IMPROVING CHILD HEALTH

THE ROLE OF POLICYMAKERS IN PREVENTION AND TREATMENT OF BIRTH DEFECTS AND DEVELOPMENTAL DISABILITIES

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SUMMARY AND ANALYSIS OF KEY POLICY OPTIONS FOR DETECTION AND PREVENTION OF BIRTH DEFECTS AND DEVELOPMENTAL DISABILITIES

A number of key policy issues were identified at the National Conference of State Legislatures’ meeting, “Improving Child Health: The Role of Policymakers in Prevention and Treatment of Birth Defects and Developmental Disabilities,” held in August 2007. This summary and analysis identifies three general areas examined in breakout groups during the meeting: preconception and prenatal period, neonatal period, and early childhood. The document highlights numerous major policy issues but is not a comprehensive examination of all policy approaches to detection and prevention of birth defects and developmental disabilities.

PRECONCEPTION AND PRENATAL PERIOD

According to the March of Dimes, the preconception and prenatal periods are critical times in the course of a woman’s reproductive life. Avoiding toxic exposures and practicing healthy behaviors can decrease the likelihood of preventable birth defects and developmental disabilities. Examples of some of the ways policymakers may help reduce the number of children with birth defects and developmental disabilities include the following:

- Educating women about the importance of taking folic acid prior to conception;
- Encouraging a pre-pregnancy checkup;
- Offering information about or treatment services for substance abuse or smoking cessation; and
- Sharing resources for carrier screening.

FOLIC ACID

Most states have conducted or have ongoing media campaigns about the importance of taking folic acid prior to and during pregnancy to prevent neural tube defects (NTDs). For some women, learning that taking prenatal vitamins with the appropriate amount of folic acid may not be sufficient if they are not able to afford them. For this reason some states have expanded their folic acid activities to include the provision of prenatal vitamins. For example, the Alabama Department of Public Health (ADPH; http://www.adph.org/perinatal/) and the Alabama Birth Defects Surveillance and Prevention Program offer free folic acid supplements to women planning a pregnancy or to women who have previously given birth to children with neural tube disorders for four months. According to the March of Dimes, states play a vital role in preventing birth defects by maintaining birth defects monitoring programs, which collect data for detecting trends and suggest areas for further research, and link families to services.

State Women, Infants, and Children (WIC) programs also may distribute vitamins, but because these programs serve women only during pregnancy and postpartum, they do not provide services to
women before they become pregnant. The Centers for Disease Control and Prevention (CDC)'s National Center for Birth Defects and Developmental Disabilities website (http://www.cdc.gov/ncbddd/folicacid/campaigns.htm) highlights other current state and local activities concerning folic acid.

Other recommendations
The CDC also makes 10 Recommendations to improve preconception health and care, which are listed below.

1. Individual Responsibility Across the Lifespan. Each woman, man, and couple should be encouraged to have a reproductive life plan.

2. Consumer Awareness. Increase public awareness of the importance of preconception health behaviors and preconception care services by using information and tools appropriate across various ages; literacy, including health literacy; and cultural/linguistic contexts.

3. Preventive Visits. As a part of primary care visits, provide risk assessment and educational and health promotion counseling to all women of childbearing age to reduce reproductive risks and improve pregnancy outcomes.

4. Interventions for Identified Risks. Increase the proportion of women who receive interventions as follow-up to preconception risk screening, focusing on high priority interventions (i.e., those with evidence of effectiveness and greatest potential impact).

5. Interconception Care. Use the interconception period to provide additional intensive interventions to women who have had a previous pregnancy that ended in an adverse outcome (e.g., infant death, fetal loss, birth defects, low birthweight, or preterm birth).

6. Prepregnancy Checkup. Offer, as a component of maternity care, one prepregnancy visit for couples and people planning pregnancy.

7. Health Insurance Coverage for Women with Low Incomes. Increase public and private health insurance coverage for women with low incomes to improve access to preventive women's health and preconception and interconception care.

8. Public Health Programs and Strategies. Integrate components of preconception health into existing local public health and related programs, including emphasis on interconception interventions for women with previous adverse outcomes.

9. Research. Increase the evidence base and promote the use of the evidence to improve preconception health.

10. Monitoring Improvements. Maximize public health surveillance and related research mechanisms to monitor preconception health.

http://www.cdc.gov/mmwr/preview/mmwrhtml/rr5506a1.htm

PRE-PREGNANCY CHECK-UP
Participants in NCSL’s meeting on birth defects and developmental disabilities focused on one of the CDC recommendations—encouraging a pre-pregnancy checkup—as a key promising strategy.
for improving the health of infants and children. This may afford health care providers an opportunity to educate patients about how to prepare their bodies for a future pregnancy by taking folic acid, eating healthy, avoiding alcoholic beverages or other substances, and quitting smoking. In addition to the CDC, organizations such as the March of Dimes recognize the value of a pre-pregnancy checkup and the potential to change damaging behaviors such as smoking that could lead to prematurity and associated complications. Interested policymakers may want to consider the state’s role in providing access to a pre-pregnancy visit and to services that the patient may need to follow up on the information provided during the check-up. For example, legislators may want to find out whether a pre-pregnancy checkup is available through state programs or private insurance and whether intervention services for women identified with substance abuse problems are available.

State involvement in the delivery of intervention services may help to ensure that women receive necessary care but also may raise questions about confidentiality, reporting requirements, and cost. Another complementary or alternative strategy that states have employed to educate women about this issue is to require that notices be posted in liquor stores or other businesses where alcohol is served about the risks of consuming alcohol during pregnancy. Some states also require distribution of information about the dangers of alcohol or drugs during pregnancy on other occasions such as in Hawaii where materials are provided prior to issuing a marriage license.

**GENETIC SCREENING**

While avoiding dangerous exposures can reduce the number of some birth defects, others are inherited. In these instances, genetic screening may help detect or prevent heritable conditions. Prior to becoming pregnant, screening can determine whether a genetic mutation is present for a disorder that causes little or no symptoms in the individual but may be passed to children if both parents carry the mutation. The results of testing can alert families to potential risk to future offspring or help prepare them for a child with special needs.

A variety of prenatal screening tools are widely available, including the following:

- chorionic villous sampling—a test done during early pregnancy that can find certain problems with the fetus
- nuchal translucency—a test using an ultrasound to measure the clear (translucent) space in the tissue at the back of developing baby’s neck to test for risk of having Down syndrome and other chromosomal abnormalities
- maternal serum screening—a blood test performed on the mother
- amniocentesis—a sample of amniotic fluid testing for abnormalities

Recent technological advances are bringing new, more extensive screening to consumers through the use of microarrays, which can screen for many abnormalities at once.

States may have a stake in preconception and prenatal genetic screening as service providers, educators, or facilitators. Preconception care may be addressed through preventive visits, interconception care and pre-pregnancy checkups. State services for pregnant women vary, and may include performance of or referral for maternal serum screening, ultrasound imaging, amniocentesis and genetic counseling. Factors to consider in determining the degree of services available through the state may include cost, consumer demand, and whether genetic counseling and other follow-up services will be provided. See California’s Genetic Disease Screening Program as an example (http://www.cdph.ca.gov/programs/GDSP/).
State legislatures play several roles to ensure that newborns with certain serious medical conditions receive necessary care in the first days of life. State newborn screening programs screen an estimated 4.1 million infants annually for genetic and metabolic disorders. Early detection and treatment of these abnormalities can prevent severe disability, mental retardation or even death and may also save states and families money by avoiding additional high medical costs and state institutional services. See New Jersey's program, New Jersey Special Child Health Services Registry (http://www.ncsl.org/print/health/LSochka807.pdf) as an example.

In 2005 an American College of Medical Genetics report, Newborn Screening: Toward a Uniform Panel and System (http://www.mchb.hrsa.gov/screening/), commissioned by the Health Resources and Services Administration, recommended screening for 29 core conditions and identified 25 other conditions that could be detected at the same time. According to the National Newborn Screening and Genetics Resource Center (NNSGRC), as of February 2008, 37 states required screening for at least 30 disorders, and some required screening for as many as 50 conditions. Only 18 states and the District of Columbia actually required screening for the core 29 conditions. In some cases whether an infant with a possibly fatal condition grows into a healthy child may still depend on the state in which he or she is born.

Pre-birth education about newborn screening to parents may also prevent children from falling through the cracks and provides an opportunity to answer their questions about the newborn screening process. According to research by NCSL and the National Newborn Screening & Genetics Resource Center (NNSGRC) (published in Pediatrics in May 2006; http://pediatrics.aappublications.org/cgi/reprint/117/5/S1/S212) 20 states require the provision of educational information to a parent or guardian prior to screening. Parent educational materials may discuss:

- the conditions that their infant will be screened for
- the need for follow-up testing
- the potential to retain samples collected
- confidentiality and privacy issues
- the right to refuse screening
- the consequences of accepting or refusing treatment

A survey of genetic services coordinators who provide follow-up services, which was conducted by NNSGRC, found “of the 50 programs with a newborn screening pamphlet, 10 programs (20%) reported that distribution was a usual activity of obstetricians; 14 programs (27%) reported that pamphlet distribution was a usual activity of prenatal classes. Although it is thought that most birthing facilities distribute program information pamphlets as part of their information packets for new mothers, only 19 programs (40%) reported having a requirement for such distribution.” The survey results also demonstrated that the contents of pamphlets varied. With support from HRSA, the American Academy of Pediatrics has developed examples of a brochure for parents (http://www.medicalhomeinfo.org/screening/Screen Materials/Newborn screening tests.pdf) and a
brochure for providers (http://www.medicalhomeinfo.org/screening/Screen Materials/Newborn screening disorders.pdf) on newborn screening that may serve as a guide for states.

Once children are identified with a condition, what happens to them in the long-term is not well understood. All states have well-established short-term follow-up procedures. The use of newborn screening Action (ACT) sheets (http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm) created by the American College of Medical Genetics may help states and providers continue to carry out effective short-term follow-up as programs expand. The ACT sheets describe the short-term actions a health professional should follow in communicating with a family whose infant has tested positive for a genetic condition. The sheet also recommends appropriate steps in the follow-up of the infant and gives an overview of the basic steps involved in determining the final diagnosis.

Long-term follow-up throughout childhood, however, varies across the country and is uncommon. An effort to develop a Newborn Screening Translational Research Network would “establish an infrastructure for research that facilitates the development of new screening methods, clinical trials for new therapeutic interventions and support longitudinal research to study the long-term health of children identified through newborn screening,” according to the Department of Health and Human Services and the Eunice Kennedy Shriver National Institute for Child Health and Human Development (NICHD). Collaboration with the new network may help states to improve their long-term follow-up programs.

The use of newborn screening samples also may prove beneficial for research into other diseases. The length of time that states require retention of newborn screening samples range from as little as one month to an indefinite period. The pool of newborn screening blood specimens is a unique opportunity to study a wide range of the population since most infants born in the U.S. participate in the program. However, government use of a child’s screening sample and information, even if for anonymous research, may raise serious concerns for families about the potential for discrimination and violations of privacy. Some states offer parents the option to have samples destroyed or retained for the purpose of research. States interested in developing consent procedures to inform parents about the opportunity to participate in research may want to consider the following:

- Whether research will be confined to research on the prevention of birth defects and developmental disabilities;
- If institutional review board requirements exist to ensure ethical research;
- How or if families may be recontacted in the event research findings may be of benefit to them; and
- Whether current protections from genetic discrimination in employment and insurance are adequate to allay fears about studies that may reveal possible genetic predisposition to future illness.

States play a vital role in preventing birth defects by maintaining birth defects monitoring programs. These programs collect data for detecting birth defects trends and suggest areas for further research. They also link people to needed services.
Developmental delays and conditions are common in early childhood, affecting at least 10 percent of children. Recent studies emphasize the importance of brain development within the first three years of life. Early developmental delays are markers for later developmental conditions such as autism, intellectual disability, hearing or vision impairment, cerebral palsy, speech and language disorders, and learning disabilities. Identifying these problems early on can help children benefit from treatment and their parents from guidance.

According to recent studies, early developmental delays are often not identified in a timely way. Many children are not identified until kindergarten entry or later—well beyond the period in which early intervention is most effective. Therefore, in many cases, opportunities to intervene early to improve children’s developmental outcomes are missed.

Pediatricians and other primary care medical providers who see children for well-child visits during the first few years of life can play a key role in the early identification of developmental delays. Pediatricians and other primary care practitioners receive training in child development and are often trusted by families. According to the American Academy of Pediatrics (AAP), pediatricians are the best informed professionals in regular contact with families during the early childhood period; thus, they are uniquely equipped to perform developmental screenings and refer families for further testing or services. Please see the AAP’s policy statement (http://aappolicy.aappublications.org/cgi/content/full/pediatrics;108/1/192).

Opportunities exist for providers to identify delays during the recommended 16 well-child visits between birth and 5 years of age. In 2008, the American Academy of Pediatrics released the 3rd edition of Bright Futures - Guidelines for Health Supervision of Infants, Children and Adolescents, American Academy of Pediatrics. These recommendations are endorsed by the Department of Health and Human Services Maternal and Child Health Bureau and materials include physician recommendations along with a schedule (http://brightfutures.aap.org/pdfs/Guidelines_PDF/20-Appendices_PeriodicitySchedule.pdf) for preventative pediatric health care.

A number of developmental screening tools are available to providers. Some tools are specific to a particular condition, like autism, while others are more general. The Centers for Disease Control and Prevention compiled this list of various screening tools (http://www.medicalhomeinfo.org/screening/DPIP/screeningtoolgrid.pdf).

Under the guidance of physicians and other providers, parents can also inform the screening process by providing key information about their children:

- Parent’s Evaluation of Developmental Status (PEDS)  
  http://www.pedtest.com/

- Ages & Stages Questionnaires (ASQ)  
  http://www.agesandstages.com/asq/download.html

The Assuring Better Child Health and Development (ABCD) Program, sponsored by the Commonwealth Fund and administered by the National Academy for State Health Policy (NASHP), assists states in improving the delivery of early child development services for low-income children and their families.
Participants in NCSL's meeting identified medical homes as a key way for states to address early childhood development. Medical homes have recently come into the spotlight as a way to provide comprehensive health care for children. Medical homes do not provide housing; rather it is a conceptual model for pediatric care that originates in a primary care setting. The AAP defines a medical home as family centered, coordinated, compassionate, comprehensive, culturally effective, accessible and continuous as a model of pediatric care.

As states increase the reach and effectiveness of health information technology (HIT) systems, they become an increasingly valuable tool for collaboration among private and public systems and among different agencies when coordinating screenings and treatment around early childhood development. HIT systems can also improve the likelihood that children will receive necessary follow-up treatment in medically underserved, rural, or frontier areas.

CONCLUSION

Legislators do not need to become experts in the field in order to promote early childhood development. Policymakers can direct attention to these issues through the bully pulpit and ensure that clinical and consumer expertise is an integral part of the development of state responses and strategies. Legislators can have a direct effect on the screenings and treatments that the millions of children enrolled in state Medicaid and SCHIP programs receive by ensuring that these programs provide the services the state requires. Policymakers can also help by encouraging the establishment of medical homes; supporting clinical practitioners; fostering coordination between public and private practices and the variety of safety net agencies responsible for early childhood development; and encouraging the use of health information technology to support the work of those in the early childhood development fields.
ADDITIONAL RESOURCES

- NCSL’s Newborn Genetic and Metabolic Disease Screening webpage
  http://www.ncsl.org/programs/health/genetics/newborn.htm
- NCSL’s State Genetics Employment Laws webpage
  http://www.ncsl.org/programs/health/genetics/ndiscrim.htm
- NCSL’s Genetics and Health Insurance State Anti-Discrimination Laws
  http://www.ncsl.org/programs/health/genetics/ndishlth.htm
- NCSL’s Medical Homes webpage
  http://www.ncsl.org/programs/health/medhom.htm
- March of Dimes
  http://www.marchofdimes.com/
  Peristats
  http://www.marchofdimes.com/Peristats/default.aspx
- Bright Futures
  http://brightfutures.aap.org/web/
- National Institutes of Health
  http://www.nih.gov/
- Eunice Kennedy Shriver National Institute of Child Health and Human Development
  http://www.nichd.nih.gov/
- National Human Genome Research Institute
  http://www.genome.gov/
- Office of Rare Diseases
  http://rarediseases.info.nih.gov/
- Agency for Healthcare Research and Quality (AHRQ)
  http://www.ahrq.gov/
- Centers for Disease Control and Prevention
  http://www.cdc.gov/ncbdd/
- Genetic Alliance
  http://www.geneticalliance.org/
- Health Resources and Services Administration
  http://www.hrsa.gov/
- American College of Medical Genetics
  http://www.acmg.net//AM/Template.cfm?Section=Home3
- National Newborn Screening and Genetics Resource Center
  http://genes-r-us.uthscsa.edu/
- Commonwealth Fund
  http://www.cmwf.org/
- National Academy for State Health Policy
  http://www.nashp.org/
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Sec. 503 of the Labor-HHS appropriations legislation:

(a) No part of any appropriation contained in this Act shall be used, other than for normal and recognized executive-legislative relationships, for publicity or propaganda purposes, for the preparation, distribution, or use of any kit, pamphlet, booklet, publication, radio, television, or video presentation designed to support or defeat legislation pending before the Congress or any State legislature, except in presentation to the Congress or any State legislature itself.

(b) No part of any appropriation contained in this Act shall be used to pay the salary or expenses of any grant or contract recipient, or agent acting for such recipient, related to any activity designed to influence legislation or appropriations pending before the Congress or any State legislature.

The National Conference of State Legislatures Staff Affairs Division responsible for this meeting is strictly prohibited from lobbying the state or federal government or taking a position on state and federal policy issues. The purpose in the meeting is solely to educate state legislators and staff; provide a forum to exchange ideas; and share expertise.