

BIRTH DEFECTS AND DEVELOPMENTAL DISABILITIES

Each year in the United States, 1 in 33 babies is born with a birth defect, amounting to 150,000 births. Another 3,000 babies are born with severe disorders that may not be obvious, but that can be detected—and treated—as early as possible, thanks to screening of dried blood spots collected at birth. In both arenas—prevention and early intervention—CDC has provided scientific leadership to laboratories, intervention programs, and media and education campaigns.

In about 25% of the cases, the causes of birth defects and developmental disabilities are known. For example, insufficient folic acid (a B vitamin) in pregnant women can lead to spina bifida (spine defects) and anencephaly (brain defects) in their infants; alcohol consumed during pregnancy can lead to Fetal Alcohol Syndrome. Because many women do not know they are pregnant until several weeks after conception, education about prevention measures (such as taking folic acid to prevent these defects, or avoiding alcohol) needs to reach women before they become pregnant. CDC is working to raise awareness of these and other prevention measures through media campaigns and specific education targeting physicians who provide routine medical care.

For the 75% of cases for which causes are still unknown, CDC supports national, regional, and local research efforts designed to discover the maternal, nutritional, genetic, and other risk factors that contribute to these conditions. Newborn screening is another detection and research tool that helps identify severe but hidden conditions as early as possible, when treatment can help make a difference. To help ensure a link between screening and follow-up, CDC is conducting a study of early detection and interventions related to hearing loss, which has potential application not only for other states but also for other conditions revealed by newborn screening. CDC also monitors the quality of newborn screening services across the country and is recognized as the worldwide expert in the most important tool for newborn screening: dried blood spot technologies.

ATTENTION-DEFICIT/HYPERACTIVITY DISORDER

WHAT IS THE PUBLIC HEALTH ISSUE?

Attention-Deficit/Hyperactivity Disorder (ADHD) is a neurobehavioral disorder that affects almost 4 million school-aged children and can last into adulthood. The symptoms characterized with ADHD are unusually high and chronic levels of inattention, impulsivity/hyperactivity, or both. Relationships with peers and family members and performance at school or work may significantly be impaired if untreated. Studies also show that ADHD patients tend to have higher injury and healthcare use rates. Many children with ADHD have other impairing behavioral or learning problems. Adolescents and adults who have ADHD and another serious mental health condition have shown in recent studies to participate in illegal substances, risky behavior, and/or criminal activities more often than children who do not suffer from ADHD.

The scientifically supported treatment for ADHD is multimodal, using both medication and behavioral therapies in combination. Controversies over the wide use and safety of medication treatments have increased over the last decades, and many questions still remain unanswered about the specific risks and benefits of medication therapies in the long-term.

WHAT HAS CDC ACCOMPLISHED?

- Sites in Oklahoma, South Carolina, and Virginia were awarded cooperative agreements to conduct community-based research on ADHD, which included population-based studies of prevalence, risk factors, coexisting conditions, and community treatment.
- The Federal Interagency Working Group on Child Mental Health (publishers of *America's Children*) and CDC have collaborated with other federal agencies to elicit heightened awareness about mental health conditions affecting children.
- CDC funded Children and Adults with Hyperactivity-Attention Deficit Disorder (CHADD) to establish the National Resource Center on ADHD.
- CDC designed and implemented a study methodology for conducting the community-based research in three locations in the United States.
- CDC published a peer-reviewed journal article on ADHD in children and associations with maternal mental health status using a national data set.

WHAT ARE THE NEXT STEPS?

- Complete data collection for community-based ADHD studies and begin data analysis in collaboration with grantees.
- Continue to work with partners to promote data collection and to apply epidemiologic methodology to the study of ADHD.
- Strengthen collaborations with federal and nonfederal partners.
- Sustain the development and distribution of ADHD health education and outreach materials.
- Work with CHADD to further develop the National Resource Center on ADHD.

DISABILITY AND HEALTH

WHAT IS THE PUBLIC HEALTH ISSUE?

Fifty-four million Americans are living with a functional limitation associated with a long-term physical, sensory, or cognitive condition. The annual economic cost associated with disabling conditions is an estimated \$300 billion. People with disabilities report higher rates of depression, anxiety, obesity, and acute pain in comparison to non-disabled persons. People with disabilities also report less access to healthcare compared to non-disabled populations.

CDC sponsors programs supporting the health, well-being, independence, productivity, and full societal participation of people with disabilities. These programs support research on risk factors for poor health and well-being; research on measures of health, functioning, and disability; data collection on the prevalence of disabilities and the health status of people with disabilities; health promotion interventions; and the implementation of public health policies related to disability and health. These initiatives also work to reduce the incidence and severity of secondary conditions such as pressure sores (decubitus ulcers), urinary tract infection, depression, and obesity.

WHAT HAS CDC ACCOMPLISHED?

- There are 16 states supported by CDC for various activities including disability surveillance, health promotion interventions, health education, and resource and policy development. The states awarded funding are Arkansas, California, Illinois, Iowa, Massachusetts, Montana, New Jersey, New York, New Mexico, North Carolina, Oregon, Rhode Island, South Carolina, Virginia, Vermont, and Washington.
- Extramural research projects have been implemented to increase the understanding of occurrence, risk/protective factors, costs associated with secondary conditions, and the impact of disability among special populations such as women, children, and racial/ethnic minorities.
- Three National Information Centers were awarded funds to provide comprehensive information and resource material to people with disabilities, their families, caregivers, and healthcare professionals. These centers include the National Center on Physical Activity and Disability, the National Limb Loss Information Center, and the Christopher and Dana Reeve Paralysis Resource Center.
- Conferences were supported to facilitate and stimulate dialogue; disseminate and exchange information; establish research and policy priorities; and outline and undertake next steps.
- CDC funded *Living Well with a Disability*, a community-based intervention which demonstrated that health promotion activities targeting people with disabilities not only improve health and wellness but also save money by reducing the frequency of medical visits.

WHAT ARE THE NEXT STEPS?

- Continue to translate research findings into practice to improve the lives of people living with disabilities, including encouraging the implementation of the *Living Well with a Disability* program.
- Develop and disseminate health information to individuals with disabilities and their families.

For additional information on this or other CDC programs, visit www.cdc.gov/program

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EARLY HEARING DETECTION AND INTERVENTION

WHAT IS THE PUBLIC HEALTH ISSUE?

Hearing loss occurs in 12,000 babies each year (3 of every 1,000). Developmental delays can occur if hearing loss goes undetected. Early Hearing Detection and Intervention (EHDI) is a national initiative that supports the early identification of infants with hearing loss. The initiative uses hearing screening; audiologic and medical evaluation; and enrollment in early intervention services when appropriate. The average age of identification of children with hearing loss is 1.5 to 3 years of age, without EHDI programs, which research suggests is already past the start of the critical period for optimal speech and language acquisition. Families and professionals can make sure the child receives intervention services at an early age if a child's hearing loss is identified soon after birth. EHDI promotes early treatment for children with hearing loss to develop communication and language skills that will last a lifetime.

WHAT HAS CDC ACCOMPLISHED?

- A multi-state study was launched to evaluate the causes of hearing loss in newborns and young children.
- A prototype database was developed to provide state profiles. All EHDI information will be available to state health departments, other government agencies, parents, and other interested parties.
- A study in Utah was financed to determine the economic costs of screening, follow-up, and diagnosis associated with EHDI programs.
- Funding was provided for a study in Hawaii to evaluate the effectiveness of early identification and intervention in children with hearing loss.
- CDC initiated a study on the accuracy of the two-stage method of screening infants for hearing loss to determine if some infants with mild loss are missed.
- CDC partnered with the Health Resources and Services Administration to fund a multi-state investigation to determine the reasons children do not complete the EHDI process and to develop strategies to reduce this "loss to follow-up."
- CDC funded 30 states to track infants who screen positive for hearing loss to determine if they receive appropriate diagnostic and intervention services.

WHAT ARE THE NEXT STEPS?

- Continue to work with states to develop a more comprehensive database for monitoring progress on the following national *Healthy People 2010* goals: all infants are to be screened for hearing loss before 1 month of age, preferably before they are discharged from the hospital; infants who screen positive for hearing loss will be referred for an audiological evaluation before 3 months of age; and infants with confirmed hearing loss will be referred for comprehensive medical and intervention services before 6 months of age.
- Move from a prototype to actual usable database for state profiles.
- Continue to conduct and disseminate results from studies into causes of hearing loss and the effectiveness of screening and early intervention programs.

IMPROVING THE LIVES OF PEOPLE WITH LIMB LOSS

WHAT IS THE PUBLIC HEALTH ISSUE?

The 1996 National Health Interview Survey indicates that nearly 12 million people in the United States have an extremity absence, or limb loss (excluding tips of fingers or toes only). The prevalence rate averages 4.9 per 1,000 persons with an extremity loss. Data from the Healthcare Costs and Utilization Project, National Inpatient Sample indicate that between 1988 and 1996 an average of 133,235 limb-loss-related hospital discharges occurred per year, and 82% of these were due to vascular conditions such as diabetes and peripheral vascular disease. The risk of amputation increases substantially with age, mainly due to age-related risks including diabetes, cancer, and cardiovascular complications. Men generally are at higher risk than women, and African Americans have a higher risk of amputations due to higher occurrences of vascular disease. People who experience limb loss often express concerns about the inadequacy of prostheses and the isolation in conjunction with a relatively rare experience.

Limb loss often results in reduced mobility and increased reliance on prosthesis for daily activities like walking and climbing stairs. While current technologies allow for a more complete range of motion and ability, the advances in products and services do not address all health concerns. Conditions for which people with limb loss may be at increased risk include pressure sores, muscle atrophy/deterioration, depression, and obesity. These conditions can be prevented through education and health promotion programs.

WHAT HAS CDC ACCOMPLISHED?

- Johns Hopkins Bloomberg School of Public Health was awarded a cooperative agreement that supports research on the epidemiology and consequences of limb loss and limb deficiency.
- CDC financed the Amputee Coalition of America to develop and operate the National Limb Loss Information Center (NLLIC). The programs and services of NLLIC include a national hotline, a website, referral services, educational curricula, youth programs, a national peer network, consumer publications, fact sheets, and a library catalog.
- CDC funded the University of Washington School of Medicine to study the impact of social support among people living with limb loss.

WHAT ARE THE NEXT STEPS?

- Ensure that people with limb loss are included in all programs designed to improve the health and well-being of people with disabilities.
- Develop additional interventions aimed at promoting general health and improved quality of life for people with limb loss and limb deficiency.
- Expand research on vulnerable populations such as ethnic minorities and people with diabetes.

IMPROVING THE LIVES OF PEOPLE WITH PARALYSIS

WHAT IS THE PUBLIC HEALTH ISSUE?

Much still needs to be done to improve the overall health of people with paralysis. Some of the secondary conditions they suffer from include pressures sores (decubitus ulcers), urinary tract infections, depression, and obesity, which can be prevented through education and health promotion programs.

CDC has established a national coordinating facility to provide educational materials, referral services, and self-help guidance for people with paralysis, their families, and caregivers. Healthcare providers and professionals also receive information on paralysis, methods to prevent secondary conditions and address quality of life issues. CDC is expanding the dissemination of educational outreach materials to increase the knowledge and understanding of paralysis among those most affected. Collaborative national relationships have been established with rehabilitation facilities, hospitals, and disability advocacy and voluntary support groups. CDC is developing the ability to measure the public health impact of paralysis through demographic and statistical research with a focus on the prevalence, causes, and extent of secondary conditions. CDC will provide leadership in helping to facilitate health promotion activities (e.g., improving physical activity, exercise and nutrition, confronting depression/isolation issues, managing weight, quitting tobacco use) among people with paralysis to enhance physical and emotional health.

WHAT HAS CDC ACCOMPLISHED?

- CDC funded two studies to address the role of supportive social relationships among people with paralysis. These studies were conducted at the Medical University of South Carolina and the University of Nevada, Reno.
- CDC has supported the Christopher and Dana Reeve Paralysis Resource Center, which was established to help improve the quality of life for people with paralysis. The center is a coordinating facility with information specialists, a library, and a website providing educational materials, referral services, and self-help guidance to those living with paralysis, their families, and their caregivers. In addition, the center funds community-based projects to improve quality of life for people with paralysis.

WHAT ARE THE NEXT STEPS?

- Assist the Christopher and Dana Reeve Paralysis Resource Center to extend services and increase the impact of these services for all individuals and families who can benefit from them.
- Ensure that people with paralysis are included in all programs designed to improve the health and well-being of people with disabilities.
- Support additional research to expand the understanding of the public health impact of paralysis.

LEGACY FOR CHILDREN™

WHAT IS THE PUBLIC HEALTH ISSUE?

The effects of parenting styles on children's developmental outcomes require further investigation to enable more readily available information. CDC is conducting a long-term study of child development at two inner-city research sites (Miami and Los Angeles). This research study will attempt to determine whether low-income mothers can improve the long-term developmental outcomes of their children through increased maternal investment of time and energy and by fostering the belief in the "power of the parent." The CDC community-based early intervention study, *Legacy for Children™ (Legacy)* is a randomized, controlled trial that will test the *Legacy* early intervention model, which focuses on improving maternal self-efficacy, building a sense of community, and increasing parental investment through weekly mothers' groups. These groups provide mothers with a time and place to play with their children, have meetings to discuss their babies' development, and explore their beliefs about parenting. *Legacy* does not provide social services, but instead relies on the strength of each mother to work through her own family challenges with the support and camaraderie of other mothers. The understanding is that each *Legacy* group functions as a time and place for participants to focus on and invest in their children's development.

WHAT HAS CDC ACCOMPLISHED?

- Adapted the main study based on lessons learned in the pilot study.
- Completed recruitment for the study at the University of California at Los Angeles (UCLA) in January 2004 and at the University of Miami in September 2003.
- Completed the intervention phase of the pilot study at UCLA in July 2003.
- Presented process data from the pilot study at national conferences.
- Updated the intervention program for mothers to keep them engaged.

WHAT ARE THE NEXT STEPS?

- Continue to present process data from the pilot study at national conferences.
- Continue to use the Legacy Working Group to guide the study, collect data, and disseminate information as it becomes available.
- Make public a manuscript on methods in the *Legacy* study and on the "Sense of Community in Legacy for Children."
- Publish a manuscript on lessons learned from the pilot study.

LONG-TERM FOLLOW-UP OF NEWBORN SCREENING

WHAT IS THE PUBLIC HEALTH ISSUE?

Phenylketonuria (PKU) is a genetic disorder that affects the body's ability to convert certain proteins found in foods. Newborn screening was implemented in the 1960s to provide early diagnosis of PKU. Identifying the disorder early allows dietary management to prevent the development of mental retardation in these children. Since then, newborn screening programs have expanded to incorporate additional conditions such as hearing loss and disorders which affect a person's metabolism, blood, or hormones. Newborn screening programs have traditionally been limited to conditions that are serious, treatable, or controllable, influenced by age of diagnosis, and with a natural history that is understood.

In the 1990s the technology of tandem mass spectrometry was introduced for population-based newborn screening. This technology allows for more accurate measurement of a broader range of conditions than previously available. For some conditions, such as PKU, the benefits of newborn screening and early treatment are generally accepted. However, for other conditions there is little information on the long-term benefit from screening and intervention. Population-based tracking and follow-up studies of children identified through the latest technology are needed to assess the public health impact of newborn screening for many of the disorders identified with this technology. Ideally, this approach will include collection of information related to treatment options, treatment compliance, and long-term outcome. The infrastructure developed for this assessment can also be used in the future for evaluation of new newborn screening programs.

WHAT HAS CDC ACCOMPLISHED?

- Created hypothesis for assessing outcomes.
- Funded Iowa and a collaboration in Oregon and Idaho to develop a database system for long-term follow-up of newborn screening.
- Finalized research protocol for the collection and analysis of medical records data.
- Developed a database to assess the long-term effects of newborn screening.

WHAT ARE THE NEXT STEPS?

- Add component of standard developmental assessment for all children within study group.
- Complete database for long-term newborn screening assessment.
- Review records, collect data, and analyze long-term assessment of newborn screening.

MUSCULAR DYSTROPHY

WHAT IS THE PUBLIC HEALTH ISSUE?

Duchenne muscular dystrophy (DMD) affects about 1 in 4,000 males and is the most common form of muscular dystrophy in children. In the absence of newborn screening, DMD is usually diagnosed around children 3 to 6 years of age. Early signs include failure to walk by 18 months, frequent falling, difficulty getting up from a sitting or lying position, and a waddling gait. As muscle deterioration progresses, children with DMD become unable to walk around 12 years of age. The disease is fatal in the teens or early twenties, due to severe respiratory and heart problems. A milder form of the disease, Becker muscular dystrophy, is caused by mutations in the same gene. The combined spectrum is referred to as DBMD. Standard birth-defects monitoring systems in the United States do not detect children with DBMD because children do not have recognizable signs or symptoms at birth. Consequently, existing birth-defects monitoring systems would need to be supplemented with additional activities to find all cases of DBMD.

In addition to Muscular Dystrophy Association clinics and other muscular dystrophy clinics, cases may be ascertained through state or regional muscular dystrophy chapters or associations, hospitals, private physicians, and diagnostic laboratories. The lack of a uniform standard of care for DBMD results in inter-clinic variation in treatment options. A long-term follow-up study of children with DBMD is necessary to evaluate different treatment options and to develop and evaluate standards of care. In addition, these activities will serve as the basis for a population-based assessment of the impact of DBMD.

WHAT HAS CDC ACCOMPLISHED?

- Convened meetings with stakeholders to identify key epidemiologic research questions as well as research needs related to families with DBMD.
- Developed a plan for monitoring activities and awarded cooperative agreements to Arizona, Colorado, Iowa, and New York.
- Financed a survey of carrier females, women who carry the mutation for DBMD, but do not have DBMD.
- Funded the Children's National Medical Center in Washington, D.C., to develop materials and conduct a family needs assessment.
- Funded the University of Iowa to develop a system that can pool data across sites and conduct a qualitative study of family needs.
- Hired a parent consultant and public health genetic counselor to assist in various aspects of the program.
- Supported the efforts of existing state partners to enable them to conduct annual interviews of families participating in the family needs assessment project.
- Work with grantees to develop data collection strategies for DBMD.

WHAT ARE THE NEXT STEPS?

- Add an additional state to the surveillance network (depending on objective review panel findings).
- Continue to work closely with stakeholders, keeping them updated as new activities become operational.
- Convene national expert meetings on newborn screening for DBMD and on treatment.
- Fund one to two states to conduct a feasibility planning study for newborn screening for DBMD.
- Work with grantees to develop optimal case finding activities.

For additional information on this or other CDC programs, visit www.cdc.gov/program

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NATIONAL RESOURCE CENTER ON ATTENTION-DEFICIT/HYPERACTIVITY DISORDER

WHAT IS THE PUBLIC HEALTH ISSUE?

Attention-Deficit/Hyperactivity Disorder (ADHD) is a neurobehavioral disorder that is characterized by problems with attention, impulsivity, and hyperactivity. The disorder affects almost 4 million school-aged children and can last into adulthood. If the condition goes untreated or unmanaged, significant difficulties in crucial areas of life such as relationships with peers and family members may occur. Performance at school or work can also be impaired.

ADHD affects between 3% and 7% of school-age children, in addition to 2% to 4% of the adult population. Almost 4 million youth ages 3 to 17 (6%) have been diagnosed with ADHD. Statistics show that boys are more than twice as likely as girls to have an ADHD diagnosis.

The National Resource Center on ADHD was opened in May 2003, to service the needs of many individuals and families seeking information and assistance related to ADHD. The center is the country's first and only national clearinghouse dedicated to evidence-based science and treatment of ADHD. Within the first 8 months of operation, the center averaged 1,200 inquiries (phone, Web, mail) per month and this figure is only expected to grow as the center expands its outreach activities. The center provides pragmatic and scientific data to clinicians, the public, and partner organizations which heighten awareness and strengthen support for ADHD.

WHAT HAS CDC ACCOMPLISHED?

- Collaborated with the Children and Adults with ADHD (CHADD) to create the ADHD National Resource Center. CHADD is a national non-profit organization providing education, advocacy, and support for individuals with ADHD.
- The center is the first and only national clearinghouse dedicated to the sharing of evidence-based science and treatment information about ADHD to the public and to professionals.
- Provides information about ADHD and associated concerns to the public and to various professional groups through their website (www.help4adhd.org), their telephone hotline (1-800-233-4050), and their electronic and onsite libraries. The materials available online and the assistance provided by telephone are available in English and Spanish.

WHAT ARE THE NEXT STEPS?

- Build capacity for the National Resource Center on ADHD to expand its ability to respond to requests by increasing the number of information specialists available to address caller needs.
- Continue to build and make available valuable and reliable resources through the online resource library and physical clearinghouse of ADHD resources.
- Evaluate the satisfaction of consumers who use the center so that program improvements can be made.
- Expand efforts to outreach to diverse communities with information about ADHD.

For additional information on this or other CDC programs, visit www.cdc.gov/program

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USING DEVELOPMENTAL SCREENING TO IMPROVE CHILDREN'S HEALTH

WHAT IS THE PUBLIC HEALTH ISSUE?

Children with developmental or behavioral disabilities are missing vital opportunities for early detection and intervention. In the United States, 12% to 16% of children have a developmental or behavioral disability including autism, mental retardation, and Attention-Deficit/Hyperactivity Disorder. However, only 50% of these children are identified as having a problem before starting school, by which time significant delays may have already occurred and opportunities for treatment have been missed.

Developmental screening is a brief assessment designed to identify children who should receive more intensive diagnosis or assessment and to provide early detection and intervention which can make a significant difference and reduce the need for more costly interventions. The health and well-being of children, especially children with developmental disabilities, can improve from developmental screenings. For example, the National Academy of Sciences concluded that early detection and intervention significantly improves outcomes for children with autism. As many as 85% to 90% of children with autism who participate in systematic intervention before they are 5 years of age gain the ability to talk, which helps them reach their full potential.

The healthcare system is one of the few places where children under 5 years of age are consistently seen and has the ideal system for tracking child development through screening. However, because of time constraints and concerns about financial reimbursement, most primary care providers either do not conduct developmental screening or rely on informal methods that identify less than half of the children with developmental and/or mental health problems.

WHAT HAS CDC ACCOMPLISHED?

- Developed a developmental screening public health research agenda and developed recommendations with key stakeholders.
- Developed a pilot screening program for autism and other developmental disabilities in Philadelphia.
- Co-sponsored and presented findings at the Health and Human Services Summit on Autism in November 2003.
- Devised an initial plan for a public health campaign to increase awareness in parents and key stakeholders on the importance of screening for autism and other developmental disabilities.
- Created intervention and the need for systematic developmental screening and assessment.
- Launched a CDC Developmental Screening website.

WHAT ARE THE NEXT STEPS?

- Acquire a manuscript on existing developmental screening data sources and systems.
- Develop other pilot screening and early intervention programs for developmental disabilities.
- Improve the capacity to monitor nationwide the integration of developmental screening into primary care and other relevant systems.
- Launch a public health campaign to increase awareness in parents and key stakeholders on the importance of screening for autism and other developmental disabilities.

For additional information on this or other CDC programs, visit www.cdc.gov/program

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