES data to confirm health disparities by disability status and identify disparities by type of disability.

4. Enhance use of the data reporting system through follow-up and technical assistance.
5. Collaborate with Division of Adolescent and School Health (CDC), Maternal and Child Health, Department of Education, and others to coordinate state priorities and planning on care for children, youth, and adults with disabilities and special health care needs.
6. Document impact of data availability on state and national programs and policies in awareness of health disparities of people with disabilities.

Four Year Outcome: Reduced proportion of people with disabilities who report delays in receiving primary and periodic preventive care due to specific barriers (HP 2020 Developmental Objective 4).

Progress Indicators:

1. Promote inclusion of people with disabilities in CDC-funded programs and policies, particularly those focused on primary and preventive care.
2. Maintain and expand efforts by State D&H programs to conduct health promotion activities to include: a) public awareness campaigns on obesity, physical activity, and smoking, preventive services (e.g., *The Right to Know Campaign*); b) state and local training on health promotion (including accessibility) for providers and people with disabilities; and c) accessibility assessments of health care facilities.
3. Assure that State D&H programs apply an inclusion approach to targeted public health campaigns /interventions (e.g., accessible medical equipment, diabetes, obesity, emergency preparedness, etc.).
4. Expand capacity to implement state-based interventions related to health care access for persons with disabilities.
5. Assure adequate technical support for State D&H programs to conduct these campaigns, trainings and assessments.
6. Incorporate a policy requirement for inclusion of people with disabilities into health promotion campaigns in State D&H FOAs.
7. Engage in strategic collaboration with NCBDDD Public Health Resource and Information Centers to promote access to primary and periodic preventive care.
8. With other agencies, build capacity to analyze health services data and have those analyses inform policies to improve health care access.

Priority 5: Incorporate disability status as a demographic variable into all relevant CDC surveys, programs and policies.

Characterize the Problem: Prevalence, Incidence and Distribution

Four Year Outcome: Increase to 90% inclusion of disability identifiers in relevant CDC surveillance systems.

Progress Indicators:

1. Document the effects of the CDC Disability and Health Workgroup on incorporating disability status as a demographic variable into all current and future CDC surveys by resolving differences among disability identifier questions or incorporating the definition recommended as a result of the ACA.
2. Develop and demonstrate validity of standard disability identifiers.
3. Coordinate efforts to modify identified surveillance system units to include disability status as demographic variable in all relevant surveys in CDC.

Four Year Outcome: Disability status is included in all CDC reports on health disparities as a means of documenting possible disparities.

Progress Indicators:

1. Provide information and data for inclusion of disability-related disparities in OSELS annual disparities report.
2. Contribute to documenting health disparities experienced by people with disabilities.
3. Contribute to inter-agency planning on implementation of Affordable Care Act and related health care reform.

Four Year Outcome: Increased awareness of disability and health across CDC.

Progress Indicators:

1. Document the effects of the CDC Disability and Health Workgroup in recognizing and communicating best practices in disability and health.
2. Identify agency best practices and showcase those programs across CDC by establishing a web presence to spotlight successful programs.

Four Year Outcome: Increase to 90% inclusion of disability in CDC programs and policies, including FOAs and accessibility of printed and electronic materials.

Progress Indicators:

1. Document the effects of the CDC Disability and Health Workgroup in influencing and implementing policies in incorporating people with disabilities into relevant CDC programs and policies by examining opportunities for disability inclusion in programs and policies, including FOAs (current and future) and accessibility of printed and electronic materials.
2. Assure that identified CDC programs include disability in relevant programs and policies, including FOAs and accessibility of printed and electronic materials.

Acronym List – Appendix I

A
AAP – American Academy of Pediatrics
ACA – Affordable Care Act
ACD – Alveolar Capillary Dysplasia
ACF – Agency for Children and Families
ACO – Accountable Care Organization
ADDM – Autism and Developmental Disabilities Monitoring Network
ADHD – Attention Deficit Hyperactivity Disorder
AHRQ – Agency for Healthcare Research and Quality
ASD – Autism Spectrum Disorder

B
BRFSS – Behavioral Risk Factor Surveillance System

C
CADDRE – Center for Autism and Developmental Disabilities Research and Epidemiology
CBDRP – Centers for Birth Defects Research and Prevention
CDR Paralysis Resource Center – Christopher and Dana Reeve Paralysis Resource Center
CMV – Cytomegalovirus

D
DBA – Diamond Blackfan Anemia
DBD – Division of Blood Disorders
DBDDD – Division of Birth Defects and Developmental Disabilities
DDT – Division of Diabetes Translation
D&H – Disability and Health
DHDD – Division of Human Development and Disability
DHDSP – Division of Heart Disease and Stroke Prevention
DHQP – Division of Healthcare Quality Promotion
DNPAO – Division of Nutrition, Physical Activity and Obesity
DRH – Division of Reproductive Health
DVT – Deep Vein Thrombosis

E
EHDI – Early Hearing Detection and Intervention Program
EHR – Electronic Health Record

F
FAS – Fetal Alcohol Syndrome
FASD – Fetal Alcohol Spectrum Disorder
FDA – Food and Drug Administration
FOA – Funding Opportunity Announcement

G
GRUMA – Largest manufacturer of corn flour and tortillas in the world

H
HP 2020 – Healthy People 2020
HRSA – Health Resources and Services Administration
HTC – Hemophilia Treatment Center

I
IHS – Indian Health Service
IMMPCt – International Micronutrient Malnutrition Prevention and Control Program
IOM – Institute of Medicine
IQ – Intelligence Quotient

J

K
KABs – Knowledge, Attitudes and Beliefs

L
LFC – Legacy for Children
LTSAE – Learn the Signs. Act Early

M
MADDSP – Metropolitan Atlanta Developmental Disabilities Surveillance Program

N
NBDPN- National Birth Defects Prevention Network
NBDPS – National Birth Defects Prevention Study
NBS – Newborn screening
NCBDDD – National Center on Birth Defects and Developmental Disabilities
NCCDPHP – National Center for Chronic Disease Prevention and Health Promotion
NCHS – National Center for Health Statistics
NCIPC – National Center for Injury Prevention and Control
NHANES – National Health and Nutrition Examination Survey
NHIS – National Health Interview Survey
NHLBI – National Heart, Lung, and Blood Institute
NIAAA – National Institute of Alcohol Abuse and Alcoholism
NINDS – National Institute of Neurological Disorders and Stroke
NSFG – National Survey of Family Growth
NTD – Neural Tube Defect

O
OD – Office of the Director
OSELS – Office of Surveillance, Epidemiology and Laboratory Services

P
PE – Pulmonary Embolism
PWD – Persons with Disabilities
PWH – People with Hemophilia

Q

R
RuSH – Registry and Surveillance System for Hemoglobinophathies

S
SCD – Sickle Cell Disease
SEED – Study to Explore Early Development
SSRI – Selective Serotonin Reuptake Inhibitors

T
TWWIA – Ticket to Work and Work Incentives Act

U
UDC – Universal Data Collection

V
VWD – Von Willebrand Disease

W
WHO – World Health Organization
WIC – Women, Infants and Children program

X

Y

Z

Appendix II

Authorities

National Center On Birth Defect And Developmental Disabilities

NCBDDD General Authorities

Authority	PHSA/ Law Section	U.S. Code Section	Summary
Detail of Personnel	214	42 USC Sec. 215	Authorizes CDC, through HHS, to detail officers or employees to other departments or workplaces to cooperate in, or conduct work related to, the functions of HHS & CDC.
Public Health Research & Investigations	301	42 USC Sec. 241	General authority for public health research and investigations.
Research, Evaluation, & Demonstrations	304	42 USC Sec. 242b	General authority for research, evaluations, and demonstrations in health statistics, health services, and health care technology assessment.
International Cooperation	307	42 USC Sec. 242l	General authority for CDC to cooperate and collaborate with governments in other nations.
Confidentiality in data collection	308d	42 USC Sec. 242k	Provides for the protection of confidential data collected under selected surveys conducted under the authority of Sections 304, 306, and 307 of the Public Health Service Act.
Health Conferences and Publication of Health Education Materials-	310	42 USC Sec. 242o	Provides general authority for conferences of state health authorities, and to publish health education and information materials.
Cooperation with State & local health departments	311	42 USC Sec. 243	Provides general authority for cooperation with State and local health authorities.
Grant Authority for Preventive Health Services	317	42 USC Sec. 247b	Provides authority for CDC to make grants to State and local health authorities for preventive health services.
Public Health Emergencies	319	42 USC Sec. 247d	Provides authority for the HHS Secretary to declare and respond to public health emergencies.
Buildings & Facilities	319D	42 USC Sec. 247d-4	Provides authority for CDC facility and capacity expansion, and includes multi-year contract authority to expand training of personnel, improve communications facilities, networks and improve capabilities for public health surveillance and reporting.

Authority	PHSA/ Law Section	U.S. Code Section	Summary
Interdepartmental Cooperation	327	42 USC Sec. 254	Preserves authority for the Secretary to furnish any materials, supplies, or equipment, or perform any services requested by another federal department, or to request materials, supplies, or equipment from another federal department.
Preparation of Biological Products	352	42 USC Sec. 263	Provides authority for CDC to prepare biological products for its own use or for the use of other agencies, public or private organizations, or individuals engaged in medical work with such product is not available by another licensed source.
CDC Foundation	399G	42 USC Sec. 280e-11	Provides authority for the establishment of the CDC Foundation and the endowed programs managed by the Foundation.
Research Project Grants and Contracts	1102	42 USC Sec. 300b-1	Authorizes CDC to conduct and support research and grants for the study of genetic diseases and hemophilic conditions.
Technology Transfer		15 USC Sec. 3710	Authorizes CDC, through HHS, to transfer federally owned or originated technology to state and local governments and the private sector.
Bayh-Dole Act of 1980, P.L. 96-517	P.L. 96-517	35 USC Sec. 200-212	Allows for the transfer of exclusive control over government funded inventions to universities and businesses operating with federal contracts for the purpose of further development and commercialization.

NCBDDD Specific Authorities

Authority	PHSA/ Law Section	U.S. Code Section	Summary
NCBDDD establishment and general authority	317C	42 USC Sec. 247b–4	Provides for the establishment of the National Center on Birth Defects and Developmental Disabilities.
Folic Acid	317J	42 USC Sec. 247b–11	Authorizes CDC to conduct programs – education, training, research, and epidemiological studies – to address the effects of folic acid in prevention of birth defects.
Muscular Dystrophy Activities	317Q	42 USC Sec. 300b-7	Authorizes CDC to conduct and support research and grants for the study of Muscular Dystrophy.
Establishment of Fetal Alcohol Syndrome Prevention and Services Program	399H - K	42 USC Sec. 280f	Authorizes the establishment of a comprehensive Fetal Alcohol Syndrome and Fetal Alcohol Effect prevention, intervention and services delivery program.

Authority	PHSA/ Law Section	U.S. Code Section	Summary
Early Detection, Diagnosis, and Treatment Regarding Hearing Loss in Infants	399M(b)	42 USC Sec. 280g-1	Establishes the National Center on Birth Defects and Developmental Disabilities (NCBDDD), and provides authority for CDC programs in surveillance, applied research, and education related to birth defects; folic acid; cerebral palsy; mental retardation; child development; newborn screening; autism; fragile X syndrome; fetal alcohol syndrome; pediatric genetic disorders and disability prevention. This section also provides specific authority for the Spina Bifida Program.
Congenital Heart Futures Act (passed in 2010 as part of Health Reform)	399 V-2	42 U.S.C. 241 et seq	Provides authority for CDC to (1) conduct (or provide grants to State and local authorities to conduct) research, education and training, technical assistance, and evaluation for folic acid prevention of birth defects and (2) to facilitate further research into the types of health services patients use and to identify possible areas for educational outreach and prevention.
Developmental Disabilities Surveillance and Research Programs	399AA	42 USC Sec. 280i	Establishes medical professional, community, social services and youth education programs focused on the effects of fetal alcohol syndrome, as well as a prevention and diagnosis program. Authorizes the Secretary to make grants to the above programs, and creates a National Task Force to foster coordination between such programs.
Combating Autism Act: Authorization of Appropriations	399EE	42 USC 280i–4	Provides for authorized appropriations to conduct the activities described in 399AA and 399BB.
Tourette Syndrome	1108	42 USC Sec. 247b-11	Provides authority for CDC to conduct (or provide grants to State and local authorities to conduct) research, education and training, technical assistance, and evaluation for folic acid prevention of birth defects.

NCBDDD Shared Authorities

Authority	PHSA/ Law Section	U.S. Code Section	Summary
Research Relating to Preterm Labor and Delivery and the Care, Treatment and Outcomes of Preterm	301	42 USC 247b–4f	Provides for the expansion, intensification, and Coordination of the activities of the Centers for Disease Control and Prevention with respect to preterm labor and delivery and infant mortality.

and Low Birth Weight Infants			
Authority	**PHSA/ Law Section**	**U.S. Code Section**	**Summary**
Interagency Coordinating Council on Prematurity and Low Birth weight	399Q	42 USC 247b–4g	Provides authority to stimulate multidisciplinary research, scientific exchange, and collaboration among the agencies of the Department of Health and Human Services and to assist the Department in targeting efforts to achieve the greatest advances toward the goal of reducing prematurity and low birth weight.
Support for Patients Receiving a Positive Diagnosis of Down Syndrome or Other Prenatally or Postnatally Diagnosed Conditions	399R	42 U.S.C. 280g et seq	Provides authority to increase the provision of scientifically sound information and support services to patients receiving a positive test diagnosis for Down syndrome or other prenatal and postnatal diagnosed conditions.
Autism Education, Early Detection and Intervention	399BB	42 USC Sec. 280i-1	Provides authority for CDC to conduct research, make grants or cooperative agreements, and provide technical assistance to States relating to newborn and infant hearing screening.
Evaluating the Effectiveness of Newborn and Child Screening Programs	1110	42 U.S.C. 300b-9	Provides for authorized appropriations from FY09 – FY12.
Interagency Coordinating Committee on Newborn and Child Screening	1114	42 USC 300b-13	Provides for the establishment of an Interagency Coordinating Committee on Newborn and Child Screening to: (1) assess existing activities and infrastructure, including activities on birth defects and developmental disabilities authorized under section 317C, in order to make recommendations for programs to collect, analyze, and make available data on the heritable disorders recommended by the Advisory Committee on Heritable Disorders in Newborns and Children under section 1111, including data on the incidence and prevalence of, as well as poor health outcomes resulting from, such disorders; and (2) make recommendations for the establishment of regional centers for the conduct of applied epidemiological research on effective interventions to promote the prevention of poor health outcomes resulting from such disorders as well as providing information and education to the public on such effective interventions.

Gaining Input from Key Informants and Partner Stakeholder Groups:

Two strategies were used to gain input from partners. Key informants (e.g., cross-cutting experts in science, program and policy) were interviewed during one hour telephone or face-to-face meetings. NCBDDD leaders – the Acting Director, Deputy Director, Division Directors and senior subject matter experts - conducted these interviews. A standard series of questions were used and interview notes were carefully taken, sorted, and considered by program staff as the strategic plan was revised.

The second partner input strategy utilized a webinar format to gain input from a large group of constituent groups. Two webinars were held to inform and gain input from the Friends of NCBDDD and major partners. The first webinar described the rationale for creating the plan and laid out draft priorities and intended outcomes. The second webinar centered on identifying the Center and Division goals and priorities, as well as the intended outcomes that would be achieved through these activities. Particular attention was paid to explaining NCBDDD's underlying rationale behind the goals and priorities outlined in the plan and offering partners an opportunity to ask questions aimed at clarifying the plan's content. Partner input was carefully catalogued and considered for incorporation into the final strategic plan.

Partner Stakeholder Groups Included:
Academy for Educational Development
Alaska Native Tribal Health Consortium
Alpha-1 Foundation
American Academy of Audiology
American Academy of Family Physicians
American Academy of Orthopedic Surgeons (AAOS)
American Academy of Otolaryngology
American Academy of Pediatrics (AAP)
American Academy of Physical Medicine and Rehabilitation
American Academy of Physician Assistants
American Association of People with Disabilities (AAPD)
American Association of Poison Control Centers
American Association of Retired People (AARP)
American Association on Health Disability (AAHD)
American Association on Mental Retardation (AAMR)
American Baby Group
American College of Medical Genetics
American College of Nurse Practitioners
American College of Obstetricians and Gynecologists (ACOG)
American College of Physicians
American College of Preventive Medicine (ACPM)
American Diabetes Association
American Foundation for the Blind (AFB)
American Heart Association
American Medical Association (AMA)
American Occupational Therapy Association
American Psychological Association

American Public Health Association (APHA)
American School Health Association (ASHA)
American Society of Hematology (ASH)
American Society of Human Genetics
American Society of Pediatric Hematologists and Oncologists (ASPHO)
American Speech-Language-Hearing Association (ASHA)
American Therapeutic Recreation Association (ATRA)
American Thrombosis and Hemostasis Network
Amputee Coalition of America (ACA)
Arc of Georgia
Arc of the United States
Arthritis Foundation
Association for Prevention, Teaching and Research
Alexander Graham Bell Association for the Deaf and Hard of Hearing
Association of Maternal and Child Health Programs (AMCHP)
Association of Programs for Rural Independent Living (APRIL)
Association of Public Health Laboratories (APHL)
Association of State and Territorial Health Officials (ASTHO)
Association of Teachers of Maternal and Child Health (ATMCH)
Association of University Centers on Disabilities (AUCD)
Association of Women's Health, Obstetric and Neonatal Nurses
Asthma and Allergy Foundation of America
Atlanta Alliance on Developmental Disabilities (AADD)
Atlanta City Schools
Autism Coalition
Autism Science Foundation
Autism Society of America
Autism Society of America (ASA)
Autism Speaks
Backus Children's Hospital
Brain Injury Association of America
Brigham and Women's Hospital
Bristol-Myers Squibb
Bubel/Aiken Foundation
Buford City Schools
Business Edge
C.S. Mott Center for Human Growth and Development
California State University
Canadian Institute of Child Health
Case Western Reserve University
Catalina Health Resource
Catholic University of America
Cavarocchi - Ruscio - Dennis (CRD) Associates
CDC Foundation
Center for Assistive Technology and Environmental Access
Center for Child Health Improvement (CHI), The
Center for Development and Disability

Center for Disability Policy & Research
Center for Health and Healthcare in Schools
Center for Health Improvement
Center for Health Services Research and Policy
Center for Population Research, NICHD, NIH
Center for Rehabilitation Technology
Center for Science in the Public Interest
Child Care Bureau, HHS
Child Neurology
Child Study Center
Child Trends, Inc
Children and Adults with Attention Deficit/Hyperactivity Disorder (CHADD)
Children's Habilitation Center
Children's Healthcare of Atlanta
Children's Medical Service - Clayton
Christopher and Dana Reeve Foundation
Christopher Reeve Paralysis Foundation
Citizens United for Research in Epilepsy (CURE)
CityMatCH at University of Nebraska Medical Center
Clayton County Schools
Cobb County Schools
Coleman Institute for Cognitive Disabilities
Commonwealth Fund
Conceive Magazine
Conquer Fragile X
Cooley's Anemia Foundation (CAF)
Council of State and Territorial Epidemiologists (CSTE)
Covering Kids & Families
Cystic Fibrosis Foundation
Dan Marino Foundation
Daniella Maria Arturi Foundation
David Geffen School of Medicine at the University of California
Decatur City Schools
Dekalb County Board of Health
Dekalb County Schools
Department of Human Genetics, Emory University School of Medicine
Department of Human Services State of New Jersey
Department of Veterans Affairs
Diamond Blackfan Anemia Foundation
Diamond Blackfan Anemia Registry
Disability Policy Collaborative
Down Syndrome Research and Treatment Foundation
Dupont Hospital for Children
Easter Seals, Inc.
EDC Health & Human Development Programs
eHealth Forum
Emory Autism Resource Center

Emory Children's Center
Emory Clinic
Emory University School of Medicine
Exceptional Parent Magazine/EP Global
Family to Family for Health Choices/Parent to Parent of Georgia
Family Voices
FAS Family Resource Institute
First Signs (Autism Group)
Florida Department of Health
Fragile X Research Foundation (FRAXA)
Fulton County Dept. Health and Wellness
Fulton County Schools
GA DHR
Genetic Alliance, Inc.
Georgia Department of Education
Georgia SIDS Project
Georgia State University
Grady Health System
Gruma, SA
Gwinnett County Public Schools
Hands and Voices
Health Resources and Services Administration
Helen Keller Foundation for Research and Education
Helen Keller National Center for Deaf-Blind Youths and Adults
Hemophilia and Thrombosis Research Society
Hemophilia Federation of America
Hereditary Hemorrhagic Telangiectasia Foundation Intenational
Hope and Light Foundation
Hunter's Hope Foundation
Inova Fairfax Hospital
International Society of Thrombosis and Hemostasis
Iron Disorders Institute
Lakeshore Foundation
Learning Disabilities Association of America
M.I.N.D. Institute
M.I.S.S. Foundation
March of Dimes
March of Dimes Birth Defects Foundation
Maria Arturi Foundation
Marietta City Schools
Marion Downs Hearing Center
MCHB Leadership Education in Adolescent Health (LEAH) Program
Meharry Medical College
Michigan State University
Morehouse School of Medicine
Mt. Sinai Center for Children's Environmental Health and Disease Prevention Research
Muscular Dystrophy Association

Muscular Dystrophy Family Foundation
National Alliance for Autism Research (NAAR)
National Alliance for Hispanic Health
National Alliance for the Mentally Ill
National Alliance for Thrombosis and Thrombophilia
National Association for the Education of Young Children
National Association of Children's Hospitals
National Association of Children's Hospitals and Related Organizations
National Association of County and City Health Officials (NACCHO)
National Association of Development Disabilities Councils
National Association of Hispanic Nurses
National Association of Nurse Practitioners in Women's Health
National Association of Pediatric Nurse Associates & Practitioners (NAPNAP)
National Association of School Psychologists
National Association of State Head Injury Administrators
National Autism Association
National Birth Defects Prevention Network (NBDPN)
National Black Nurses Association
National Center for Gender Issues and Attention Deficit/Hyperactivity Disorder
National Center on Physical Activity
National Council of La Raza
National Council on Disability
National Council on Folic Acid
National Down Syndrome Society
National Foundation for Facial Reconstruction
National Fragile X Foundation
National Healthy Mothers, Healthy Babies Coalition
National Hemophilia Foundation (NHF)
National Hispanic Medical Association
National Institute on Alcohol Abuse and Alcoholism
National Marfan Foundation
National Medical Association
National Multiple Sclerosis Society
National Organization on Disability (NOD)
National Organization on Fetal Alcohol Syndrome
National Perinatal Association
New Jersey Medical School
Northside Hospital Perinatal Loss Offices
Oral Health America
Organization for Autism Research (OAR)
Organization of Teratology Information Services
Pan American Health Organization (PAHO)
Parent Project Muscular Dystrophy (PPMD)
Parents of Infants and Children with Kernicterus
Partnership for Prevention
Philadelphia College of Ostepathic Medicine
Reaching for the Stars

Research and Training Center for Children's Mental Health
Research and Training Center on Independent Living
Research and Training Center on Rural Disability
Research Autism
Research!America
Robert Wood Johnson Foundation
Rollins School of Public Health, Emory University
Saint Louis Arc
Saint Louis University School of Medicine
SHARE Atlanta: Pregnancy and Newborn
Shepherd Center
Sickle Cell Disease Association of America, Inc. (SCDAA)
SMA Foundation
Special Olympics
Spina Bifida Association of America
State Adolescent Health Coordinators Network
Teratology Society
The Arc of Georgia
The Arc of the United States
The Joey Company
The Marcus Institute
The NPD Group Inc.
The What to Expect Foundation
Tourette Syndrome Association
Trisomy 18 Foundation
Trust for America's Health
Tuberous Sclerosis Alliance
United Cerebral Palsy Associations (UCPA)
United Cerebral Palsy Research and Educational Foundation
University of Arizona
University of Georgia
University of Illinois at Chicago
University of Maryland
University of Miami
University of Missouri-Columbia
University of Nevada School of Medicine
University of Oklahoma Health Sciences Center
University of Texas School of Public Health, The
University of Virginia
University of Washington FAS Diagnostic Clinic
Unlocking Autism
USF College of Public Health
Yale School of Medicine and School of Nursing
Zero to Three

Overview of Major Birth Defects

Appendix IV

Major birth defects are abnormalities that can affect the structure or function of an organ. In the United States, major birth defects affect an estimated 3% of live births and they are the leading cause of infant death. There are more than 4,000 different known birth defects, ranging from minor to profound.

Birth defects develop during pregnancy, and most occur during the first three months of gestation. They may be caused by genetic, environmental or unknown factors. In many cases, the cause is thought to be an interaction of genetic and environmental factors.

Many experts classify birth defects into major categories: structural or metabolic defects, and defects caused by congenital infections. The most common example of a structural birth defect is congenital heart disease. Heart defects account for about one-third to one-quarter of all birth defects, accounting for one case per 100 – 200 births. Neural tube defects (spina bifida and anencephaly) are also structural defects, as are orofacial clefts (cleft lip, cleft palate, and combinations of the two).

Congenital infections may also cause birth defects, and rubella (German measles) and cytomegalovirus (CMV) are well known causes. Congenital rubella caries the highest risk for birth defects (about 20%), but because of universal immunization in the United States, it is now rarely seen. CMV is probably the most common congenital infection and is associated with intellectual disability and hearing loss.

Detailed case definitions exist for individual major birth defects, and in clinical practice, these defects are documented using the World Health Organization's International Classification of Diseases, 9th Revision, Clinical Modification.

In the United States, birth defects have accounted for over 139,000 hospital stays during a single year, resulting in $2.6 billion in hospital costs alone.