Evidence-Based Practice in Spina Bifida:

Developing a Research Agenda

May 9–10, 2003

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Centers for Disease Control and Prevention

National Institute of Child Health & Human Development

National Center for Medical Rehabilitation Research

Interagency Committee on Disability Research

Agency for Healthcare Research and Quality

National Institutes of Health, Office of Rare Diseases

Spina Bifida Association of America

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Edited by Gregory S. Liptak, MD, MPH

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Introduction

Gregory S. Liptak, MD, MPH
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Spina bifida (meningomyelocele) is the most complex birth defect compatible with life. As a result, treatments and interventions to optimize the development and function of individuals who have spina bifida are complex as well. As care has improved, the number of individuals with spina bifida surviving into adulthood has increased. Yet, the consensus has been that most of the research on which current treatments are based has been done in less than optimal ways. Further, very few studies have been published regarding care for adults. Thus, the evidence base for current care is shaky at best.

For several years, the Spina Bifida Association of America (SBAA) and the Spina Bifida Foundation have tried to improve the evidence-base for spina bifida. In June 2002, the Professional Advisory Council (PAC) of the SBAA met in Orlando, Florida, to develop a prioritized list of topics that they felt needed to be reviewed and discussed to determine the soundness of current recommendations and the need for future research. After determining the topics for review, the PAC appointed experts in the field to perform critical reviews of the topics from the current literature. The International Classification of Functioning, Disability and Health (ICF) developed by the World Health Organization (http://www3.who.int/icf/icftemplate.cfm) was chosen as the framework for the outcomes addressed by the scientific articles. In a similar fashion, the levels of evidence adapted from the Centre for Evidence-Based Medicine (www.cebm.net/levels_of_evidence.asp) were chosen as criteria for evaluating the quality of the published articles.

Funding for a conference was provided by the Agency for Healthcare Research & Quality, the National Center on Birth Defects and Developmental Disabilities at the Centers for Disease Control and Prevention, NIH's Office of Rare Diseases, the National Institute of Child Health and Human Development, and the Department of

Education's National Institute on Disability and Rehabilitation Research. Participants for the conference which was entitled, "Evidence-Based Practice in Spina Bifida: Developing a Research Agenda," were chosen by the funders and by SBAA. The goal of the meeting was to develop a national research agenda for spina bifida.

During the conference, reviewers were given 15 minutes to present their critical survey of the literature. After several topics were presented, small group discussions that lasted one hour each were held. These sessions were designed to identify gaps and studies needed to fill gaps and to discuss priorities for future research. General discussions with the entire group were held to review these priorities. Many of the review articles will be submitted for publication in refereed scientific journals. Others will be posted on the SBAA web site. This compendium is a review of the findings of the conference and includes recommendations for future research.

Note: in most instances, the terms spina bifida, meningomyelocele and myelomeningocele are used interchangeably in this document.

Welcoming Remarks, May 9, 2003 Centers for Disease Control and Prevention

José Cordero, MD, MPH
Director of the National Center on Birth Defects and Developmental Disabilities
Centers for Disease Control and Prevention, Atlanta, Georgia

On behalf of our director, Dr. Julie Gerberding, and our center, I would like to welcome you to this important conference. We are here as the result of a successful, year-long, collaborative effort. We are grateful for the hard work of all our partner agencies: the Agency for Healthcare Research & Quality, NIH's Office of Rare Diseases, the National Institute of Child Health and Development, and the Department of Education's National Institute for Disability and Rehabilitation Research.

We are also grateful to you…each one of you…who has taken your time to give to this conference. Thank you for the work you will be doing in the next two days, and for the effort you have already put forth by preparing and reading the review papers. That is a task in and of itself.

It is wonderful to see this unification. We are excited by the triumphs that will come from continued work with our partners, our fellow governmental agencies, the Spina Bifida Association of America, universities, and health care providers. We will change the lives of people — present and future, who are living with spina bifida — as well as the lives of their families.

We have seen real progress toward our mutual goals: We have worked hard to educate the public about the importance of folic acid for women during their reproductive years. We have just analyzed data that shows blood folate levels are increasing and fewer children are being born with spina bifida and anencephaly. We have worked with the

Spina Bifida Association of America for many years. Currently, we are joining forces to increase the number of women taking folic acid who have already had a child born with spina bifida.

And, in Fiscal Year 2003, Congress gave our center 2.0 million dollars to begin the National Spina Bifida Program. This program will enable us to take the recommendations of this conference and turn them into action. We will be funding specific research projects on spina bifida through university affiliates of the Association of University Centers on Disability (AUCD). We are developing an outreach program through the American Academy of Pediatrics on optimal management of secondary and related conditions. We are also working on a new physical conditioning module for individuals with spina bifida. Additionally, we are improving our data collection, and the dissemination of information, about secondary conditions including physical, functional, social, psychosocial, and financial. The National Spina Bifida Program is just one example of our mission to promote the health and wellness of individuals living with spina bifida.

Bringing together these diverse talents, and the efforts of our various organizations, can reap exciting and hope-giving benefits. Our efforts here…in these two days…can bring hope and help to the world's children, and their families, for generations to come.

Agency for Healthcare Research and Quality

Denise Dougherty, PhD
Senior Advisor, Child Health
Office of Priority Populations Research
Agency for Healthcare Research and Quality
U.S. Department of Health and Human Services

On behalf of the Agency for Healthcare Research and Quality (AHRQ) and our Director, Dr. Carolyn Clancy, I am delighted to welcome you all to this landmark meeting.

AHRQ was excited to help support this meeting because it fits so well with AHRQ in two specific areas:

People with disabilities are one of AHRQ's priority populations, as directed by the Congressional law that reauthorized AHRQ in December 1999.[Agency for Healthcare Research and Quality, #339]

AHRQ has as its goals improving the quality, outcomes, and cost/use/access of health care for all Americans. Knowing the evidence base for prevention and treatment of secondary conditions among people with spina bifida is a necessary first step to making health care more responsive.

AHRQ previously held a series of meetings designed to develop a research and translation agenda relevant to people with disabilities.[Clancy, 2002 #336] [Perrin, 2002 #337] AHRQ's new policy on the inclusion of priority populations, including people with disabilities, is making it clear that AHRQ seeks applications to improve health care for this population. [Agency for Healthcare Research and Quality, #338] We are looking forward to participating in this meeting to learn more about what AHRQ and its partners might do to make life better for people with disabilities.

National Institutes of Health

Duane Alexander, MD
Director, NICHD, NIH

The National Institute of Child Health and Human Development is pleased to join in welcoming you and to be a co-sponsor of this conference. The Institute has long held an interest in spina bifida. One of our major areas of research is birth defects, and as one of the most common and most serious of these, much attention has been focused on spina bifida. We have developed and made available to the scientific community for study animal models of this condition, and supported studies of the neurodevelopmental and genetic origins of the anomalies. Our epidemiologists, in studies in Ireland where the prevalence of spina bifida is particularly high, have identified a specific gene defect that predisposes women to bear children with spina bifida, especially if their diets are low in folate.

We also support research on rehabilitation for persons with spina bifida who, in common with persons with spinal cord injury, have problems with mobility, bowel and bladder function and infection, and sexual function.

Our major current research is a clinical trial evaluating the effectiveness, along with the safety for mother and fetus, of fetal surgery to close the neural tube defect *in utero* in the second trimester of pregnancy. Animal studies and some clinical experience in humans suggest better preservation of motor function below the lesion and reduced incidence of hydrocephalus requiring a shunt if the spinal defect is closed during pregnancy. This randomized trial, involving 200 pregnancies, will provide the answer as to whether there are benefits to prenatal closure that outweigh the risk and trauma.

Much progress has been made from research on spina bifida, and NICHD intends to continue its broad and strong research in this field. We wish you a successful conference, and eagerly anticipate your recommendations.

National Institute on Disability and Rehabilitation Research

Theresa B. San Agustin, MD
Program Officer, National Institute on Disability and Rehabilitation Research
U.S. Department of Education

Good morning. I am representing Steven James Tingus, Director of the National Institute on Disability and Rehabilitation Research (NIDRR) in the U.S. Department of Education. NIDRR is pleased to be a sponsor of this important gathering. This workshop fits well into the mission of NIDRR's long range plan: "To generate, disseminate, and promote the use of knowledge that will improve the ability of disabled individuals to perform regular activities in the community, and increase the capacity of society to provide full opportunities and supports for participation."

NIDRR is strongly committed to President Bush's New Freedom Initiative which drives current federal policy supporting persons with disabilities. The goals of NFI are to increase access to assistive and universal-designed technology, expand educational opportunities, integrate Americans with disabilities into the workforce, and promote full access to community life. We are pleased that the topics focusing on spina bifida during this conference contribute to those goals.

NIDRR has two general mechanisms for funding research related to spina bifida: field-initiated projects and fellowships. Field-initiated projects are initiated by investigators focused in a specific research area, such as spina bifida. They are funded for $150,000 per year for three years. Decisions for funding are made by standing panels. NIDRR Fellowships are awarded to individuals from field-initiated topics for 12 months. Levels of award are based on the experience of the applicant.

In the past 15 years, NIDRR has funded 19 projects specifically related to spina bifida. Two recent projects include a study of neuro-

psychological functioning and psychosocial adjustment in adolescents with spina bifida and an evaluation of a new material and method for custom-fitting modular ankle-foot orthoses. NIDRR and Mr. Tingus look forward to the research agenda generated from this meeting, and support the great work being done on behalf of those with spina bifida and their families. Thank you for participating in this effort.

To find information about NIDRR grants please access the *Federal Register* at http://www.ed.gov/funding.html or contact Donna Nangle at NIDRR Donna.Nangle@ed.gov.

Summary of Findings

Gregory S. Liptak, MD, MPH, Professor of Pediatrics, University of Rochester Medical Center

Current Studies

As suspected prior to the formal reviews, much of the clinical care provided to individuals who have spina bifida is based on studies that have used weaker designs. Some of the care provided to children and much of the care provided to adults is based on expert opinion (Evidence Level 5). Much of the care provided to children is based on case series (a series of individuals, usually selected from a single spina bifida program studied without the use of a control group) (Evidence Level 4). Very few studies that qualify as level 1, e.g., randomized control trials or representative cohort studies, have been performed in any topic on individuals with spina bifida. Also, most studies have concentrated on the ICF category of Body Functions and Structures, but not on Activities or Participation.

Research Priorities

The following table illustrates the top two or three priorities for each of the topic areas discussed during the conference.

Topic	Priorities
Neurosurgery	Optimal assessment and treatment of hydrocephalus
	The role of tethered cord release in long-term management
	The long term neurological priorities, challenges, complications, and treatments for adults
Neuropsychology and Learning	Evaluation of the core processing deficits associated with neuro-anatomical abnormalities
	Determination of the earliest indicators of learning difficulties
	Identification of the instructional and developmental interventions that are most effective in facilitating learning

Topic	Priorities
Urology	Optimal proactive therapy of urinary function in individuals who are at risk for renal deterioration Evaluation of neuromodulation of the bladder Interventions using tissue engineering
Sexuality	Natural history of sexual maturation
	Range of sexual functional abilities
	Optimizing psycho-sexual development
Self Care	Factors that affect the teaching and learning of self care Assessment of ways to measure self care Optimizing the use of assistive devices
Family Functioning	Evaluation of family resilience and family dysfunction
	Optimizing parenting
Socialization	Determination of the prevalence and nature of social challenges Determination of risk factors for impaired socialization Optimizing socialization
Independence	Assessment of current functioning of adults
	Secondary conditions in adolescents and adults
	Optimizing self-determination and independence
Education and Employment	Factors that predict performance in school Participation in the labor force Models of transition from school to work
Behavioral Health & Mental Health	Developmental trajectories of mental health/behavioral health issues
	Optimizing mental health
Reproductive Issues for Women with Spina Bifida	Optimal management of preconceptual issues, e.g., precocious puberty, birth control, and anatomical changes Optimizing outcomes of pregnancy (in women with spina bifida) Optimizing preventive care issues like osteoporosis, breast and gynecologic cancer, and heart disease
Orthopedics	Optimal management of hip dislocation
	Prevention and management of osteoporosis
	Optimal management of kyphosis and scoliosis
Mobility	Evaluation of an upright mobility program Optimizing mobility changes during adolescence
Gastroenterology and Nutrition	Optimizing bowel management
	Optimizing nutrition
Integument	Pathophysiologic factors that affect skin breakdown Optimizing preventive skin care Optimal treatment of skin breakdown
Latex Allergies	Latex sensitization and allergy in babies and young children
	Latex allergy in adults
	Optimal preventive management

Implications

Many of the recommendations suggested the following three-step approach: (1) Establish the prevalence and nature of a particular issue or problem, (2) Identify risk factors that increase the likelihood of an adverse outcome, and (3) Develop and test an intervention strategy. For future research to be valid (i.e., methodologically sound) and generalizable to other individuals with spina bifida, studies should utilize samples that are representative of the population in general and large enough to be statistically sound. This typically entails collaboration among several centers. Longitudinal studies were also recommended often.

One strategy that has been used successfully with conditions such as childhood cancer is the use of integrated research networks, i.e., groups of institutions that work with each other to develop collaborative research projects. Funding organizations could support this type of collaboration by paying for meetings involving researchers from across the country in order to develop and plan collaborative projects. Lessons from the highly successful research program in immunizations include the following: (a) Need for strong scientific base and rationale, (b) Quantification of disease burden and target group, (c) Critical role of "champions," (d) Availability of models (e.g., theoretical models for social science research), and basic science counterparts (e.g., for urological research), (e) Meticulously executed clinical trials, and (f) Measurements of public health impact [National Vaccine Advisory Committee, Pediatrics, October 1999].

The task now is to use the hard work of everyone involved in this effort to develop future research to fulfill Dr. Cordero's dream of "exciting and hope-giving benefits" to all individuals who have spina bifida.

Neurosurgery

Rationale

Spina bifida (meningomyelocele, occult spinal dysraphism, and other variants) is primarily a neurological condition. The disorder results from failure of the tissues that will become the brain and spinal cord to develop normally during very early development. Brain abnormalities may include the Chiari type II malformation and hydrocephalus, particularly in those with myelomeningocele. Other abnormalities of the spinal cord and brainstem such as syrinx (accumulation of spinal fluid within the spinal cord) and split cord can cause symptoms. Tethering (abnormal restraint) of the spinal cord can occur in individuals and cause deterioration.

Abnormalities of the spinal cord lead to signs and symptoms such as paralysis and weakness of the legs, loss of feeling, and loss of normal bowel and bladder control. Abnormalities in the brain can lead to disorders of breathing, difficulty swallowing, and nonverbal learning disorders. The hydrocephalus itself requires surgical treatment and is associated with problems such as precocious puberty. Ventricular shunt failure is a common occurrence requiring neurosurgical intervention. Tethering of the spinal cord can lead to progressive neurological deterioration such as weakness or loss of function.

Summary of Current Information

The following topics regarding the management of children and adults with spina bifida were reviewed: (1) hydrocephalus; (2) Chiari malformation; (3) tethered spinal cord; (4) syringomyelia; (5) neurosurgical causes of scoliosis; (6) spinal cord tethering in adults; and (7) use of adjunctive tests to predict neurological deterioration in patients with spina bifida. Although fetal surgery is a critical topic, it was not reviewed, given the ongoing randomized controlled trial currently underway, which should answer some of the questions related to this issue.

Hydrocephalus

No standard protocol for the management of hydrocephalus currently exists. Guidelines for ongoing care include regular follow-up in a

dedicated multidisiplinary clinic. Surveillance imaging studies of the brain are performed, although the interval of examination remains indeterminate. Several options for the first ventricular shunt include: (a) initial surgery for hydrocephalus at the time of closure of the myelomeningocele if the hydrocephalus is severe (including ventriculoperitoneal and ventriculo-subgaleal shunts), (b) shunt surgery later depending upon clinical circumstances, and (c) expectant monitoring without surgery for the first 5 months of life for moderate stable ventricular enlargement. Endoscopic third ventriculostomy remains an option in the management of these patients based upon the strength of several relevant articles, although the exact success rate remains unknown. Whenever evaluating any deterioration in the patient with spina bifida, it is always necessary to consider shunt malfunction before undertaking other forms of neurosurgical treatment.

Chiari Malformation

Chiari type II malformation remains an important cause of death and disability for patients with spina bifida. Most individuals present in infancy, and the outcomes are dependent upon both age and presenting symptoms. The results of early surgery for symptomatic Chiari malformations are mixed, with some studies suggesting a benefit, and others failing to find a benefit to early and aggressive surgery. Shunt malfunction produces signs or symptoms referable to the Chiari malformation, and up to 20% of shunt malfunctions may be missed if determination of shunt malfunction is based solely upon enlargement of ventricular size. The ideal surgical technique has not been explored in a systematic manner. Critical questions related to Chiari malformation include the following: What patients should be selected for surgery? What operation is most effective? What are the results and adverse effects of surgery? What clinical or radiographic parameters may predict the need for intervention in this population?

Tethered Spinal Cord

Spinal cord tethering is universally present on imaging studies in individuals who have meningomyelocele but is symptomatic in only 11–27%. Common presenting features include back or leg pain, reduced sensory or motor function in the legs, deteriorating gait, increasing spas-

ticity (stiffness) in the legs, orthopedic deformities of the legs, change in bowel or bladder habits, and scoliosis. Most symptoms are either improved or stabilized, and few deteriorate after untethering. Because virtually all individuals who have tethered spinal cord will have an abnormal MRI scan, the decision to perform surgery on an individual to untether the spinal cord continues to be based on clinical findings with the help of adjunctive studies such as cystometrogram or manual muscle testing. No single technique for the original surgical closure of the meningomyelocele at the time of birth has been identified that will completely prevent tethering. Some children will require more than one untethering surgery. However, factors that identify that group have not been identified.

Tethering in Adults

Very few papers dealing with this topic have been published. Based on the available studies, the presenting features of adult tethered cord syndrome are similar to those seen in childhood. No natural history study has yet been performed to evaluate the frequency with which deterioration from tethering occurs in adults with spina bifida. The success rate of untethering is uncertain. The data upon which to recommend early aggressive spinal cord untethering are mixed and methodologically flawed and further studies are needed.

Syringomyelia

Syringomyelia (accumulation of spinal fluid within the spinal cord) is visible on imaging studies in 11–77% of persons with spina bifida. Its presence frequently appears to be coincidental and not associated with significant symptoms; the frequency of symptomatic syringomyelia is 1.8–5%. However, the signs and symptoms of syringomyelia overlap with those of the Chiari malformation, tethering, and even shunt malfunction. Moreover, no long-term studies of syringomyelia have been published that would help determine in which patients syringomyelia might become symptomatic, what features might predict symptoms, or whether or when treatment may improve symptoms or signs due to syringomyelia. Little data exist on what treatments work better than others. Long-term outcome studies are clearly needed to understand the

natural history of syringomyelia, better define the contribution of syringomyelia to deterioration, and evaluate various treatment options.

Neurosurgical Causes of Scoliosis

Uncertainty exists whether or not Chiari malformation, syringomyelia, and/or spinal cord tethering can be causes of isolated scoliosis in the person with spina bifida. The incidence of scoliosis appears to be most dependent upon patient age and sensorimotor level; progression of scoliosis, on the other hand, is most dependent upon the severity of the initial curve (direct relationship), patient age (inverse relationship), and ambulation status. Studies examining the contribution of tethering to scoliosis are few and have significant methodological flaws that make analysis very difficult. No data suggest that Chiari malformation or syringomyelia contribute to scoliosis in patients with spina bifida regardless of level. No data suggest any correlation between signs or symptoms of tethering, Chiari malformation, or syringomyelia and scoliosis.

Reliability of Adjunctive Tests of Neurological Functioning

Adjunctive tests include ultrasonographic, MRI, and CT scans of the neuraxis; electrophysiological testing such as somatosensory evoked potentials (SSEP); urodynamic testing; manual muscle testing; and gait analysis. The available data do not shed much light on the efficacy and reliability of these tests. The use of regular surveillance CT scans in individuals without clinical signs and symptoms does not identify shunt malfunction frequently enough to warrant its routine use. Neither spinal ultrasound nor dynamic MRI can reliably correlate the radiographic imaging of tethering with clinical symptoms. Two studies of SSEPs failed to find any predictive value for neurological deterioration. Although urodynamic testing has been recommended as an adjunctive test for deterioration in patients with spina bifida, no published study to date has followed patients serially and in a systematic fashion. Several studies have identified improvements in urodynamics postoperatively in patients undergoing tethered cord release. Manual muscle testing is another adjunct that is used frequently in many centers to identify neurological deterioration but its reliability has yet to be established. Finally,

gait analysis is a promising tool that may identify and/or quantify deterioration, but it has not yet been studied in any detail in this population.

Specific Recommendations

The following were identified as research priorities:

1. Assessment and treatment of hydrocephalus in patients with spina bifida

What is the best practice for assessing the patency and monitoring shunt malfunction in patients with shunted hydrocephalus? What are the long-term outcomes for patients with known disconnected or otherwise malfunctioning shunts who are currently asymptomatic? Can shunt independence reliably be established in a subset of these individuals? At what point does ventricular enlargement have an impact on neuropsychological growth and development, and at what point should shunting be considered for the presently asymptomatic patient with stable asymptomatic moderate ventricular enlargement? Do patients with stable unshunted ventricular enlargement deteriorate later in life? Are they predisposed to adverse outcomes in terms of neuropsychological assessments, quality of life issues, and long-term complications? Finally, what is the role for endoscopic third ventriculostomy — What are the patency rates, long-term outcomes, complications, and failure rates for this procedure, and for which patients should it be considered?

2. The role of tethered cord release in the long-term management of patients with spina bifida

What are the long-term outcomes for patients undergoing tethered cord release? Which patients would most benefit from this type of surgery? What are the rates of rethering? Finally, funding should be directed toward improving our understanding of the pathophysiology, biochemistry, and molecular biology of spinal cord tethering, and investigating novel and creative approaches to minimizing or preventing spinal cord tethering, both after primary closure and after untethering procedures.

3. The long-term neurological priorities, challenges, complications, and treatments for adults with spina bifida

Adults are comprising an ever increasing proportion of the population of people with spina bifida. A cross-sectional, population-based

study of adults with spina bifida should be undertaken emphasizing long-term follow-up, complications, challenges, and neurological outcomes for this group. Outcome measures should use the World Health Organization model of disability and examine survival, cognitive and functional outcomes, quality of life issues, cost of care, and access to healthcare services in a coordinated fashion. Although not specific to neurosurgical issues, studies of various models of transition to adult healthcare services should be undertaken to find the best practice in caring for these individuals. Finally, training grants should be offered to help neurosurgeons whose practice involves adults to better understand the issues facing this population and to learn best care for them.

4. Identification of patients with neurological deterioration from hydrocephalus, shunt malfunction, tethering, Chiari, and syringomyelia

Reliable, objective, quantitative, and rigorously applied tools are required for assessing neurological function in this population — both to follow function serially, identify deterioration when it occurs, and assess the impact of various neurological procedures in trying to improve or stabilize function. Adjunctive tests should be objective, sensitive, specific for deterioration from an identifiable neurological cause, validated objectively, have practical value, and correlate with functional outcomes. Research to identify novel ways to assess and follow neurological function should be sought.

5. The role of neurosurgical causes of scoliosis in people with spina bifida

How important are neurosurgical processes such as tethering, Chiari malformation, syringomyelia and hydrocephalus on the development and progression of scoliosis in this population? What, if any, are the root neurosurgical causes of scoliosis? How do treatments for these conditions impact the incidence and/or progression of scoliosis in this population?

Neuropsychology and Learning

Rationale

Although spina bifida is typically thought of as a disorder of the spine and spinal cord, it involves the entire nervous system, including the brain. Children with meningomyelocele often have abnormalities such as Chiari type II malformation, hypoplasia of the cranial nerve nuclei, thinning of the posterior cerebral cortex, and abnormal architecture of the corpus callosum and other midline structures. Most children (80%) with meningomyelocele have intelligence in the normal range, yet they often have specific learning disabilities. Typically they are stronger in language and weaker in perceptual and motor skills. Older research has documented that children with meningomyelocele have lower nonverbal than verbal IQ scores, with verbal IQ scores often in the average range. They have difficulty with pragmatic communication, e.g., they use and understand single words but have significant problems with language at the level of text and discourse. They demonstrate significant problems with short-term memory, especially list learning. They often appear to have problems with attention and executive function; however, evaluation of their memory, attention, and executive functions may reflect difficulties with other core processes. The neuropsychological challenges of meningomyelocele are present early in life, are comprehensive in nature, and are a significant impediment to academic learning, social competence, and adaptive functioning throughout the life span. Thus, they directly impact the long-term outcomes and level of independence of individuals with meningomyelocele.

In school, math is commonly impaired more than word recognition skills, and problems with writing and reading comprehension are common. Neurological impairments can affect learning outside of school as well in areas such as self-care, social skills, and adaptive behavior skills. The relationships among learning, achievement, motivation, and learning outcomes appear to be complex.

Summary of Current Information

Although numerous studies have been published evaluating cognitive function in children with meningomyelocele, they have methodological limitations. First, they have generally been conducted on school-

aged children, so that the origins of cognitive difficulties (in infants and preschoolers) and the long-term outcomes of compromised cognition (in young adults) are not understood. Second, these studies have not analyzed the range of cognitive outcomes in the same individuals, so the interrelations among domains are not adequately understood. Third, the considerable heterogeneity in cognitive profiles associated with variations in the neural phenotype, hydrocephalus and its treatment, and environmental factors have not been studied. Finally, it is likely that differences within and across neuropsychological and learning domains are due to impairment in a smaller set of core processes involving timing, attention, orientation and regulation, motor control, and visual perception. These core processes are strongly linked with the variable expressions of brain dysmorphology characteristic of meningomyelocele (cerebellum, midbrain, posterior cortex, and corpus callosum). The impact of brain dysmorphology on core processes is likely moderated by variable impacts of the early care giving environment of the child.

Little evidence exists beyond clinical and educational experience indicating how neuropsychological factors, physical disabilities, limits to accessibility, learned helplessness, and environmental factors all contribute to challenge active participation in the process of learning for individuals with meningomyelocele. Furthermore, little is known about what instructional techniques and developmental interventions are effective in promoting learning. Although extensive bodies of research are available in the fields of special education, developmental intervention, and assistive technology, few studies address the unique learning challenges of individuals with meningomyelocele.

Therefore, understanding about cognitive function, learning, and development in individuals with meningomyelocele is fragmented because research has been largely discipline-specific, with studies of genetics, development, brain imaging, and cognitive outcomes conducted in parallel rather than in an integrated fashion. Understanding is incomplete because very few studies have used recent developments in genetic analysis, structural and functional brain imaging, and in-depth analysis of cognitive functions. Understanding is also unidimensional, because the multiple factors that affect outcomes have not been studied using a multidimensional framework.

Specific Recommendations

A coordinated program of research will be necessary in order to further knowledge in this area. The high degree of individual variability necessitates a large sample size (>500 subjects) involving multiple sites. A transdisciplinary approach is required including neurological studies (e.g., neuroimaging, functional MRI, evoked potentials), genetic evaluations, neuropsychological evaluations, academic assessments, ratings of social and adaptive behavior, and family environment/parenting evaluations. Valid sampling techniques are needed to ensure representative participation across ethnicity and social status with appropriate matched comparison groups. The inclusion of longitudinal data is essential to understanding the development of neuropsychological challenges.

Neuropsychology

1. The first research study recommended is an evaluation of the core processing deficits associated with the neuroanatomical abnormalities of meningomyelocele. Core processing deficits include timing, attention orientation and regulation, motor control, and visual perception. Despite patterns that individuals with meningomyelocele share in common, a great deal of individual variability occurs in individuals. These difficulties emerge early in development, can be modified by experience, persist across the life span, and are related to genetic factors, level of the lesion, and neurological abnormalities. The relationship between anomalies in brain structure and the core processing deficits, as well as the relationship between core processing deficits and functional outcomes over the course of development, are not well understood now. Also, the neuropsychological development across the first three years of life for children with meningomyelocele is not well known.
2. The second study would examine the ways in which core processing deficits impact on adaptive behavior, social competence, and academic learning throughout the life span. For example, although individuals with meningomyelocele are perceived as having problems with emotional development, motivation, and perceptions of self and the environment, it is not clear how the core processing deficits affect these aspects of functioning and participation.

3. The third study would evaluate the environmental factors that can interact with core processing deficits to affect adaptive behavior, social competence, and academic learning throughout the life span. Can these environmental factors either potentiate or ameliorate the functional effects of the core processing deficits? Can specific environmental interventions be identified that could prevent some of the cognitive dysfunction found in individuals with meningomyelocele? If so, what type of intervention would be most promising and when in the life span would that intervention be most likely to be effective?
4. The final study would be to take the information learned from Study 3 (above) to develop and test a specific intervention, whose goal would be to ameliorate the adverse cognitive and functional effects of the neurological changes found in individuals with meningomyelocele. For example, some evidence exists that reading disability is preventable in many children (without meningomyelocele) and that intensive interventions can be effective with older children who have severe reading difficulties.

Learning

1. The first study would be to determine the earliest indicators of learning difficulties and the ways in which children could be evaluated so that these challenges are identified and ameliorated. While early intervention is probably the most effective strategy, the learning challenges of meningomyelocele may present in subtle ways during early childhood such that parents and educators do not address them; or the child may not qualify for current Birth to Three (IDEA) or other special education services. Parents and school personnel need ways to identify children at risk of later learning challenges so that those at risk can receive preventive services.
2. The second study would be to identify the instructional and developmental interventions that are most effective in facilitating the learning of individuals with meningomyelocele. While it appears that identifying and intervening to ameliorate learning challenges during the preschool and elementary years is critical, it is not clear what early intervention strategies would be the most effective. For exam-

ple, are home-based models of intervention for parents and providers effective with the youngest children?

3. The third study would be to determine which models of early intervention are most effective with preschoolers with meningomyelocele and to identify the ways in which parents and specialists could best provide these services.
4. The fourth study would be to identify the most effective strategies for disseminating information about the learning challenges of meningomyelocele to parents and teachers. Parents are often not informed of the learning challenges of spina bifida and need education to ensure that they are informed and can advocate for their child during the preschool years and beyond. Many schools will see only a handful of students with spina bifida so they will not gain a familiarity with the types of learning challenges they present.

Urology

Rationale

The bladder and urethra are controlled by nerves that leave the spinal cord in the lower sacrum. Therefore, bladder dysfunction is present in virtually all individuals with meningomyelocele. Even individuals with sacral lesions and typical leg movement usually have urinary problems. In addition, individuals with meningomyelocele also have a higher incidence of malformations of the kidneys, including horseshoe kidney and absent kidney.

The bladder has two major functions: 1) to store urine, and 2) to empty the urine once the bladder is full. Problems with the bladder may lead to incontinence; failure to completely empty the bladder of urine can lead to infections of the bladder and/or kidneys. Over 90% of infants with spina bifida have normal renal function at birth. If unattended, however, about 50% will have renal deterioration. For example, the combination of a tight bladder outlet and increased tone in the bladder may cause urinary reflux and hydronephrosis and produce kidney damage over time, especially if urinary tract infections occur. Renal damage is the single most common cause of morbidity and mortality in individuals with meningomyelocele; death from chronic kidney failure still occurs in adults with meningomyelocele. The goals of urological care are to 1) maintain normal renal function, and 2) achieve social continence.

Summary of Current Information

Several data bases were reviewed and over 450 articles were assessed regarding the evidence for current urologic practice. Present urologic management is based primarily on evidence classified as level 4 and 5. Very few studies classified as level 1 or 2 are available to support current therapy.

Each individual with spina bifida has a distinct physiologic outcome. Bladder and ultimate renal function cannot be predicted by simply determining the location of the spinal lesion. Thus, urodynamic evaluations are used to determine bladder function. Urodynamics refers to a group of diagnostic procedures that are performed to evaluate bladder function. Parameters that can be measured include pressure, flow, and

electrical activity. From the available literature it appears that urodynamic testing is a reliable, reproducible method that can identify particular risk factors affecting both lower urinary tract (bladder) and upper urinary tract (kidney) function. Many urologists provide proactive therapy that is based on risk factors determined by urodynamic testing. While proactive therapy is appealing, no convincing evidence shows that it is more beneficial than the conventional treatment of upper tract abnormalities once they have occurred. The earliest form of proactive intervention in spina bifida would be closure of the spinal lesion in the fetus. Currently, preliminary reports do not support any urologic benefits from fetal back closure. This reinforces the need to continue to pursue other areas of early proactive therapy that may prevent the urologic sequelae of spina bifida.

When lower urinary tract abnormalities affect upper urinary tract function, intervention centers on both medical management, which includes intermittent catheterization and medications, and operative intervention, particularly augmentation cystoplasty (enlarging the bladder). Medical and surgical therapies have been shown to prevent further upper urinary tract damage, and in many cases improve upper urinary tract status. However, no evidence supports the benefits of one particular therapy over other treatments, particularly with regard to operative intervention and the use of various bowel segments in reconstructing the bladder. In addition, when urinary reconstruction is undertaken, there are particular complications, particularly the potential for bladder rupture, which may be more prevalent in individuals with spina bifida. Because of these various risk factors, research is being done to evaluate alternative forms of bladder reconstruction. The most promising appears to be tissue engineering, the development of laboratory-grown cells, tissues, or organs to replace or support the function of defective or injured body parts. No evidence-based studies support the use of tissue engineering in humans, but its success in animal models is encouraging.

Kidney damage is a direct result of abnormal innervation to the bladder, which results in hostility factors. In order to prevent these changes from occurring, neuromodulation, stimulating the nerves either outside or within the bladder, may be of benefit. Neuromodulation has been undertaken in a crude fashion in the past. While some trends have

been identified, no definite benefits from this intervention have been found. But this area remains appealing for future research.

Urinary continence is an important social function. Achieving urinary continence in individuals with meningomyelocele currently is achieved with a combination of interventions including intermittent catheterization, medications, and surgery such as urinary reconstruction of the bladder neck and augmentation cystoplasty. Most therapies to achieve continence require the use of intermittent catheterization. While this is beneficial, it is associated with an increased incidence of urinary infection. In addition, individuals who are unable to perform self catheterization may have limited independence.

As the population of individuals with spina bifida ages, interests shift to other areas, particularly related to quality of life and sexuality. Very little information exists that assesses erectile dysfunction and fertility in males with spina bifida. Further information in this area is greatly needed [see section on Sexuality].

Specific Recommendations

It was the consensus of the urologic panel that future research efforts in urologic management should include the following: (a) prevention of lower urinary tract dysfunction through basic science studies such as neuromodulation, (b) prevention of upper urinary tract changes through a multiinstitutional trial investigating proactive therapy, and (c) treatment of urologic problems associated with spina bifida particularly tissue engineering for bladder augmentation.

1. The first study recommended is a multiinstitutional randomized controlled trial evaluating proactive therapy of urinary function in individuals who are at risk for renal deterioration based on hostility factors determined through urodynamic testing. This study would determine whether proactive therapy is more beneficial than observational therapy. Questions that would be asked include: Does proactive therapy decrease the need for operative intervention in order to preserve renal function? Is continence improved with proactive therapy and can this also limit operative intervention? Is early intermittent catheterization readily accepted by families and, subsequently, children? Does earlier intervention with intermittent catheterization lead to a greater acceptance of self catheterization at

an earlier age? What is the relationship between renal function and social function, e.g., might measures that improve renal function worsen activities and participation?

a. However, two major methodological issue would have to be addressed before this study could be undertaken. The first is the evaluation of renal function. Currently, upper urinary tract deterioration is defined by radiographic images of hydronephrosis and vesicoureteral reflux. Yet, renal functional impairment can be objectively assessed through multiple modalities including nuclear imaging, e.g., DMSA renal scan, and assessment of the following: glomerular filtration rate, effective renal plasma flow, urinary concentrating ability, creatinine clearance and parathyroid hormone levels. It is unclear which combination of these tests is best to evaluate renal function. Therefore, research to evaluate these outcome measures should be undertaken to determine which combination is most indicative of physiologic and functional outcomes (i.e., the gold standard).

b. The second issue is the validity of urodynamic testing. A prospective trial would be necessary to validate the urodynamic assessment and screening of individuals. Factors related to testing that need to be evaluated include the appropriate rate and temperature of fill, the testing medium, the measurement of compliance, the significance of uninhibited contractions, voiding or leak point pressure, and the importance of a post void residual. The prognostic significance of current risk factors such as leak point pressure, detrusor sphincter dyssynergy, and compliance need to be further elucidated. Testing parameters such as sensitivity, specificity, positive and negative predictive values, and likelihood ratios for diagnostic tests used in individuals with meningomyelocele should be determined. Sensitivity analysis using ROC curves to evaluate the interpretation of urodynamic testing would be valuable as well.

2. One proactive approach to the prevention of neurologic dysfunction of the lower urinary tract would be through neuromodulation of the bladder either extravesically or intravesically. This would first require an animal model having spina bifida, which does not exist. Therefore, we support the development of an animal model for spina bifida that would allow evaluation of the effects of neuromodulation. These include the differences between intra- and extravesical stimulation, the best timing for stimulation, the duration of

stimulation, and evaluation of the cellular changes that occur in the bladder prior to and subsequent to stimulation.

3. Fetal surgical intervention for meningomyelocele has not yet shown any significant benefit for urologic function. We strongly support the current multicenter randomized controlled trial that is funded by the NIH. Consistency across sites in the evaluation of bladder function particularly as it relates to urodynamic testing is mandatory. In addition, it was felt that the use of EMG assessment of the external urinary sphincter in this study would provide the best objective measure of neurogenic dysfunction and denervation.
4. Tissue engineering has the potential to revolutionize operative management of urinary reconstruction. While there is no scientific human data, a study is currently ongoing. At present, the development of lower urinary tract hostility factors due to nerve innervation dysfunction cannot be prevented. Therefore, current therapies are designed to treat the dysfunction. Tissue engineered bladder tissue to be used for augmentation and bladder neck reconstruction including external urinary sphincter may dramatically improve care. While this area is exceedingly important, tissue engineering impacts multiple areas of urologic management. Thus, this research will likely obtain funding from many sources including the private sector. We strongly support the need for continuing research in the area of tissue engineering but could see funds earmarked towards urologic care of individuals with spina bifida better utilized in the area of proactive therapy.
5. As individuals with meningomyelocele age, quality of life issues related to sexuality and fertility become increasingly important. Factors that need to be evaluated include the number of males who have erectile dysfunction, the number who are interested in achieving satisfactory intercourse, and the number interested in fathering children. Once this data is accumulated, an assessment should be made regarding the direction therapeutic intervention should take [see Sexuality section].

Sexuality

Rationale

Humans are sexual beings. Sexuality, an issue that may entirely be overlooked in people with spina bifida, includes (a) puberty, (b) sexual functioning including fertility, and (c) psychosexual development. These three aspects of sexuality are related to each other and all can be affected by spina bifida. For example, many girls with meningomyelocele and hydrocephalus experience precocious puberty. Erectile dysfunction and retrograde ejaculation, which are common in adult men with spina bifida, and urinary and fecal incontinence, which are common in all adults with spina bifida, directly affect sexual function. Both males and females with spina bifida typically have decreased sensation in the perineum, which can impair the ability to experience orgasm. Nonverbal learning disorder, which affects social interactions and self-esteem, is frequent in spina bifida and affects psychosexual development. Societal attitudes towards people who have disabilities also affect self-esteem and sexual self-image.

Summary of Current Information

Relatively few studies specifically examining sexuality in individuals who have spina bifida have been published; therefore, the reliability of available information is not high. Precocious puberty occurs in 10–30% of girls with spina bifida and hydrocephalus; girls with precocious puberty have elevated sex hormone levels as well as growth retardation. The average age of menarche of girls with spina bifida is earlier than for other girls (10.2 v. 12.8 years). Treatment of precocious puberty with analogues of gonadotropin releasing hormones (e.g., leuprolide) can stop the progression of puberty, stop menses, and decrease sex hormone levels. However, predicted adult height has not been improved by this treatment. Boys rarely experience precocious puberty. They do, however, have a higher than expected incidence of undescended testicles (cryptorchidism) (15–25%).

Approximately 75% of males with spina bifida are able to have erections. These usually are achieved reflexively by stimulation rather than occurring by psychogenic means, and the ability to sustain these erections in order to have intercourse is uncertain. Generally, the lower the

level of lesion, the greater the likelihood of being able to have erections. Sildenafil (Viagra®) has been shown to improve erectile dysfunction in men with spina bifida. Retrograde ejaculation, where the semen goes into the bladder rather than egressing from the urethra, is common, but the exact frequency is unknown. Many men are infertile, but the frequency is uncertain. The technique of artificial insemination is promising but its future role in this population is unknown. More females than males report having had sexual intercourse, but it is uncertain how frequently orgasm occurs. Women with spina bifida are capable of becoming pregnant and should be aware of birth control. Genital and perineal skin breakdown, including penile and vaginal ulceration, can occur with intercourse because of decreased sensation; awareness of this should be part of sex education for people with spina bifida.

Adults with spina bifida are described as being more socially isolated than those without disabilities. This may decrease opportunities for sexual intimacy. Problems with understanding social cues related to nonverbal learning disabilities may make intimacy more difficult as well. Although sexual abuse of individuals (especially females) with spina bifida has been reported, the frequency is unknown.

Specific Recommendations

1. Puberty

What is the natural history of sexual maturation in individuals who have spina bifida?

Specific questions that should be answered include the following: (a) when do these children enter and complete puberty? (b) How often does precocious puberty occur and what are its consequences? (c) what factors predict precocious puberty? (d) what are the consequences (both positive and negative) of gonadotropin releasing hormone suppression in the treatment of precocious puberty? and (e) what other neuroendocrinologic changes occur during puberty?

These questions could be answered by the following studies: (a) a cross-sectional survey of children ages 3–18 years, examining growth and pubertal development. This would also include information regarding their hydrocephalus and shunt status, level of lesion, ambulation, Tanner staging and age at menarche. For children at selected ages, e.g., 12, 15, 18, questions could include health-related quality of life and

measures of psychosocial adjustment, thereby examining the consequences of precocious puberty; (b) a multicenter trial of gonadotropin releasing hormone suppression; and (c) basic science studies of hypothalamic function and hydrocephalus.

2. Sexual function

What is the range of sexual functional abilities (e.g., erection, ejaculation, arousal/orgasm) in individuals with spina bifida? Specific questions include the following: (a) What is the nature of spermatogenesis, ejaculation, and the range of fertility in males? (b) What factors predict sexual function and fertility? (c) What is the pathophysiology of sexual dysfunctions? and (c) What are effective treatments/management options?

These questions could be answered by the following studies: (a) cross-sectional survey of sexual function in a community-based representative sample, (b) cross-sectional study of adult males examining fertility and ejaculation, with analyses of factors associated with infertility and potential interventions to preserve fertility, (c) basic science (developmental biology) research to understand the effects of spina bifida on hypothalamo-pituitary-gonadal function and autonomic nervous system, (d) assays of infant males with spina bifida to examine human chorionic gonadotropin and sex hormone levels in early infancy, (e) longitudinal cohort study of infants who have had video urinary dynamic studies to determine if early findings predict subsequent sexual function, (f) evaluation of newer therapies, e.g., electrical bladder stimulation (its effect on autonomic function related to sexual function), genital stimulation, or nerve bypass therapies to improve sexual function and perineal sensation.

3. Psychosexual development

What happens in the psychosocial development of individuals who have spina bifida? Specific questions include the following: (a) what are the expectations and needs of families who have children with spina bifida? (b) what can promote adaptation and healthy psychosexual development? (c) what are the effects — both positive and negative — of bladder and bowel programs/interventions on psychosexual development? E.g., are independence in catheterization and bowel management

related to positive sexual outcomes? and (d) what are the knowledge gaps and attitudes among clinicians?

These questions could be answered by the following studies: (a) survey of practitioners to highlight knowledge gaps and attitudes for continuing education efforts, (b) focus groups with parents of infants, children, and adolescents to determine their needs and attitudes, (c) comparison of sexual development with individuals who have other chronic conditions and disabilities, and (d) development and implementation of interventions to promote adaptation and psychosexual development.

Self Care

Rationale

The term self care often is used interchangeably or in combination with terms such as self-management, self-regulation, and independence. Examples of areas of self care include eating, grooming, bathing, bladder and bowel management, and dressing. Children with spina bifida have been shown to have problems in areas of self care; these problems can arise from limitations in cognition, poor organizational skills, specific visuo-spatial problems, impairment of the upper extremities, limited mobility, or learned helplessness. Many children with spina bifida develop activities of daily living at a very slow rate; many adults fail to achieve independence. Other factors such as family functioning and expectations, education and training provided to the individual with spina bifida, and support may affect self care as well.

Summary of Current Information

Children with spina bifida have been found to have lower scores on various measures of self care, such as the Pediatric Evaluation of Disability Inventory (PEDI) and the Functional Independence Measure for Children (WeeFIM). Children with spina bifida score lower on these measures than do children without disabilities; their scores show a significant correlation with their clinical neurological levels, walking ability, intelligence, and bowel and bladder functions. Their pattern of impaired self care differs from children who have other disabilities. For example, children with spina bifida have problems with continence, and mobility/activities while those with hydrocephalus (exclusively) have worse scores on school activities, worries, sight, and communication. Important areas of self care include bladder and bowel management, hygiene, prevention of skin breakdown, and self-medication. Little evidence exists regarding the development of self care as a process or the nature of transitions in care. Areas unexplored include optimal timing for initiation and the typical sequence of events. In addition, little evidence exists on self care outcomes, including factors that affect (retard or facilitate) it, and regarding the evaluation of specific outcomes. Although some information is available about the employment status of adults, little is known about specific self-care skills, including their acqui-

sition and retention. Assistive technologies can help the self care of individuals with spina bifida, yet information about their actual use is rare. For instance, the prevalence of use of different types of assistive devices by people with spina bifida is unknown., nor are the benefits achieved by the use of these devices known.

Specific Recommendations

Several questions have been raised in the area of self care. These include the following:

1. What are the most effective and developmentally appropriate methods to teach self-care behavior? What successful intervention programs currently exist? What methods, e.g., peer mentoring, are effective? Can the use of guidelines/checklists improve the teaching of self care? What barriers to the development of self care exist? How can they be overcome?
2. What is the optimal way for individuals with spina bifida to learn self care? What factors in the child, e.g., nonverbal learning disability or upper extremity function, affect the method of learning? What parental and cultural factors need to be considered in designing an intervention? Can interventions be standardized (manualized)? What is the best developmental age to learn certain skills, e.g., self-catheterization? What is the best way to address the transition to adulthood to insure that individuals have the self-care skills they will need to be independent?
3. What instruments are optimal to evaluate self-care outcomes (e.g., hygiene, self-medication)?
4. What are the best assistive devices to help individuals with spina bifida with their self care? When should their use be introduced? How should their costs and benefits be measured?

These questions can best be answered by two types of studies: The first would be surveys to evaluate the current status of programs for self care and assistive technology. Surveys of adults with spina bifida should be included. The second type of research would be intervention studies that use control groups, are multidisciplinary and involve children from multiple centers. Formal protocols for improving specific self-care areas need to be developed. Measures that assess the person's developmental status and ability to use assistive technology would be critical for the

intervention. Homogeneous grouping of the individuals would also be important to minimize confounding factors.

Family Functioning

Rationale

Families who have a child with spina bifida face the same issues as families who have a child with other chronic conditions. These include the following: (1) the loss of the ideal child — this can lead to unresolved grief with associated states of denial, sadness, anger, and guilt; (2) daily burden of care — someone has to insure that the child is catheterized, receives appropriate monitoring, and takes medications; (3) increased expenses, including financial and loss of time; (4) lost opportunities such as the mother who cannot return to school or work in order to take care of the affected child; (5) neglected siblings; (6) confusing systems of health and other care; and (7) social isolation, especially because few family members or friends understand spina bifida. These may lead to clinical dysfunction and high levels of family stress. Parents and their child with spina bifida may have difficulty attaching, and parents may have increasing conflict that leads to isolation from the rest of the family (often the father), separation, or divorce. Children with spina bifida may view their parents as too intrusive or controlling — parents may have difficulty "letting go" as their children approach adulthood. On the other hand, the joys and challenges of having a child with spina bifida may strengthen some families, who demonstrate remarkable resilience. In order to address these issues, programs that deliver care to individuals with spina bifida have provided formal and informal education and support to families. However, the effectiveness of these interventions is uncertain.

Summary of Current Information

A review of published literature, with its few methodologically sound papers, shows that parents have higher levels of psychological distress (41% in one study) and that 12–25% of families exhibit clinical levels of family dysfunction. As might be expected, families most at-risk are those with a single-parent or older parents, those of lower means (socioeconomic status), and those in ethnic minority groups. Although all families are disrupted by spina bifida, many also show considerable resilience. Despite the needs of families that have a child with spina bifida, no studies documenting family therapy or similar interventions have

been published! Family-based interventions in other pediatric chronic conditions such as diabetes and cystic fibrosis have been shown to be effective in enhancing family function, decreasing conflict, and improving medical adherence (compliance).

Specific Recommendations

Previous studies in this area have utilized small, nonrepresentative samples, and often ignored groups like Latinos or those of decreased means. Many were not based on any theory and had no comparison group(s). Therefore, future studies should be coordinated, theory-driven programs of research with a longitudinal approach and multidisciplinary and multisite involvement that utilize control groups.

1. The first question to be answered is, What factors are associated with family resilience and with risk of family dysfunction in the face of the challenges of spina bifida? A study that is longitudinal, multidisciplinary, and multisite can answer this question and continue the work already published. This study should isolate areas of resilience that can be built upon with subsequent interventions. It should have large enough sample size, utilize mediational models, and focus on children of specific ages.
2. What family-based services, supports, and interventions can optimize parenting, prevent stress-related challenges, and effectively improve clinical dysfunction? These studies can adapt empirically supported interventions that have been successfully used with other families that have a child with a chronic condition (e.g., diabetes). The targets of the family interventions should clearly be delineated and include outcomes such as medical adherence, parenting/marital stress, sibling adjustment, independent functioning, and transition to adulthood. The intervention should be standardized and adapted for use with ethnic and minority families. The intervention should target families at greatest risk, e.g., those that are poor, have a single-parent, older parents, or are from minority backgrounds. These interventions should have a long enough follow-up to allow evaluation of changes that occur over time and their persistence.

Socialization

Rationale

Social competence, including the use of the peer group and other community contacts as support in the face of stressful events, has been shown to be critical to academic success, independence, vocational adjustment and quality of life. However, individuals who have spina bifida have been reported to have impaired social skills during childhood, in part because of nonverbal learning disorders and limited access to their peer group because of decreased mobility. Adolescents and adults with spina bifida also report isolation and have been reported to spend a great deal of time in solitary activities like watching television.

Summary of Current Information

Although, parents, clinicians, and educators have consistently found that most individuals with spina bifida demonstrate difficulties with social competence, this area has received little attention in the research literature. The conceptual definition of social competence and the way it is operationalized are not well established or consistently applied. For example, social skill behaviors, more general conceptions of social competence, rates of social activity, and the establishment of friendships have not been differentiated or systematically addressed. Most studies draw from one or more local spina bifida programs, fail to report the percentage of potential subjects who decline to participate, and do not use control groups. Few studies have utilized the currently accepted "gold standard" social assessment techniques, naturalistic observation and social behavior rating scales. Between 33% and 50% of individuals with spina bifida experience social isolation — as measured by decreased participation in social activities and time engaged with peers. Limited mobility is one factor that has been linked with social isolation. Participation in mainstream educational programs may be related to higher social competence; however, this finding may reflect a sampling bias, i.e., children who are not mainstreamed generally have lower levels of functioning already. Most measures of medical status are not related to social functioning. Family functioning and intellectual level may be related to social function. Social skills training programs and experiences at camps may affect social skills; however, the literature on social skills

training emphasizes the importance of identifying and training specific skills rather than addressing social functioning in general, which most programs for children with spina bifida have done.

Specific Recommendations

1. The first study recommended is one that will establish the prevalence and nature of the social challenges associated with spina bifida. This can be achieved by examining a representative multisite sample of individuals with an appropriate comparison group. Assessment of subjects should (a) address specific social skills and behaviors, general ratings of social competence, frequency of social activity, and the quality of relationships; (b) include behavior ratings by trained observers, peers, parents, and teachers; and (c) apply observational measures in key settings (e.g., playground, classroom, peer play in the home).
2. The second study would be to identify individuals who are at high risk for problems with socialization. This would entail an examination of the relationships between social challenges and factors that contribute to their occurrence. Neuropsychological functioning (such as the presence of nonverbal learning disabilities), communication skills, ambulation, continence and its management, CNS status, physical appearance, dimensions of family functioning, community and social factors (e.g., SES, status of peer group), and personality variables should be examined as potential risk or resiliency factors.
3. The third priority would be to identify interventions that are effective in improving socialization. This would include the following: (a) preventive interventions at key transition points aimed at parents, teachers, peers, and the individual with spina bifida; (b) social skills instruction in various clinical and educational settings addressing key behaviors known to promote social success (e.g., joining a group of peers, and conversation skills), behaviors related to managing the demands of spina bifida (e.g., explaining about one's disability, and asking for help at school), and social behaviors that can be barriers to adherence (e.g., excusing oneself to attend to self care); (c) interventions addressing peers and their response to children with disabilities, and (d) techniques for the integration of disabled and

nondisabled peers (e.g. Circle of Friends, and Peer Mentorship) and for the provision of opportunities to practice social skills in inclusive settings. These investigations would address the positive and negative interactions between the individuals with spina bifida and their peers.

Independence

Rationale

Independence includes a number of factors including self care, transition, employment, and quality of life. Functional assessments describe a person's strengths and limitations in self care (feeding, dressing, grooming, bathing, continence), mobility, communication/social, and cognitive abilities and are important aspects of independence. A number of factors can affect the development of independence in a person with spina bifida. These include impairments of mobility and upper extremity function, learning disorders, neurogenic bladder and bowel, prolonged or recurrent illnesses (e.g., decubitus ulcer or urinary tract infection), poverty, social isolation, and societal views of disability. Thus, individuals with spina bifida are at increased risk of reduced independence and underemployment, and many individuals experience difficulties with effective health care transitions.

Summary of Current Information

The evidence to date regarding autonomy, independence, and quality of life is very limited. There is some suggestion that adolescents with spina bifida have lower autonomy than those without. Parents of these children tend to be "overprotective" compared to parents of children without disabilities. However, autonomy is generally ill-defined and does not include areas like personal choice in daily life, choice in management of spina bifida, and issues of choice in school. Several potential measures of self-determination have been developed. Although degree of disability is linked with measures of independence, it is not directly associated with health-related quality of life; parental hope and the attitudes of adolescents with spina bifida have been somewhat predictive of quality of life. In studies of adults with spina bifida, social secondary conditions are not always linked to severity of disability, although the presence of a ventricular shunt correlates negatively with social outcomes such as employment.

Specific Recommendations

Two major areas require answers: (1) What levels of self-determination, independence, and quality of life do persons with spina bifida

attain? What factors are associated with this independence and what interventions make a difference to facilitate independence? (2) What do the follow-up studies of adults document about independence outcomes in spina bifida, and what evidence exists on effective interventions to maximize transitions, including health care transitions? The following four assessments are recommended:

1. A multicenter, representative survey of adults to assess their current living, psychosocial and medical statuses, and to identify how they are currently accessing health care services.
2. A longitudinal study looking at the course and impact of secondary conditions in adolescents and adults with spina bifida.
3. Examination of the factors promoting self-determination and independence. This research will need to be broad-based, encompassing individual trait, family, neurocognitive, and social factors that may influence independence and self-determination over the developmental trajectory.
4. Development of prospective interventional studies based on experiential or research-based knowledge of precursors to achieving self-determination. This may require the creation of new assessment tools, particularly in children, and will likely also need to include a strong parent educational component.

Education and Employment

Rationale

With improved care, more individuals with spina bifida are surviving into adulthood. A number of factors associated with this condition, however, may affect their ability to participate in education or assume meaningful employment. Individuals with spina bifida may have nonverbal and verbal learning disabilities as well as problems with attention that can affect performance in school, type of employment, as well as social integration. They have limited mobility secondary to paraplegia and often have impaired fine motor abilities (use of their arms and fingers). They may have problems accessing transportation. They may have recurrent medical problems such as urinary tract infection, skin breakdown, and ventricular shunt malfunction that can prevent them from participating in school or work for an extended time. They may have urinary or fecal incontinence, which can affect social interactions. If a job opportunity does not offer them adequate insurance coverage, they may be better off financially remaining unemployed and staying on Medicaid.

Summary of Current Information

Even though most children with spina bifida are eligible for the Early Intervention Program for Infants and Toddlers with Disabilities under Part H of IDEA (Individuals with Disabilities Education Act), few studies have examined the effects of this program on the development of these children. Because of the complexity of spina bifida, a wide variety of services may be required to optimize the development of children with spina bifida. However, no studies of such interventions in school settings have been published. Children with spina bifida may be classified in a variety of ways, e.g., orthopedically impaired or learning disabled — or they may be classified under Section 504 of the Rehabilitation Act — and receive widely different services. Also, few longitudinal studies of children with spina bifida have been published — most of the findings have been based on cross-sectional surveys.

Despite improved survival among individuals with spina bifida, few studies have been published that examine educational achievement after high school, entry into the world of work, or full participation in com-

munity life for people with spina bifida. Some studies have shown that the percentage of young adults with spina bifida continuing their formal education into college (34%) is lower than that of the general population (47%). In addition, young adults with spina bifida tend to be one or two years delayed in educational attainment compared to age-matched peers and typically have worse performance in certain subjects (like mathematics) than other individuals. Similarly, the employment rate has been found to be lower in individuals with spina bifida than in those without disabilities (33% vs. 69%) or even in those with other disabilities like cerebral palsy. Unemployment has been found to be related to cognitive abilities, and when employed, individuals with spina bifida are more likely to be working in supported or sheltered employment.

Specific Recommendations

Because children with spina bifida are extremely diverse, it is critical that any research project provides documentation and consistency in the description of participants. This would allow homogeneous groupings that would improve the validity of findings. Similarly, control groups should be utilized that are matched on characteristics directly relevant to the outcome variables of interest (e.g., functional abilities), and not just on those variables are easy to use for matching (e.g., age and IQ). This would increase the sensitivity of research for academic and social outcome studies. Longitudinal studies are preferable to cross-sectional studies, and studies should be cross-disciplinary (e.g., a collaboration between health and education researchers).

1. School

Specific questions include the following: (a) What factors and combinations of factors — e.g., impairments, environmental support, nature of the school — predict the performance in school of children with spina bifida? (b) What types of early intervention programs are most effective for preschool children? (c) What educational accommodations are effective with children and youth with spina bifida? (d) What are the characteristics of children and youth who are successful in different types of programs, e.g., inclusive model vs. pullout? (e) What types of transition programs are effective in helping youth make the passage to post-secondary education and work? and (f) What type of training

would improve teachers' abilities to optimize the education received by these individuals?

These questions could be answered by the following studies: (a) longitudinal study of preschool children to identify factors that predict academic success, (b) experimental intervention of different types of early intervention to determine effectiveness for children with various presentations of spina bifida, (c) longitudinal study of school-aged children with matched control group to evaluate school performance and education accommodations — e.g., what happens in inclusive versus segregated educational environments and the mechanisms through which the effects of those environments are transmitted to children with spina bifida, (d) experimental intervention to develop an effective transition program, and (e) a survey to identify characteristics of individuals who have been successful in education.

2. Work

Specific questions include the following: (a) Is progress being made in participation of persons with spina bifida in the labor force? (b) What are specific characteristics of persons with spina bifida who are successfully employed? (c) What incentives would improve employment in this population — e.g., insurance, federal assistance, or training grants? (d) What models for transition are effective across different levels of school and from school to work?, and (e) What are the costs and benefits of various models?

These questions could be answered by the following studies: (a) the studies outlined above under the category of school would be useful for the work arena as well. In addition, the following studies are proposed to address specific work-related questions: (b) experimental intervention designed to increase positive transitions of adolescents to the world of work, e.g., the use of prevocational and vocational skills and/or peer support groups beginning in middle adolescence, and (c) experimental intervention (perhaps on the policy level) to address job issues such as inadequate health insurance and discrimination in hiring.

Behavioral Health & Mental Health

Rationale

Children with spina bifida are theoretically at risk for developing problems with psychosocial adjustment, including low self-esteem. During the preschool years, the achievement of independence may be impaired by problems with mobility and bladder and bowel control. A sense of industry that develops in the school-age child may be reduced by the child's learning disorders as well as by his or her inability to compete with peers in sports. Difficulty in the school setting may exacerbate a preexisting poor self-image that many of these children have as a result of their physical disabilities. The feeling of being different can impair the establishment of peer relationships in both school and community. The child's self-esteem also may be lowered if he or she must continue to wear diapers or care for an ostomy. Some children may develop learned helplessness during childhood. They believe that unpleasant or aversive stimuli cannot be controlled and, therefore, cease trying to fix an aversive circumstance, even if they can exert some influence. During adolescence, lowered self-esteem may relate to a poor body image and difficulty in dealing with the sexual changes and feelings. Problems for the young adult with meningomyelocele may include increasing social isolation, a realization that the disability is permanent, and sexual dysfunction.

Summary of Current Information

Some findings support the notion that children with spina bifida experience lower mood and self-worth. They also consider themselves less competent in academic areas. Children with spina bifida may have more internalizing behavior problems as opposed to externalizing behavior problems. Some studies have found a higher-than-expected rate of depression in individuals with spina bifida, although the findings are not consistent. Social isolation appears to be an important marker for future mental disorder. Most of the studies published have been cross-sectional, involving small numbers of subjects. Literature regarding benefits of leisure/therapeutic activities to improving mental health outcomes in persons with spina bifida remains essentially descriptive. The presence of neurocognitive and core learning deficits in people

with spina bifida may affect measurement, especially the use of self-report tools. Measures of behavior may reveal elevation in scores compared to norms, but identifying if those behavior and mental health issues are significant clinically may be difficult. Furthermore, elevated scores on behavior rating scales do not necessarily equate with careful psychiatric descriptions and diagnosis. Models of providing mental health services to persons with spina bifida have not been developed and various barriers exist to providing services in the community, school or in medical centers.

Specific Recommendations

Mental health and behavioral health should be studied in the context of the whole person (i.e., all the domains of disability), family, school, community, and leisure activities. Issues exist regarding measurement. These include the following: measurements should include strengths-based (resiliency) approaches; mental health should include concepts of self-help, cognition, socialization, "maturity," leisure, and physical health. Research should delineate developmental trajectories of mental health/behavioral health issues; research should utilize models of mental health to optimize ways to deliver mental health services to persons with spina bifida. Research should target a spectrum of possible interventions (counseling, cognitive therapy, biofeedback, medications, etc.) and settings (community, school, hospital/clinic).

1. A multicenter, multidisciplinary, interventional, longitudinal study should be developed adhering to the criteria listed above. Children receiving two different intervention strategies can be compared and the benefits and costs of each one can be determined.

Reproductive Issues for Women with Spina Bifida

Rationale

Spina bifida can affect reproductive health in women. For example, many girls who have meningomyelocele with hydrocephalus experience precocious puberty. Females with spina bifida typically have decreased sensation in the perineum. Nonverbal learning disorder, which affects social interactions and self-esteem, is frequent in spina bifida and affects psychosexual development. Societal attitudes towards people who have disabilities also affects self-esteem and sexual self-image. Hip contractures can decrease abduction, flexion or extension of the hip joint, which can affect coitus, delivery of a baby and the ability to perform a pelvic examination. Vertebral anomalies and kyphoscoliosis may predispose pregnant women to disk problems or back pain. Ventriuculo-peritoneal shunts may fail during pregnancy. Latex allergies may affect the use of condoms as well as gynecologic and obstetric care. Women who have had complex urological reconstructions such as augmentation cystoplasty may require special monitoring during labor and delivery to avoid injury to the reconstructed bladder and outlet. Urinary tract infections, which already are common in individuals with spina bifida, increase during pregnancy and can lead to morbidity for the pregnant woman, premature delivery and possible adverse consequences to the fetus. Women with spina bifida have an increased risk of having offspring with spina bifida and other birth anomalies. Thus, genetic counseling and the use of high dose folic acid are recommended as part of periconceptual and pregnancy care.

Summary of Current Information

Although it has been documented that precocious puberty occurs in 12–16% of girls with spina bifida and hydrocephalus, it is not certain if girls with spina bifida without hydrocephalus are at risk for precocious puberty. No specific information is available regarding the qualities of menstruation in women with spina bifida. No studies examining specific gynecologic problems or reproductive endocrine dysfunction unique to women with spina bifida have been published. Little is known whether intraperitoneal gynecological procedures can affect ventriculo-peritoneal

shunt function. Virtually no research has been published using standardized criteria to document the incidence of sexual dysfunction in women with spina bifida; little is known about sensation, lubrication, or orgasms during sexual intercourse. The need for birth control and the safety of various methods for women with spina bifida has not been addressed. Case reports have shown that women with spina bifida have recognized secondary conditions such as recurrent urinary tract infections that may influence pregnancy. Also, the physiologic process of pregnancy and the effects of the growing fetus may exacerbate the secondary conditions of spina bifida. Kyphoscoliosis may become more pronounced, ventriculoperitoneal shunts may malfunction requiring revision and genitourinary diversions may fail. The occurrence of infertility in women with spina bifida is uncertain. No studies have been published exploring the effects of menopause in women with spina bifida. Likewise, information regarding specific incidence rates of reproductive cancers in women with spina bifida is nonexistent.

Specific Recommendations

Since so little is known about the reproductive health issues concerning women with spina bifida, studies that define the unique problems and needs of these women should be initiated. Collaboration with organizations such as the American College of Obstetrics and Gynecology, the Maternal Fetal Medicine Network of the National Institute of Child Health and Human Development, and Urological societies should be considered. Comprehensive, longitudinal, multicenter studies should be initiated to examine the three main areas listed below. Results of these studies should then direct intervention studies.

1. Preconception issues include the following: (a) precocious puberty — this includes the pathophysiology, effects of pharmacological delay, relationship between linear growth and independence, the use of gonadotrophin hormones, and the use of growth hormone; (b) menstrual disorders — these include the unique problems and effects of endocrine cycles on secondary conditions of spina bifida and vice versa, mobility problems with menstrual management, side effects of medications on menstruation, and long-term effects on osteoporosis when menstruation is stopped by medication or surgery; (c) safe birth control; and (d) anatomical changes of spina bifi-

da and their effects on the female reproductive system, including pelvic floor descent and kyphoscoliosis.

2. Pregnancy outcomes can be evaluated by a prospective, longitudinal, multicenter cohort study of young women. They would be followed over time to evaluate issues such as menstruation patterns, fertility, sexual concerns, the course of pregnancy and labor, methods of delivery, outcomes of newborns (including the occurrence of congenital abnormalities), health care during pregnancy, and the occurrence of spontaneous loss (abortion and miscarriage).
3. Preventive Health Care Interventions should be developed and examined scientifically. Protocols can be developed for issues such as the frequency of pap smears, mammograms, and menopause-related screening — including DEXA scanning, risk factors for arteriosclerosis (LDL, HDL, hypertension, diabetes, smoking, obesity, physical inactivity, and atherogenic diet), and evaluation of endocrine function. Issues related to this topic include access to health care, educational intervention for women with spina bifida, and continuing education for health care providers including guidelines for care.

Orthopedics

Rationale

Children with meningomyelocele have orthopedic conditions that are both primary and secondary. The primary conditions arise from failure of neural tube closure and consist of abnormalities of the vertebral bodies such as hemi-vertebrae and butterfly vertebrae. These lead to kyphosis (which may be present at birth) and scoliosis. Kyphosis and scoliosis typically worsen as the child ages, and are more common with higher level lesions. Abnormal innervation of the muscles of the back, pelvic obliquity, and tethering of the spinal cord probably contribute to these conditions as well — thus they have a secondary component. Secondary orthopedic conditions occur because of abnormal muscle function around joints and decreased mobility, although other factors may contribute as well. Joint contractures and deformities of the lower extremities occur commonly and include club foot, calcaneus and other foot and ankle deformities, knee and hip contractures, and dislocated or subluxed hips. Rotational deformities of the tibia and femur (e.g., coxa valga) also occur. Joint contractures or deformities may be present at birth or may develop as the child ages. They may occur across joints that have uneven muscle pull (e.g., the hip in a child with an L3-4 deformity where flexion occurs without extension) or in joints where all the muscles are paralyzed (e.g., the hip in a child with a thoracic level lesion who sits in a wheelchair all day). Osteoporosis of the lower extremities with or without pathological fractures frequently has been reported. A host of surgical procedures to address these orthopedic conditions have been developed; some of them have fallen out of favor.

Summary of Current Information

The goals of orthopedic management of the child with meningomyelocele include the following: maintain alignment of the extremities and joints, maximize range of motion, stabilize the spine and extremities, maximize function, provide comfort, and protect the skin. Although a vast number of articles has been published related to orthopedic care, most have been case series of children treated in a specialized center without a control group. For example, the only orthopedic article found related to spina bifida that was a controlled trial was a

study showing that these individuals have a higher occurrence of osteoporosis. One area where current practice has changed is the management of hip dislocation. Formerly these were treated surgically. However, no evidence has documented that this is advantageous for comfort or for function. In this population, pain from hip dislocation is rare unless the child has had surgery. Thus, the risks of surgery are greater than the risks of having dislocations, and the use of surgery to realign hips has fallen out of favor. Controversy also exists regarding the treatment of kypho-scoliosis in these children. For example, the optimal age for repairing congenital kyphosis is unclear. Nor is it clear which patients with scoliosis benefit from sacral instrumentation or a one stage versus two-stage operation. Most orthopaedic studies have evaluated pathophysiologic outcomes, e.g., the degree of spinal curve, but have not evaluated functional outcomes like activities, e.g., ambulation, and participation.

Specific Recommendations

All studies of orthopedic interventions should provide classifications of the children that allow generalizations to be made. A consistent definition of level of lesion should be used in all studies e.g., the lowest functioning nerve root motor level with a definition of minimum strength, or the most distal sensory level. All dimensions of outcome in the WHO International Classification of Functioning, Disability and Health (Body structure and function, Activities, and Participation) should be included as outcomes. These categories should be viewed broadly, e.g., including bone density as an outcome. Environmental factors should be included as potential confounding variables. The treatment should include postoperative rehabilitation as well as the surgical procedure itself.

Questions that should be answered include the following: (a) can factors be identified that predict which children will develop hip dislocation? (b) can nonsurgical interventions prevent hip dislocation? (c) what is the best treatment for hip flexion contractures? (d) what is the best treatment for unilateral hip dislocation? (e) what is the best treatment for kyphosis (e.g., decancellation vs. excision, length of fusion, and type of stabilization) and when should it be performed? (f) what is the best

treatment for scoliosis? (g) can risk factors for osteoporosis be identified? and (h) what is the best treatment for osteoporosis?

These questions could be answered by the following methods: (a) review the current databases that have been collected in centers like the University of Washington and Northwest University to evaluate predictors for hip dislocation and joint conractures, (b) convene pediatric orthopedists from around the country to develop standard criteria discussed above (e.g., what constitutes level of lesion) and collaborative trials, (c) perform controlled trials to treat unilateral hip dislocation, (d) perform collaborative controlled trials to treat kyphosis, (e) perform collaborative controlled trials to treat scoliosis, (f) perform a longitudinal cohort study to identify factors associated with the development of osteoporosis, and (g) perform controlled trials of various treatments for osteoporosis.

Mobility

Rationale

The higher the level of the lesion in meningomyelocele and the greater the muscle weakness, the more ambulation will be impaired. Even in children with low-level lesions, some impairment in mobility occurs. Most infants with meningomyelocele learn to belly crawl as their first means of mobility. Infants with strong voluntary hip flexion and some knee movement may eventually assume the all-fours creep. Children with sacral level lesions learn to walk well by 2 or 3 years of age with bracing at the ankles or no bracing at all. Children with mid-lumbar paralysis often require crutches and bracing up to the hip. Children with thoracic or high-lumbar paralysis may eventually stand upright and walk but only with support of the hips, knees, and ankles. This support may be provided by extensive bracing and/or mobility devices such as a parapodium, reciprocal gait orthosis (RGO), or hip-knee-ankle-foot orthosis (HKAFO) used in combination with crutches or a walker. As children with lumbar level lesions approach adolescence, most will rely increasingly on wheelchairs for mobility. This occurs for several reasons: their center of gravity and relative strength change, upright mobility requires a lot of energy, and mobility using a wheelchair is much faster than mobility with the other devices. Because most children with meningomyelocele will not become effective community ambulators, the supplemental or primary use of a wheelchair is considered at least by early adolescence since it offers the advantages of speed, efficiency, and attractiveness. Motorized wheelchairs can be used as early as a 24-month developmental level. Formal gait analysis using video cameras and sensors linked to computers has been used to analyze the gait of children with meningomyelocele and guide subsequent orthopedic management.

Summary of Current Information

The likelihood of ambulation has been found to be a function of several factors, including the location of the lesion and associated muscle weaknesses, cognitive functioning (especially nonverbal performance), impaired hand function, visuo-spatial function, executive function, complications like joint contractures and shunt infections, the

involvement of the parents, and the therapy program. For those children with a combination of high-level lesion and mental retardation, where walking may not be a realistic goal, wheelchair training is started early. Several studies have been performed assessing the energy efficiency of ambulation in orthotic devices, with conflicting results regarding the metabolic demands of children walking in HKAFOs or RGOs. None of the mobility aids seems to be more effective than any other in maintaining extension at the hip and knee. The velocity of walking seems to be faster with HKAFOs than with RGOs, though some controversy exists. Differences in velocity may be due to variations in gait patterns (swivel vs. swing-through), and the types of assistive devices used to walk (walker vs. crutches). Upright ambulation with either the HKAFO or the RGO theoretically offers both physiologic and psychological benefits; these have not been proven. Both orthotics decrease velocity and increase oxygen cost. In children with low-level lesions, ankle-foot orthoses (AFOs) are beneficial in terms of increasing velocity of walking, decreasing time spent in double limb phase of gait and decreasing hyperdorsiflexion of the ankle in midstance phase of gait. Most studies have looked at velocity or energy control walking as the functional outcome.

Specific Recommendations

1. The first study proposed would be an evaluation of the benefits, costs, and functional outcomes of an upright mobility program. This study would examine the ease of getting around in the environment such as the home or school, getting to and from the school, the store, a recreational activity, and participation in sports as the functional outcome. A randomized controlled trial to study the efficacy of bracing and ambulation in the children with thoracic and upper lumbar levels of paralysis would be ideal. Factors such as the type and duration of training, bone density (osteoporosis), financial costs, visuo-spatial abilities, decubitus ulcers, upper extremity functioning, self care (e.g., ability to catheterize), continence, and use in varied environments should be evaluated. An alternative to a randomized controlled trial would be a prospective clinical trial in which the parents choose either a walking training-orthotics program or an early wheelchair program. If part of the program

includes upper body strength training, its role in facilitating upright mobility could be evaluated as well.

2. The second study recommended is a natural history study to evaluate the change in mobility that occurs during adolescence from upright to wheelchair. Questions that could be answered include the following: (a) when is the change physiological and when does it represent a problem such as tethered spinal cord? (b) is their an optimal time to introduce wheeled mobility? and (c) what happens to mobility during adulthood?

Gastroenterology and Nutrition

Rationale

Most children who have spina bifida have bowel problems including constipation and incontinence. Abnormal neuronal migration in the gastrointestinal tract can decrease gastrointestinal motility. Because of difficulty swallowing related to the Chiari II malformation some children with spina bifida have decreased fiber in their diet. In addition, decreased physical activity and decreased abdominal muscle tone contribute to constipation. Because of abnormal sacral nerves, children with spina bifida have dysfunction of the external rectal sphincter and puborectalis muscle. They also have abnormal rectal and anal sensation and lose the ability to differentiate between gas, liquid stool and solid stool. These changes lead to fecal incontinence, which can be socially devastating. Many children with spina bifida have decreased activity levels because of paraplegia; they may have altered hormones (e.g., precocious puberty and decreased growth hormone); they may have different diets because of difficulty swallowing. All these factors can place them at increased risk for developing overweight and obesity.

Summary of Current Information

Attempts at bowel management typically include encouraging foods that are high in fiber. Timed potty sitting is tried in preschool children. Daily laxatives such as polyethylene glycol and lactulose commonly are used. Suppositories, manual disimpaction, and laxatives also are frequently used. Daily enemas and biofeedback have been attempted. Very few randomized controlled trials have been conducted in this area, however; none of the current treatments has been found to be superior to any other. In addition, "successful" bowel management is not defined and effectiveness of management options is not clearly established. Two newer surgical procedures, one that connects the appendix to the colon (Malone) and the other that provides a direct connection between the abdominal wall and the colon (cecostomy), allow irrigation of the colon on a regular basis and are called antegrade colonic enema or ACE procedures. Percutaneous insertion of a tube into the descending colon also has been attempted. These approaches have been evaluated in children for whom more conventional bowel techniques have failed. Success

rates for the ACE procedures range from 70–90% with complication rates of 15–80%. The cecostomy seems to be a somewhat better procedure. However, virtually all studies are of nonconsecutive cases from ill-defined, nonrepresentative samples. In most studies, previous treatments are uncertain and the outcomes measured have been very limited. Likewise, little evidence has been published on the decision-making factors that influence the implementation and achievement of bowel management program(s) in individuals with spina bifida and/or their families. Little is known regarding bowel management in adults with spina bifida.

Children with spina bifida, particularly those with thoracic to L-2 lesions, are at increased risk for obesity as a result of their decreased energy expenditure. About two thirds of these children are significantly overweight. However, factors other than activity level seem to affect the occurrence of overweight and obesity. Because published studies have been limited, the factors leading to obesity remain uncertain. Little is known regarding nutrition in adults with spina bifida.

Specific Recommendations

1. In order to evaluate risk factors and identify treatments for constipation and fecal incontinence, two studies are proposed. The first would be a survey using a representative sample, possibly a cohort of individuals with spina bifida, to evaluate the natural history of these issues including the natural history of bowel dysfunction, prevalence of constipation and fecal incontinence, and currently used therapies. The second study would be a graded intervention trial beginning with standardized medical treatments then progressing to surgery. Potential confounding factors that should be examined include rectal sensation and tone, level of lesion, GI motility, diet, physical activity, medications, and body habitus. When surgery is used, it should follow standardized protocols and follow-up. Outcomes include continence, which would have to be defined *a priori,* activities, and participation, e.g., swimming, sports, dating, sleeping over, and sexual function.
2. The second area of concern is nutrition, including overweight and obesity. Studies would encompass an evaluation of prevalence and risk factors as well as morbidity. Factors to be examined include

body composition, which could be evaluated using DEXA scan, energy expenditure, family BMI, activity level including time spent in sedentary activities (television, video games, Internet), growth hormone, onset of puberty, associated nutrition and malnutrition including albumin and pre-albumin, diet, respiratory problems, occurrence of decubitus ulcers, and depression. Once this study is completed, an intervention based on the findings from the first study could be initiated.

Integument

Rationale

Skin sores or decubitus ulcers frequently occur in children with meningomyelocele, whose weight-bearing surfaces (e.g., feet, buttocks) are insensitive to pain. Edema, diminished movement and deep vein thrombosis may contribute to these breakdowns. These children may sustain injuries that they do not feel. In addition, the effect of spina bifida on autonomic nerves may affect the vascular supply to insensate areas, and sores that are present may require extensive periods to heal, in a way similar to those who have diabetes. This problem becomes more frequent during adolescence and, if not caught early, may require prolonged hospitalizations for debridement, skin grafting, and intravenous antibiotics. Failure of skin ulcers to heal in a reasonable amount of time may be related to an underlying infection (e.g., of the bone), requiring surgical debridement and intravenous antibiotic treatment; it may also be related to chronic contamination with urine or stool or to inadequate intake of protein and other nutrients.

Summary of Current Information

In one study, over a 13-year period, 75 children with meningomyelocele spent the equivalent of 17 years in hospital days at a cost of more than 2 million dollars. Treatment frequently results in long-term immobility, confinement to bed or a wheelchair and the necessity of plastic surgery. In a review of 193 patients referred to an adult spina bifida program, pressure sores or skin breakdown were a current problem for 8 patients (4.1%) and a previous problem for 74 (38.3%). In a review of published literature, no studies were found that examined the pathogenesis of pressure sores in individuals with spina bifida. Similarly, very few studies have addressed the issue of prevention of decubiti in this population. In general, the management and treatment of pressure ulcers have been guided by the 1994 AHCPR (now AHRQ) Guidelines for Pressure Ulcer Treatment. These guidelines were created by a panel of experts in the field of pressure ulcer treatment, but do not specifically address children or individuals with spina bifida. Nonsurgical management such as debridement, the use of special dressings, and vacuum closure has been evaluated in samples that did not include individuals with

spina bifida. Several case series of various surgical techniques (such as skin flaps) to treat decubiti in individuals with spina bifida have been published. Few conclusions about the effectiveness of these techniques can be made based on these studies.

Specific Recommendations

The prevention of skin breakdown is a priority. However, the currently published literature cannot allow determination of risk factors. Nor can it be used to decide which preventive measures are most effective. Therefore, the following recommendations are made:

1. Evaluate pathophysiologic factors that increase the risk of developing skin breakdown. These include incontinence, orthopedic factors such as pelvic obliquity and joint contractures, level of lesion, shear forces, neuropathic foot, and psychological factors (such as depression and learning disabilities).
2. Once the risk factors are known, preventive programs should be undertaken to identify those that are effective and efficacious.
3. Develop intervention programs, based on the guidelines of the AHRQ to determine if they effective for individuals with spina bifida. Methods used to treat pressure sores in other populations can be evaluated to determine if they are effective in individuals with spina bifida.

Latex Allergies

Rationale

In the early 1990s, several reports of children with spina bifida having severe allergic reactions to latex were published. Most of these occurred in the operating suite; most were anaphylactic; some were fatal. It was thought that the sharp increase in demand for latex gloves due to the HIV/AIDS epidemic had led to poor quality control, more antigenic gloves and, therefore, to severe allergic reactions to these products. Using a variety of testing methods, 25–73% of children with spina bifida have been found to be sensitive to latex. Some of these seemed to have no problem with latex, while others would develop symptoms just by walking into a room that contained latex products. It was found that between 30% and 72% of latex-sensitized individuals actually had latex allergies. As a result of these high prevalence rates and the serious nature of the reactions, latex avoidance has been recommended for all individuals with spina bifida from birth through adulthood. Avoidance is recommended in all environments, including the home, and especially during surgery, dental procedures, and invasive diagnostic studies.

Summary of Current Information

A review of published literature has identified factors that are associated with latex allergies. Not all studies, however, show the same associations. Factors found to be associated in many studies include the number of previous surgeries and the presence of allergies (atopy) in the person with spina bifida. Severe anaphylactic reactions are more likely to occur in individuals with spina bifida who have had more severe allergic reactions in the past and who have daily rectal disimpaction. Testing for latex sensitivity has been done using skin and serum tests. No clear superiority of one method over another has been identified. Several studies have shown that children provided with latex precautions have a lower occurrence of latex allergies than historical controls. However, this secular decrease in latex allergies may not be related to preventive measures but to higher quality latex in the current environment. There is some evidence that latex avoidance in surgery can decrease the occurrence of severe reactions. It is not clear whether routine avoidance can prevent latex allergies. No evidence has been pub-

lished that addresses how to most effectively teach or manage latex avoidance programs.

Specific Recommendations

Very little is known about the occurrence of latex sensitization or allergy in adults who have spina bifida. Most earlier research did not evaluate measures of adherence to protocols in the homes or other environments where children spend extensive time (like the school). Creating latex-safe environments can be difficult and requires education and change in behavior.

1. The first research project recommended would be a longitudinal, natural history study (perhaps as a part of, or an adjunct to, the Management of Myelomeningocele Study [MOMS] conducted by the NIH) to assess the development of latex sensitization and allergy in babies and young children. The study should include periodic serum testing, assessment of exposure to latex and cross-reacting foods, analysis of all allergic episodes, evaluation of how families learned about latex allergy, barriers to latex avoidance, and quantitative assessment of adherence to latex precautions in various environments.
2. The second study would be to evaluate latex allergy in adults with spina bifida (possibly through SBAA). The research should include: Serum testing for antibodies, clinical history of reactions, assessment of exposure to latex and cross-reacting foods, and limitations to full participation in activities due to latex allergy or fear of latex reactions.

Appendix 1: Participant List

U.S. Department of Health and Human Services
U.S. Department of Education

Spina Bifida Association of America
Spina Bifida Foundation

Evidence-Based Practice in Spina Bifida: Developing a Research Agenda

The Washington Court Hotel
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Appendix 2: Conference Agenda

U.S. Department of Health and Human Services
U.S. Department of Education

Spina Bifida Association of America
Spina Bifida Foundation

Evidence-Based Practice in Spina Bifida: Developing a Research Agenda

The Washington Court Hotel
Washington, D.C.
May 9–10, 2003

Agenda

Friday, May 9, 2003

7:00 a.m. – 7:45 a.m.	*Registration and Continental Breakfast*
7:45 a.m. – 8:10 a.m.	**Welcome and Introductions**
	Jose Cordero, M.D., M.P.H., Director, National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention
	Denise Dougherty, Ph.D., Acting Associate Director, Office of Priority Populations Research, Agency for Healthcare Research and Quality
	Duane Alexander, M.D., Director, National Institute of Child Health and Human Development, National Institutes of Health
	Theresa San Agustin, M.D., Program Officer, National Institute on Disability and Rehabilitation Research, U.S. Department of Education
	The Honorable Christopher H. Smith (R-NJ), Congressman, U.S. House of Representatives
8:10 a.m. – 8:30 a.m.	**Meeting Overview,** *Gregory Liptak, M.D., M.P.H.*
8:30 a.m. – 9:30 a.m.	**Panel 1—Denise Dougherty, Ph.D., AHRQ, Moderator** **Orthopaedics,** *Mary Clark, M.D.* **Neuropsychology,** *Jack Fletcher, Ph.D.* **Integument and Vascular,** *Donna Zahra, Ph.D., A.R.N.P.* **Neurosurgery 1,** *Mark Dias, M.D., F.A.A.P.*
9:30 a.m. – 9:40 a.m.	*Morning Break*

9:40 a.m. – 10:40 a.m.	**Group Sessions for Panel 1** *Group A: Orthopaedics* *Group B: Neuropsychology* *Group C: Integument and Vascular* *Group D: Neurosurgery 1*
10:45 a.m. – 11:40 a.m.	**Panel 2—Robert Jaeger, Ph.D., ICDR, Moderator** **Mobility,** *Andrew King, M.D.* **Work and School,** *Rune Simeonsson, Ph.D., M.S.P.H.* **Neurosurgery 2,** *Mark Dias, M.D., F.A.A.P.* **Urology 1,** *David Joseph, M.D.*
11:45 a.m. – 1:15 p.m.	**Lunch and Presentation** *Harold Pote,* President, Spina Bifida Foundation *Judy Woodruff,* Prime Anchor, CNN
1:30 p.m. – 2:30 p.m.	**Group Sessions for Panel 2** *Group A: Mobility* *Group B: Work and School* *Group C: Neurosurgery 2* *Group D: Urology 1*
2:30 p.m. – 2:45 p.m.	*Afternoon Break*
2:45 p.m. – 3:45 p.m.	**Panel 3—Lou Quatrano, Ph.D., NICHD, Moderator** **Self-Care,** *Patricia Braun, R.N.C., C.P.N.P., M.A., M.S.N.* **GI and Nutrition,** *Patricia Braun, R.N.C., C.P.N.P., M.A., M.S.N.* **Behavior and Mental Health,** *Richard Adams, M.D.* **Urology 2,** *David Joseph, M.D.*
4:00 p.m. – 5:00 p.m.	**Group Sessions for Panel 3** *Group A: Self-Care* *Group B: GI and Nutrition* *Group C: Behavior and Mental Health* *Group D: Urology 2*
5:30 p.m. – 7:00 p.m.	**Dinner**
7:15 p.m. – 8:15 p.m.	**Panel 4—David Joseph, M.D., Moderator** **Discussion of Priorities,** *All Participants*

Saturday, May 10, 2003

7:00 a.m. – 8:00 a.m.	*Continental Breakfast*
8:00 a.m. – 9:00 a.m.	**Panel 5—Rosaly Correa-de-Araujo, M.D., M.Sc., Ph.D., AHRQ, Moderator** **Women's Issues,** *Amie Jackson, M.D.* **Socialization,** *Mark Merkens, M.D.* **Learning,** *Jack Fletcher, Ph.D.* **Sexuality,** *Adrian Sandler, M.D.*
9:00 a.m. – 9:10 a.m.	*Morning Break*
9:10 a.m. – 10:10 a.m.	**Group Sessions for Panel 5** *Group A: Women's Issues* *Group B: Socialization* *Group C: Learning* *Group D: Sexuality*
10:15 a.m. – 10:45 a.m.	**Panel 6— Don Lollar, Ed.D., CDC, Moderator** **Family,** *Grayson Holmbeck, Ph.D.* **Independence,** *Kathleen Sawin, D.N.S., R.N., C.S., F.N.P., F.A.A.N.*
10:50 a.m. – 11:50 a.m.	**Group Sessions for Panel 6** *Group A: Family* *Group B: Independence*
11:50 a.m. – 1:00 p.m.	**Lunch and Presentation** **Fetal Surgery,** *Catherine Shaer, M.D.*
1:15 p.m. – 2:30 p.m.	**Final Discussion,** *All Participants*