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COMMENTARY

Epidemiology as a Guardian of Children's Health: Translating Birth Defects Research into Policy

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INTRODUCTION

The intentional coupling of epidemiology with the practical implications of policy has been a much desired, but often elusive, goal. At times, information provided by sound epidemiological research has gone unnoticed; the voices of celebrities, public opinion, or industries have held sway in health policy development. This lost influence of epidemiology can result in inaction, wasted resources and/or failure to develop effective health initiatives. It can even harm those whom public health was meant to protect. However, when solid epidemiological research has been provided to, understood, used and implemented by policy makers, the efforts of epidemiologists and policy makers can result in truly effective health initiatives. Examples abound of such successful epidemiologically based policy initiatives related to children's health, in particular children with special health care needs like birth defects. Some of the most successful examples include folic acid dietary supplements during pregnancy for prevention of neural tube defects, newborn testing for phenylketonuria and dietary management, and recommendations for the limitations on alcohol ingestion during pregnancy for the prevention of fetal alcohol syndrome.

Despite epidemiological and medical advances in the causes and treatment of children with birth defects and despite an increase in health policy initiatives, additional work must be done to improve the health of the nation's children with special health care needs like birth defects. A better understanding and increased knowledge of the epidemiology of children with birth defects is a high priority due to the maternal and child health indicators in *Healthy People 2010* (1). It is also important for needs assessment, program planning and development, service delivery, and program evaluation for Medicaid and the State Children's Health Insurance Program and other State Title V Maternal and Child Health Block Grant related activities for this population (2). This is particularly salient issue given the current climate of health care reform and the need to assure children with special health care needs like birth defects have health insurance coverage. The purpose of this commentary is to describe some past and current research and policy initiatives regarding children with birth defects. We also explain the role epidemiologists can play in translating birth defects research into policy.

EPIDEMIOLOGICAL PROFILE OF CHILDREN WITH SPECIAL HEALTH CARE NEEDS

The Maternal and Child Health Bureau (MCHB) defines children with special health care needs as children who have or are at an elevated risk for a chronic developmental, physical, emotional, or behavioral condition and who additionally need health and related services of a form or quantity beyond which is required by children in general (3). Using the MCHB definition, in the United States, the prevalence rate of children with special health care needs ranges from 12%–19% (4).

Children with birth defects represent a subset of children with special health care needs due to their increased risk of developmental and chronic conditions. The March of Dimes defines a birth defect as an abnormality of function, structure, or body metabolism that is present at birth and results in mental and/or physical disability (5). Annually, birth defects affect about eight million children worldwide, which is about 6% of births worldwide (6). In the United States, 120,000 infants each year are born with a birth defect, affecting about 1 in 33 infants (5). Prevalence rates for children with special needs, including those with birth defects, can vary by a variety of factors such as: geographical residential location; maternal age, race/ethnicity, and prenatal exposures; and child's condition, age, sex, and race/ethnicity (4, 5).

Birth defects remain the leading causes of infant deaths in the United States (7, 8), accounting for over 20% of all infant deaths. Birth defects can impair the development of speech, hearing, and feeding capabilities and often impair psychomotor and cognitive skills, thereby creating physical and emotional stress for children and their families. Such problems can result in significant direct and indirect costs. Direct costs include costs to the health system itself and indirect costs include costs related to mortality, morbidity,

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Selected Abbreviations and Acronyms

CDC = Centers for Disease Control and Prevention
MCHB = Maternal and Child Health Bureau
NBPDS = National Birth Defects Prevention Study

and disability and out of pocket expenses. Birth defects account for more than a third of all pediatric hospitalizations. Hospital costs attributable to birth defects are more than $2.6 million annually in the United States (9). To date, we know genetic, environmental, maternal illness, or a combination of these factors can cause birth defects; however, about 60%–70% of causes of birth defects are unknown (5). Because of the high prevalence and costs of birth defects, understanding the causes of various birth defects is an important public health goal.

EPIDEMIOLOGY AND POLICY: CHILDREN WITH SPECIAL HEALTH CARE NEEDS

Successful Policy Initiatives

Specific research initiatives in the epidemiology of children's health have resulted in significant improvements in longevity and quality of life among children with special needs like children with birth defects. For over 40 years in the United States, established newborn screening programs for phenylketonuria and other devastating inborn errors of metabolism and for congenital hypothyroidism have potentially helped protect an estimated several thousands of infants and children from developing severe mental retardation due to early identification and treatment of these conditions (10). Public concern over a link between vaccinations and autism has caused many parents to refuse vaccinations for their infants and children. It has already resulted in decreased vaccination rates and the resurgence of preventable communicable diseases, disability and death in children. This link has been clearly refuted by epidemiological research and reported by the Institute of Medicine (11). This confusion also may have the unintended effect of diverting appropriate research and resources away from autism research, early identification and treatment.

Recommendations for folic acid fortification of the diets of pregnant women have resulted in an estimated 70%–80% decrease in the prevalence of spina bifida, a type of neural tube defect. Using birth defects surveillance data, neural tube defect trends significantly decreased in North America, Australia, and Europe in 2003 due to implementation of these recommendations (12).

Other sound epidemiological studies on children have noted the association of prenatal alcohol exposure and the increased risk of fetal alcohol spectrum disorders. Warning labels on alcohol containers now indicate that there is no established safe amount of alcohol consumption during pregnancy (13).

Current Initiatives in Epidemiology of Children's Health

An impressive number of government organizations sponsor research on children's health issues. The Centers for Disease Control and Prevention (CDC) sponsor the National Birth Defects Prevention Study (NBDPS) (14), the MCHB manages the National Survey of Children with Special Health Care Needs (15), and the National Institutes of Health conducts the National Children's Study (16, 17). The initial planning and development of the National Children's Study began about 8 years ago and is led by many partners and collaborators. These include the United States Environmental Protection Agency, the United States Department of Health and Human Services, the Eunice Kennedy Shriver National Institute of Child Health and Human Development and the National Institute of Environmental Health Sciences of the National Institutes of Health (16, 17). The March of Dimes focuses on birth defects and prematurity to include a broad environmental approach to children's health. Other organizations are interested in promoting children's health, particularly children with birth defects, among them are the National Birth Defects Prevention Network World Health Organization, the American Academy of Pediatrics, and specialty organizations devoted to specific birth defects. Each of these organizations depends on ongoing epidemiological research. These studies, organizations, and initiatives speak to pressing, unresolved health care needs of the nation's children.

Current Research Initiatives for Children with Birth Defects

The CDC has promoted several public health research initiatives for children with birth defects. These have included identifying priority research areas for children with specific birth defects like Down syndrome (18) and orofacial clefts (i.e., cleft lip with and without cleft palate) (19). The CDC also funds the NBDPS (14).

The National Center on Birth Defects and Developmental Disabilities at the CDC convened a workshop in 2006 to review current research, to identify gaps in knowledge, and to prioritize a public health research agenda for orofacial clefts (19). This workshop identified several areas of epidemiological research in orofacial clefts that warranted funding and additional research: phenotype characterization to better define etiologically homogeneous categories of orofacial clefts; effects of nutrition and nutritional supplements on the risk of orofacial clefts; early screening methodology to identify learning outcomes; social marketing campaign and smoking cessation; long-term outcomes;