

Table of Contents

2	Overview of Promoting Excellence Workgroups	14	Recommendations for Educators
4	Definitions	14	Recommendations for Professional Organizations
4	Introduction	14	Recommendations for National Patient Advocate Organizations
5	The Genetic Basis	15	Recommendations for the Huntington's Disease Society of America (HDSA) and Other Community Huntington's Disease Resources
6	Clinical Manifestations and Epidemiology	15	Recommendations for Caregivers
7	Huntington's Disease and Palliative Care	16	Recommendations for Policy-Makers
8	Total Functional Capacity Scale	16	Recommendations for First Responders
8	Methodology	16	Recommendations for Health Care Delivery Systems
9	Conclusions	18	Summary
10	Recommendations to the Field: A Call for Actions	20	HDSA Centers of Excellence
11	Recommendations for Researchers	22	Huntington's Disease Peer Workgroup Members
12	Recommendations for Public and Private Funders	24	Appendix CD-ROM Table of Contents
12	Recommendations for Hospice Programs		
12	Recommendations for the Centers of Medicare and Medicaid Services (CMS)		
13	Recommendations for Health Care Practitioners		



*O*VERVIEW OF PROMOTING
EXCELLENCE WORKGROUPS



In 1997, *Promoting Excellence in End-of-Life Care*, a national program of The Robert Wood Johnson Foundation, received 678 Letters of Intent in response to a Call for Proposals for fewer than 25 grants to develop innovative palliative care programs. In an effort to tap this interest and energy and prompt rapid change, “Peer Workgroups” were created, composed of interested clinicians and researchers who are working to extend palliative care to special populations of patients in challenging domains and specific niches of medicine.

Seven workgroups were created in areas with critical patient need that indicated readiness to address end-of-life issues: Amyotrophic Lateral Sclerosis (ALS); The Children’s International Project of Palliative/Hospice Services (ChIPPS); Critical Care; End-Stage Renal Disease (ESRD); HIV/AIDS; Huntington’s Disease (HD); and Surgeon’s Palliative Care. Each workgroup was asked to assess the existing evidence base; to identify resources, gaps and products for the field; and to develop *Recommendations to the Field*.

The *Promoting Excellence* Huntington’s Disease Peer Workgroup, convened in October 2001, consisted of 16 members representing clinicians, psychologists, nurses, social workers, ethicists, therapists and educators with experience in the care of people with HD, and a family caregiver whose children have Huntington’s Disease. Chaired by Richard Dubinsky, M.D., Associate Professor in the Department of Neurology at the University of Kansas Medical Center, the group divided into three subcommittees who met over 18 months to address the Workgroup’s charge: Care, Education, and Research and Public Policy.

This report from the HD Peer Workgroup is written to advance palliative care for patients with Huntington’s Disease and their families, and offers *Recommendations to the Field* to improve the comfort and quality of life of patients, families and caregivers affected by this disease – throughout the disease, and particularly in its advanced stages. Palliative care for people with HD and their families can serve as a model of palliative care for other neurodegenerative diseases such as multiple sclerosis, stroke, Parkinson’s and Alzheimer’s diseases.

Definitions

The following definitions were adopted by the HD Peer Workgroup to assist readers of this report.

The beginning of the end of life

The point at which the person is no longer able to live independently.

Palliative care

Interdisciplinary team-based care, focused on comfort and quality of life for patients and their families, with the goal of improving overall quality of life rather than prolonging life.

Health care practitioner

All the professional and volunteer providers of health care for people with HD. This includes but is not limited to physicians, nurses, therapists, social workers and aides.

Community HD resources

Include HD clinics, HDSA support groups, Centers of Excellence and chapters.

Long-term care facilities (LTCF)

Adult day care centers, respite services, assisted living, intermediate and skilled nursing homes.

Spirituality

A sense of one's place and comfort in the larger universe. Although spirituality is often associated with organized religion, spirituality can exist outside of a formal religion or a religious place.

Introduction

A successful corporate accountant begins uncontrollably fidgeting. He loses motor control, and then he loses his job. Another woman, unable to speak, grieves the death of her son by screaming down a nursing home's halls. A man stumbles down the street and is arrested for intoxication. He isn't drunk. He has Huntington's Disease.

These scenarios are not atypical for the 30,000 people in the United States with Huntington's Disease (HD), an inherited neurodegenerative disorder with onset usually between 30 to 50 years of age, and a disease course spanning two or more decades to death. It involves progressive motor, cognitive and behavioral symptoms from onset until death. No cure exists. The disease, which is often misinterpreted, is underreported and understudied.

Its tragedies, however, are many. Each child of a person with Huntington's Disease is at risk to develop HD. As the disease progresses, a veil drops between people with HD and their friends and family, slowly robbing them of their ability to communicate and prompting, according to one study,* almost 30 percent to attempt suicide at least once, generally in the early stages of the disease.

Perhaps most disturbing is that the disease seems wrought with conditions that are neither costly nor difficult to correct. However, many staff, burned out by patients' demands, fail to understand the special requirements of HD, such as the need for more frequent snacks to combat the weight loss associated with the disease.

A major consequence of the combination of cognitive and behavioral problems and the long course of the disease is that many people with HD have consumed most financial resources by the time they need them most – toward the end of life.

*See Appendix C, Research and Policy Subcommittee Report, on enclosed CD.



The Genetic Basis

The genetic mutation that causes HD is an unstable expanded trinucleotide repeat on the short arm of the 4th chromosome. In this area of the genome, the gene normally has fewer than 30 CAG repeats. When there are 40 or more CAG repeats, HD is fully penetrant. A gene is fully penetrant when all gene carriers manifest the illness. When there is reduced penetrance, some carriers may not develop signs of the illness. When there are 36 to 40 CAG repeats in the huntington gene, the disease is less than fully penetrant, but due to the instability in the mutation, a full-length mutation may be passed to the next generation where HD will appear. When there are 30 to 35 repeats, the carrier will not develop the disease, although a longer CAG repeat length may be transmitted to the next generation. The trinucleotide repeat instability is more pronounced in male gene carriers, which leads to the rare appearance of juvenile onset in some children of affected men (and occasionally women). Because of this inherent instability, the mutation may arise *de novo* in a family.

“Illness and disability are a family affair. The diagnosis that made our family member need care happened to us as well. It is our diagnosis just as much as it’s theirs. I have a psychosocial form of the disease, just as my husband has a clinical one.”

– A Family Member



Clinical Manifestations and Epidemiology

The mean age at onset of HD is around 37 years old, but ranges between two and 85, with some cases falling outside this range. In individual cases, disease onset may be difficult to pinpoint, particularly when behavioral and psychiatric changes are considered. In patients who are closely followed, a zone of onset appears spanning three to five years. The presenting symptoms vary. In a review of the nature of onset in 510 HD patients, Di Maio et al. found that neurological symptoms heralded the disease onset in 59 percent of subjects, while psychiatric symptoms were first noted in 23 percent, and 18 percent had both at onset.¹

Studies of motor onset in the Lake Maracaibo, Venezuela community in which HD is endemic, showed that patients first develop subtle changes in volitional eye movement and clumsiness before they show frank motor changes due to the disease. Initially, people with HD exhibit involuntary,

spasmodic movements of limbs and facial muscles. Over the course of years, they lose normal motor functions and become bed bound.² This is indicative of most patients with HD.

Juvenile HD, which has a strikingly different phenotype, begins with akinesia, rigidity and dystonia and is often accompanied by myoclonic muscle tremors and twitches or seizures. The progression of HD occurs independent of any therapeutic intervention. It remains unclear whether or not progression rate relates to the CAG repeat length, thus limiting the prognostic value of genetic testing for disease severity and progression.^{3,4,5}

The prevalence of HD is estimated to be between three and 10 people/100,000 people of European descent.^{6,7} (See corresponding chart below.) Most patients are identified by the presence of a suggestive neurologic course in a patient with a positive family history. However, in a study of symptomatic patients undergoing confirmatory genetic tests in British Columbia, nearly 25 percent

Geographic region	Prevalence
Grampian/NE Corner of Scotland (Simpson, 1989)	9.94/100,000
Ireland (Morrison, 1995)	6.4/100,000
New South Wales (McCusker, 2000)	6.3/100,000
Glasgow/West of Scotland (Bolt, 1970)	5.2/100,000
Mauritius, Independent Island Republic in the Western Indian Ocean (Hayden, 1981)	4.6/100,000
Hong Kong (Chang, 1994)	3.7/1,000,000
South Africa (Hayden, 1980)	0.0/1,000,000

The burden of caregiving and the process of seeking out community resources to ensure quality care as the disease progresses must not fall solely on HD family members.

had negative family histories. Although in many cases the family history is obscured by misdiagnosis, obfuscation or competitive mortality, researchers estimate that as many as 8 percent of HD patients with negative family histories may reflect new genetic mutations arising from borderline premutations.⁸ Moreover, the study of HD likely suffers from under reporting of patients. Studies of mutational flow suggest that accurate diagnosis is poor with late onset HD.⁹ Current epidemiologic data show that there are 25,000 to 30,000 HD patients in the United States. On average, five first-degree relatives are at risk for each affected person.

Huntington's Disease and Palliative Care

The HD Peer Workgroup chose to define the beginning of the end of life in HD as the point at which the person is no longer able to live independently. Palliative care is defined as interdisciplinary team-based care, for people with life-threatening illness or injury, which addresses physical, emotional, social and spiritual needs and seeks to improve quality of life for the ill person and his or her family. Patients need palliative care well before they are near the end of life. In fact, the World Health Organization defines the beginning of the need for palliative care for people with cancer as the time of diagnosis. Demonstration projects and pilot studies funded by The Robert Wood Johnson Foundation showed that early introduction of palliative care to people with incurable diseases can enhance the quality of life throughout illness while reducing hospital admissions, allowing more patients to be cared for at home and die at home.¹⁰ Preliminary findings indicate that care can be

improved while diminishing the overall medical expense.

In a similar fashion, the need for palliative care in Huntington's Disease begins when a diagnosis of HD is first considered. Because a patient's ability to communicate diminishes over the course of the disease, it is at the beginning that the need for information about HD is the greatest. The future can be discussed and plans made for later in the course of the illness when the need for palliative care intensifies. A palliative care health worker can then coordinate care according to the patient's preferences, and stand ready to handle problems before crises arise. A team of clinicians can work to improve care and quality of life, emphasizing continuity of care, communication, symptom management, crisis prevention and early crisis management. Early introduction of palliative care, a seamless provision of services of gradually increasing intensity, is imperative.

The progression of HD is divided into five stages defined by the patient's score on the Total Functional Capacity (TFC) Scale, which details level of function in the domains of workplace, finances, domestic chores, activities of daily living and requirements for unskilled or skilled care.^{11,12} The interdisciplinary palliative care team assists patients and their families in articulating their wishes and the goals of care, and developing a plan to achieve those goals. When a person with HD is no longer able to live independently, palliative care turns toward adaptation to advanced illness and disability and issues of life completion and closure, while continuing to provide comfort and support for the person with HD and his or her family.

No cure is presently available for Huntington's Disease. Researchers, however, have made great strides in understanding the basic mechanisms

causing HD that may lead to potential treatments, through studies funded publicly and through HDSA. The Huntington Study Group (www.Huntington-study-group.org), as well as other individuals and groups, have conducted clinical studies aimed at reducing disease progression and the impact of symptoms. Those involved in research and in care hope that a treatment and cure for HD are developed in the near future. In the interim, the goal must be to provide the best possible care for people with HD and their families. National organizations such as the Huntington’s Disease Society of America (www.HDSA.org) and the Huntington Society of Canada (www.HSC-ca.org) are invaluable in providing education and support to those with HD as well as funds for care and research. Health care alone cannot provide the necessary support to HD patients and families. Integrated care and connections that utilize resources from the entire community provide the best care.

The burden of caregiving and the process of seeking out community resources to ensure quality

care as the disease progresses must not fall solely on HD family members. Non-traditional approaches that integrate attention to personal care and comfort in addition to curative treatment and efforts to modify the course of the disease can improve overall quality of life for HD patients and their families.

Methodology

The Care, Education, and Research and Public Policy Subcommittees identified needs of the HD community and searched for available resources using electronic and other databases, delineating those resources currently available and those that need to be developed to provide optimum care for people with HD and their families.

- The Care Subcommittee focused on the domains of autonomy, dignity, meaningful social interaction, communication, comfort, safety and

Total Functional Capacity (TFC) Scale

	Engagement in occupation	Capacity to handle financial affairs	Capacity to manage domestic responsibilities	Capacity to perform activities of daily living	Care can be provided at...
STAGE I (TFC 11-13) (0-8 YEARS)	USUAL LEVEL	FULL	FULL	FULL	HOME
STAGE II (TFC 7-10) (3-13 Y)	LOWER LEVEL	REQUIRES SLIGHT ASSISTANCE	FULL	FULL	HOME
STAGE III (TFC 3-6) (5-16 Y)	MARGINAL	REQUIRES MAJOR ASSISTANCE	IMPAIRED	MILDLY IMPAIRED	HOME
STAGE IV (TFC 1-2) (9-21 Y)	UNABLE	UNABLE	UNABLE	MODERATELY IMPAIRED	HOME OR EXTENDED CARE FACILITY
STAGE V (TFC 0) (11-26 Y)	UNABLE	UNABLE	UNABLE	SEVERELY IMPAIRED	TOTAL CARE FACILITY ONLY

“Less attention...is paid to caregiver grief, that relentless ongoing process that is brought about, not by a loved one’s death, but by the changed aspects of their life, and inevitably of our own.” —A Caregiver

order, spirituality, enjoyment, entertainment and well-being, nutrition and functional competence.

- The Education Subcommittee determined what educational resources were needed throughout the continuum of HD, investigated available publications and resources on palliative and end-of-life care in HD and determined needed improvements in the education of health care practitioners, as well as in the counseling and self-care education extended to HD patients and their families and in the training of community workers.
- The Research and Public Policy Subcommittee determined that improvement of end-of-life care required a thorough understanding of how and where HD patients spent their final years, who provided care, primary sources of funding and pertinent policies that affected care. This subcommittee investigated published literature in the areas of epidemiology, financing and policy to identify gaps in research and to make recommendations that address unmet needs in these areas.

Subcommittee reports are contained in the Appendices on the enclosed CD.

Conclusions

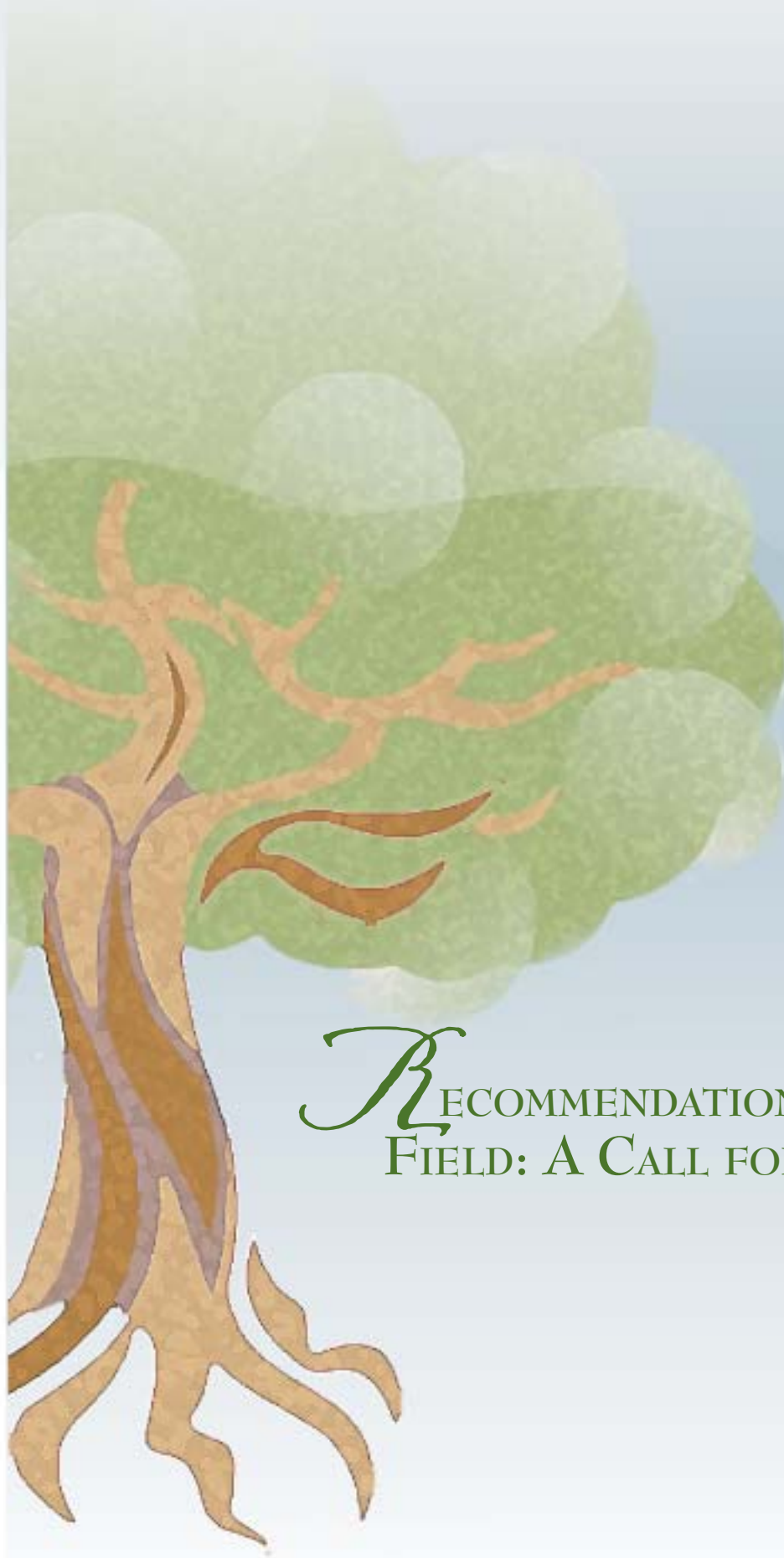
Based on its research, the Huntington’s Disease Peer Workgroup asserts that fundamental changes are needed in the care of people with Huntington’s Disease. Health care delivery systems can no longer focus solely on cure and saving life. They must also focus on enhancing the well-being and quality of life of people with HD and their families through-

out the course of their illness. Goals of care extend beyond alleviation of medical symptoms and suffering to include aspects that encompass the whole person and the role of work, leisure and recreation in his or her life. Comfort, safety, spirituality and emotional well-being are also not adequately addressed by a health care system that concentrates on attempts to cure disease rather than aid and provide comfort.

This report represents the HD Peer Workgroup’s findings. Its *Recommendations to the Field* issue a call to action to improve care and quality of life for people with HD and their families. The Workgroup advocates that these *Recommendations to the Field* are complemented by needed changes in the national health care policy agenda. These changes include providing medication benefits and in-home therapies that maintain function and provide palliative care for people with HD to the extent possible and desired in their own homes, and providing support and respite for caregivers.

By understanding the scope of problems faced by people with HD and their families, rationally examining new models of health care delivery and redefining the current system, we can enhance palliative and end-of-life care for the person with HD. We can also improve support for families in caregiving – and in grief. And we can educate, counsel and support HD families across generations – those at risk and affected. The *Promoting Excellence in End-of-Life Care* HD Peer Workgroup offers the following *Recommendations to the Field* as a starting point for needed change.





*R*ECOMMENDATIONS TO THE
FIELD: A CALL FOR ACTIONS



The *Promoting Excellence in End-of-Life Care* Huntington's Disease Peer Workgroup calls on all involved with Huntington's Disease (HD) to:

- Foster close connections among people with HD and their families, friends and the community, as a conduit for important emotional support for the patient. As the disease progresses, these connections often become more difficult, while at the same time, all-the-more important.
- Maintain communication with HD-affected people in homebound or long-term care facilities. As the disease progresses, communication difficulties become more profound for the patient, making it increasingly important that others preserve social networks and help HD-affected people make their needs known.
- Screen for and respond to spiritual distress expressed by patients with HD and their families, extending spiritual care to them and supporting their spiritual experience. The potentially changing spiritual experience and needs of people with HD must be met throughout the course of the disease.

Recommendations for Researchers

Despite remarkable research being conducted that is aimed at a cure and reducing symptoms, more encompassing research is needed to better understand the impact of the disease on the lives of patients and their families, appropriate settings to care for patients and the role of clinicians in providing care.

The *Promoting Excellence* Huntington's Disease Peer Workgroup recommends that the National Institutes of Health (NIH) and other researchers:

- Study patients' barriers to adopting advance care plans and advance care directives to improve use and utility of the documents.
- Conduct population-based epidemiological research in HD.
- Identify the demographic features of people with HD, their caregivers, and the types of professionals who care for people with HD, by stage of illness. This data can support development of recommendations for appropriate allocation of health care resources.
- Study caregiver demographics and the relationship between caregiver burden and the clinical features of HD.
- Identify caregiver issues in HD to provide better medical management of people with HD and their families.
- Compare the chronic versus home care settings to identify risk factors for placement in long-term care facilities, demographics of patients and resources required in each setting.
- Conduct interventional studies employing rehabilitative services to provide data about ways rehabilitative services can improve care in HD.
- Study the risk factors for imminent death in order to provide better access to hospice care until the Centers for Medicare and Medicaid Services (CMS) change the six-month life expectancy rule to receive reimbursement for care provided by the Medicare Hospice Benefit (explained in detail in Appendix A, Addendum C on enclosed CD).
- Develop evidence-based guidelines that clinicians can use to assess capacity in arenas such as decision-making abilities.
- Determine the collision rates among drivers with HD to assess traffic safety problems and to examine the consequences of driving cessation.

Recommendations for Public and Private Funders

The *Promoting Excellence* Huntington's Disease Peer Workgroup recommends that public and private funders:

- Expand pharmacy formularies to include all the medications that benefit people with HD.
- Base utilization review and insurance reimbursement (public and private) on individualized assessments performed by qualified clinicians familiar with HD, rather than on general categorical assessments based on other illnesses.
- Support ongoing maintenance and analyses of national HD databases. These large databases of people with HD and their families, maintained by the Huntington Study Group and others, are critically important infrastructure and repositories of data (further explained in Appendix C on enclosed CD).
- Fund interventional studies employing rehabilitative services, evaluating the effects of rehabilitative services on the level of function, symptoms and quality of life for people with HD.

Recommendations for Hospice Programs

The *Promoting Excellence* Huntington's Disease Peer Workgroup recommends that hospice programs:

- Develop assessment criteria for determining the end stage of HD to guide hospice referral and admission until CMS changes the six-month life expectancy rule of the Medicare Hospice Benefit.
- Promote the full use of current eligibility guidelines to extend hospice services to patients with late stage HD and their families.
- Expand their capacity to care for HD patients and families and coordinate the health care and social support services they need.

Recommendations for the Centers for Medicare and Medicaid Services (CMS)

The *Promoting Excellence* Huntington's Disease Peer Workgroup calls on CMS to:

- Change the current six-month life expectancy rule under the Medicare Hospice Benefit so that access to hospice and palliative care by people with end-stage HD is unimpeded. The six-month life expectancy rule is explained in Appendix A, Addendum C on the enclosed CD.
- Offer higher reimbursement rates for Medicaid-funded HD nursing homes to provide patients with better access to quality nursing home care and other programs of comprehensive palliative care.
- Expand home-based care services coverage through Medicaid to enable people to stay at home or to provide alternatives to long-term care placement.
- Expand Medicaid coverage, where necessary, to provide for psychiatric, psychological and behavioral services needed by people with HD in the early and middle stages of the disease.
- Expand Medicaid coverage with a prescription drug benefit to symptomatic adults in the early stages of HD on the basis of a confirmed diagnosis of chronic, progressively degenerative disease rather than on the basis of a financial means test or current level of functional impairment.
- Develop tools for disease management and case management explicitly tailored to HD in order to provide continuity of care and coordination of services.
- Fund respite services and other family-oriented services to family caregivers of people with HD, recognizing that bereavement services are necessary as families grieve the chronic degenerative illness of a loved one well before the person's death.
- Provide coverage for long-term and rehabilitative services with the goal of sustaining func-

tional capacity and independent living to the extent possible.

- Provide psychosocial and behavioral services in the home setting in addition to traditional medical and nursing services, and adapt the team-based, interdisciplinary approach taken in hospice and palliative care near the end of life for use during earlier stages of HD when death is not imminent.

Recommendations for Health Care Practitioners

The *Promoting Excellence* Huntington's Disease Peer Workgroup recommends that physicians, nurses, therapists, social workers, aides and other health care practitioners:

- Discuss advance care directives early in the course of illness when people with HD retain the capacity to execute them. Educate people with HD on the need to develop advance care plans and complete Advance Directives to promote their autonomy and to ensure that care through the end of life is consistent with their values and preferences and those of their families.
- Increase awareness of criteria and protocols for admission to hospice and palliative care systems.
- Help family caregivers obtain devices that ensure patient safety, prevent injury and contribute to activities of daily living. As families cope with the decline in functional mobility and work to prevent injury, they also work to foster independence and maintain a healthy balance between determination to live fully and acceptance of functional decline.
- Treat people with HD in a dignified and caring manner throughout the course of their lives. Even when communication is severely limited or not possible, people with HD continue to understand if they are treated with dignity and respect, as do their families.
- Develop clinical evaluation tools and treatment guidelines for disease management and case

Consider...

Jack led a full and vibrant life. He earned his Ph.D. and two Master's Degrees at a major university and, as a professor, enjoyed influencing the lives of young and old alike with his vibrant, upbeat personality.

Jack's life changed dramatically after his HD diagnosis. Jack felt the loss keenly when he could no longer safely drive his car to work every day or manage the complicated demands of his position. With his autonomy and independence dramatically affected, he fell into a deep depression. With the help of an understanding wife, as well as a qualified and sensitive occupational therapist, Jack learned to utilize public transportation. This adaptation allowed him to maintain his connection with co-workers and continue working in a newly established office arranged by his department for his use.

Returning home from work one day, Jack began to cross the street, but with his slowed gait and poor judgment he did not reach the other side before the light changed, and he was hit by an oncoming car. Although his injuries were minor, he and his family were again faced with changes in his life and lifestyle due to HD.

Jack eventually agreed to attend a local day care center, although he maintained that he did not need anyone watching over him, while his wife worked. His outgoing personality and positive attitude quickly endeared him to the staff and residents, however, and Jack soon became a staff assistant rather than a resident. He helps others with activities and chats with residents and staff. Encouraging and supporting others allows him to maintain his dignity and independence to the greatest extent possible.

management explicitly tailored to HD, and include attention to continuity of care and coordination of services.

- Include quality of life considerations such as contentment and happiness of the person with HD in the scope of clinical interventions.
- Ensure adequate nutrition for people with HD, who often require unusually high caloric intake to maintain body weight.
- Offer counseling and preventive therapy to HD patients and family members as they adapt to and cope with the progression of HD.
- Routinely assess the functional capacity and competent decision-making ability of people with HD.

Recommendations for Educators

The *Promoting Excellence* Huntington's Disease Peer Workgroup calls upon educators in medicine, nursing, social work, physical, occupational and speech therapy to:

- Develop core curricula that include the following: the affective and behavioral manifestations of HD; assessment of capacity to function independently; comfort and quality of life; family issues; and issues of life completion and life closure.
- Teach the importance of advance health care planning and the use of Advance Directives, and the importance for primary health care providers to identify and educate patients about Advance Directives earlier in the course of illness while they retain the capacity to execute them.
- Learn skills to evaluate, select and acquire adaptive equipment, assistive technology and home adaptation designed to enable the person with HD to stay at home for as long as possible. This training must include information about the availability of services for people with HD at various stages.

Recommendations for Professional Organizations

The *Promoting Excellence* Huntington's Disease Peer Workgroup proposes that The American Medical Association (AMA), the American Neurological Association (ANA) and American Society of Human Genetics (ASHG):

- Develop specific training programs for physicians and genetic counselors regarding the implications of HD genetic testing and counseling and involvement of all members of affected patients' families, whenever possible and appropriate.
- Modify the AMA Education for Physicians for End-of-Life Care (explained in Appendix B on enclosed CD), by developing new curricula to address specialized training needs of providers working with people with HD and other neurodegenerative diseases.

Recommendations for National Patient Advocate Organizations

The *Promoting Excellence* Huntington's Disease Peer Workgroup calls upon national patient advocate organizations to:

- Develop and disseminate information that stresses the efficacy of individualized treatment techniques designed to improve quality of life, while recognizing the affected people's right to decline medical intervention.
- Inform caregivers about the need for medical alert bracelets or similar devices.
- Encourage caregivers to enroll their family member in programs such as the *Safe Return Program* of the Alzheimer's Association (<http://www.alz.org/ResourceCenter/Programs/SafeReturn.htm>).
- Provide articles, Web sites and mailings about palliative care that enhance education for families.

Recommendations for The Huntington's Disease Society of America (HDSA) and Other Community HD Resources

The *Promoting Excellence* Huntington's Disease Peer Workgroup recommends that HDSA and other community HD resources:

- Inform affected patients' families – and the public – about HD;
- Develop tools to assess the functional capacity of people with HD. Three relevant sub-domains of functional capacity are social, professional and driving competence (best assessed by a trained driving instructor).
- Develop educational resources about the nature of psychological, spiritual and social suffering of people with HD and their families, as well as the nature of loss and concomitant grief for families dealing with HD as motor and behavioral symptoms increase.
- Expand educational resources regarding the impact of presymptomatic genetic testing on family members.
- Study informational and educational needs of patients and their families, and expand and enhance patient and family resources on the HDSA Web site as needed.
- Connect the person with HD and their family with local resources including HDSA support groups, chapters, Centers of Excellence and community HD clinics. These vital connections preserve the autonomy of people with HD and assist all involved in meeting the challenge of care.

Recommendations for Caregivers

The *Promoting Excellence* Huntington's Disease Peer Workgroup calls on caregivers to:

- Continually reassess and strive to improve levels of comfort. Comfort is more than just alleviation of pain or suffering, encompassing security and safety, and coping with stress and

Consider...

Marguerite is a beautiful woman who, before the onset of HD, enjoyed ballet, singing, modeling in fashion shows, occasional extra film assignments and participating in stage productions. She married at an early age and has two children. Although her husband loves her and is compassionate, the rest of the world has not been quite as sympathetic nor have people in her community understood the nature of her illness and the accompanying personality and behavior changes. In time, her friends avoid her as do her child's school staff, grocery clerks and even medical staff. It becomes clear that her family needs to orchestrate a support group around her:

Since it is no longer safe for Marguerite to drive, the family secures transportation from a county transportation service for the handicapped.

Her aunt invites her to join her church Bible study class and a women's group that has frequent activities throughout the year. These activities encourage meaningful social interaction and Marguerite feels renewed spiritual strength.

Family gatherings also change. Many now take place at Marguerite's home to make it more convenient, to promote her self-esteem and create happy memories for her husband and her children while she is still living at home. Family and friends respond to the family's invitation to help by orchestrating some of these gatherings. As Marguerite's abilities decline, family and friends adapt tasks and activities to meet her abilities.

These subtle but powerful changes produce a much happier and content person as Marguerite realizes self-worth and happiness. Being appreciated by others reaffirms her as a person. The quality of her life does not decline although it is altered to accommodate the changes brought on by Huntington's Disease.

the feeling of contentment. As HD advances, stress imparted by the progressive loss of abilities and independence leads to great discomfort across many aspects of a person's life.

- Pay heightened attention to the safety needs of people with HD.

Recommendations for Policy-Makers

The *Promoting Excellence* Huntington's Disease Peer Workgroup calls on policy-makers to:

- Visit long-term care facilities and spend time interacting with residents and staff in order to better understand unmet needs within long-term care facilities.
- Require insurance underwriters to create policies and administrative guidelines that enable and encourage people with HD to work at whatever level they are capable of performing without sacrificing needed benefits and services.
- Preserve federal and state insurance benefits for people with HD when disease renders them no longer employable.
- Enhance prescription drug and care benefits and expand home-based services helping people with HD remain at home for as long as comfortably possible.
- Fund formal demonstration projects that build upon the recommendations in this report and on integrated, life prolonging and palliative care models in other settings.
- Enact legislation that states and national health care providers and organizations adopt training and reference tools regarding recognition of the signs and symptoms of HD.

Recommendations for First Responders

The *Promoting Excellence* Huntington's Disease Peer Workgroup recommends that Emergency Medical

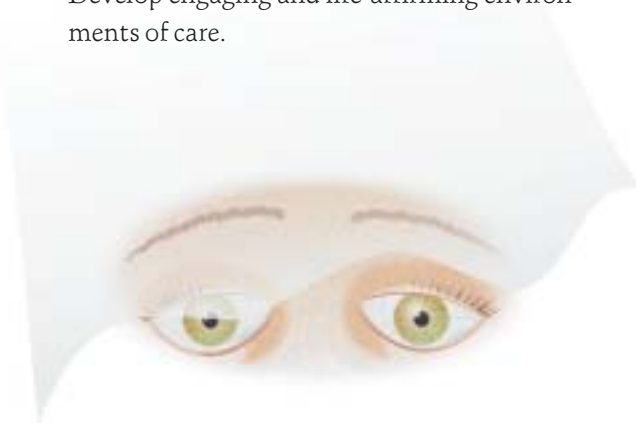
Technician (EMT) personnel, rescue teams, fire and law enforcement and other first responders:

- Develop and enhance educational tools regarding recognition of the signs and symptoms of HD, with training and reference tools adopted by state as well as by national health care providers and organizations.
- Carry a *Medical Quick Reference Card* that would enable first responders to question whether aberrant behavior is due to mental or organic illness rather than intoxication or illicit drugs.

Recommendations for Health Care Delivery Systems

The *Promoting Excellence* Huntington's Disease Peer Workgroup calls upon health care delivery systems to:

- Promote models of care to enhance participation in the full spectrum of daily living, including work, leisure and recreation.
- Design coverage for long-term and rehabilitative services using the goal of sustaining as much functional capacity and independent living as possible, given the progression of the disease and the individual's level of functional impairment.
- Provide respite services and other family-oriented services to family caregivers of people with HD, recognizing that bereavement services are necessary as families experience the chronic degenerative illness of a loved one, well before the time of death.
- Develop engaging and life-affirming environments of care.



Consider...

Mandy is a young lady with Huntington's Disease. Although she is only 25 years of age, she began showing symptoms at the age of 11. After a formal diagnosis at the age of 17, her attitude was positive. She stayed on top of the latest HD research and dealt with her limitations. She participated in whatever she could and enjoyed a peaceful environment created to accommodate her lifestyle and needs. She participated in Karaoke with the family as backup singers, and she danced from a wheelchair with the help of family and friends.

Mandy is now at Stage IV of Huntington's Disease. Her family foresees a transition to the next level of the disease in the near future. Although her speech is difficult to understand, Mandy can still communicate her needs and wishes. When the family misinterprets what she is trying to communicate, they assume a loving and playful attitude in trying to decipher her words rather than expressing frustration or hopelessness. Mandy is usually the first one to start laughing at their hilarious interpretations! She currently receives speech therapy. The family is now looking ahead to when even limited communication will be impossible. They have contacted a computer company that offers equipment to assist disabled people with communication needs.

The family has talked about Advance Directives. They have contacted an elder law attorney to draft the necessary documents. This has all been done in a positive and natural course of life spirit. Mandy would like to be placed in an assisted living facility when a home setting is no longer adequate for the care that she requires. She feels she will be able to maintain a greater sense of dignity and care in a professional care setting.

The family has, as a group, selected burial plots and discussed the funeral arrangements they would like to have. Mandy, being of young heart, communicated her desire to wear a red, fitted, spaghetti strap dress for her funeral. Her mother assured her that she would get her dress, and the entire topic became a festive discussion, such as planning a party.

One of the most comforting and reassuring communications from family and friends to Mandy is the knowledge that she is loved and that she will be well taken care of and never left alone.

Summary

Huntington's Disease has devastating physical, emotional, cognitive, social and financial consequences. Most individuals affected by the disease leave the workforce early, become ineffective parents and partners, consume medical resources and end their lives in long-term care facilities. Families are exhausted by the overlapping intergenerational care needs caused by the hereditary nature of HD. Improving care at the end of life requires in part a thorough understanding of how and where people with HD spend their final years, who provides care, what the primary sources of funding for care are and what pertinent policies affect care.

Now is the time to call for formal demonstration projects that build upon the recommendations in this report and on integrated concurrent life-prolonging and palliative care models in a variety of health care settings. Demonstration projects are needed that integrate cutting-edge neurological care with hospice and palliative care in a concurrent manner. New best practice models for completing the continuum of care for HD patients and their families must be developed. Recognition and achievement of these recommendations will not only dramatically improve the lives of individuals living with Huntington's Disease, but also those of people with other neurodegenerative diseases such as multiple sclerosis, Parkinson's disease, Alzheimer's disease and stroke.

¹ Di Maio L, Squitieri F, Napolitano G, Campanella G, Trofater JA, Conneally PM. "Onset symptoms in 510 patients with Huntington's disease." *Journal of Medical Genetics*, 30(4): 289-292, 1993.

² Penney JB, Jr, Young AB, Shoulson I, Starosta-Rubenstein S, Snodgrass SR, Sanchez-Ramos J, et al. "Huntington's disease in Venezuela: 7 years of follow-up on symptomatic and asymptomatic individuals." *Movement Disorders*, 5(2): 93-99, 1990.

- ³ Brandt J, Bylsma FW, Gross R, Stine O, Ranen N, Ross CA. "Trinucleotide repeat length and clinical progression in Huntington's Disease." *Neurology*, 46: 527-531, 1996.
- ⁴ Kiebertz K, MacDonald M, Shih C, Feigin A, Steinberg K, Bordwell K, et al. "Trinucleotide repeat length and progression of illness in Huntington's disease." *Journal of Medical Genetics*, 31(11): 872-874, 1994.
- ⁵ Illarioshkin SN, Igarashi S, Onodera O, Markova ED, Nikolskaya NN, Tanaka H, et al. "Trinucleotide repeat length and rate of progression of Huntington's disease." *Annals of Neurology*, 36(4): 630-635, 1994.
- ⁶ Maat-Kievit A, Losekoot M, Van Den Boer-Van Den Berg H, Van Ommen GJ, Niermeijer M, Breuning M, et al. "New problems in testing for Huntington's disease: The issue of intermediate and reduced penetrance alleles." *Journal of Medical Genetics*, 38(4): E12, 2001.
- ⁷ McCusker EA, Casse RF, Graham SJ, Williams DB, Lazarus R. "Prevalence of Huntington disease in New South Wales in 1996." *The Medical Journal of Australia*, 173(4): 187-190, 2000.
- ⁸ Almqvist EW, Elterman DS, MacLeod PM, Hayden MR. "High incidence rate and absent family histories in one quarter of patients newly diagnosed with Huntington disease in British Columbia." *Clinical Genetics*, 60(3): 198-205, 2001.
- ⁹ Falush D, Almqvist EW, Brinkmann RR, Iwasa Y, Hayden MR. "Measurement of mutational flow implies both a high new-mutation rate for Huntington disease and substantial underascertainment of late-onset cases." *American Journal of Human Genetics*, 68(2): 373-385, 2001.
- ¹⁰ *Financial Implications of Promoting Excellence in End-of-Life Care*. Missoula, Mont.: *Promoting Excellence in End-of-Life Care*, a national program of The Robert Wood Johnson Foundation, 2002.
- ¹¹ Shoulson I. "Huntington's disease: Functional capacities in patients treated with neuroleptic and antidepressant drugs." *Neurology*, 31: 1333-1335, 1981.
- ¹² Shoulson I, Fahn S. "Huntington disease: Clinical care and evaluation." *Neurology*, 29(1): 1-3, 1979.



HDSA Centers of Excellence:

please note that all contact information was accurate at time of printing.

NORTHEAST

New England HDSA Center of Excellence

Charlestown, Mass. 02129

Director: Steven Hersch, M.D., Ph.D.

Center Contact: Yoshio Kaneko

Phone: 617/724-2227

Email: ykaneko@partners.org

HDSA Center of Excellence at the University of Rochester

Rochester, N.Y. 14620

Directors: Ira Shoulson, M.D.; Peter Como, Ph.D.;

Karl Kiebertz, M.D.

Center Contact: Leslie Briner

Phone: 585/273-4147

Email: leslie.briner@ctcc.rochester.edu

HDSA Center of Excellence at Columbia Health Sciences/NYS Psychiatric Institute

New York, N.Y. 10032

Director: Karen Marder, M.D.

Center Contact: Deborah Zeck Thorne, C.S.W.

Phone: 212/305-9172

George G. Powell HDSA Center of Excellence at North Shore University Hospital

Manhasset, N.Y. 11030

Director: Andrew Feigin, M.D.

Center Contact: Mary Ellen Benisatto

Phone: 516/869-9527

HDSA Center of Excellence at Johns Hopkins University/Johns Hopkins Hospital

Baltimore, Md. 21287

Directors: Christopher Ross, M.D., Ph.D.

Adam Rosenblatt, M.D.

Center Contact: Debbie Pollard

Phone: 617/955-2398

Email: dpollard@jhmi.edu

HDSA Center of Excellence at the University of Virginia

Charlottesville, Va. 22908

Director: Madaline Harrison, M.D.

Center Contact: Pat Allinson, M.S.

Phone: 434/924-2665

Email: psa9d@virginia.edu

SOUTHEAST

HDSA Center of Excellence at Emory School of Medicine

Atlanta, Ga. 30329

Directors: Randi Jones, Ph.D.

J. Timothy Greenamyre, M.D., Ph.D.

Center Contact: Joan Harrison, G.N.P.

Phone: 404/728-6364

Email: jharri2@emory.edu

HDSA Center of Excellence at University of Alabama

Birmingham, Ala. 35233

Director: Leon Dure, M.D.

Center Contact: Stacey Mantooth

Phone: 205/996-7850

Email: smantooth@peds.uab.edu

MIDWEST

HDSA Center of Excellence at Ohio State University

Columbus, Ohio 43210

Director: Sandra Kostyk, M.D., Ph.D.

Center Contact: Nonna Stepanov

Phone: 614/688-8672

Email: stepanov-1@medctr.osu.edu

HDSA Center of Excellence at University of Iowa Hospitals and Clinics

Iowa City, Iowa 52242

Directors: Jane Paulsen, Ph.D.; Henry Paulson, M.D.;

Robert Rodnitsky, M.D.

Center Contact: Anne Leserman

Phone: 319/353-4307

Email: hdinfo@uiowa.edu

**HDSA Center of Excellence at
Hennepin County Medical Center**

Minneapolis, Minn. 55415

Director: Martha Nance, M.D.

Center Contact: Beryl Westphal, APRN

Phone: 612/873-60243 or 612/873-2595

**HDSA Center of Excellence at
Washington University School of
Medicine**

St. Louis, Mo. 63110

Director: Joel Perlmutter, M.D.

Center Contact: Melinda Kavanaugh, M.S.W.

Phone: 314/362-3471

Email: kavanaughm@neuro.wustl.edu

WEST AND PACIFIC NORTHWEST

**HDSA Center of Excellence at
Colorado Neurological Institute**

Wheat Ridge, Colo. 80033

Director: Lauren Seeberger, M.D.

Center Contact: LeAnn Hansen, LCSW

Phone: 720/371-5806

Email: leannahansen@hotmail.com

**HDSA Center of Excellence at
University of Washington**

Seattle, Wash. 98195

Director: Thomas Bird, M.D.

Center Contact: Debbie Olsen (general info)

Phone: 206/616-2135

SOUTHWEST

**HDSA Center of Excellence at
University of California
Davis Medical Center**

Sacramento, Calif. 95817

Directors: Vicki Wheelock, M.D.;

Teresa Tempkin, R.N.C., M.R.N., A.N.P.

Center Contact: Teresa Tempkin

Phone: 916/734-6278

Email: teresa.tempkin@ucdmc.ucdavis.edu

**HDSA Center of Excellence at
University of California**

San Diego, Calif. 92093

Directors: Jody Corey-Bloom, M.D., Ph.D.;

Joanne Hamilton, Ph.D.

Center Contact: Jody Goldstein

Phone: 858/622-5854

Email: jlgoldstein@ucsd.edu

**HDSA Center of Excellence at
Baylor College of Medicine**

Houston, Texas 77030

Director: Joseph Jankovic, M.D.

Center Contact: Christine Hunter, R.N., CCRC

Phone: 713/798-3951

Email: chunter@bcm.tmc.edu

Huntington's Disease Peer Workgroup Members

Richard Dubinsky, M.D. (Chair)

Associate Professor
Department of Neurology
University of Kansas Medical Center
Kansas City, Kan.

Tetsuo Ashizawa, M.D.

Professor and Chair
Department of Neurology
The University of Texas Medical Branch
Galveston, Texas

Gary Barg

Editor in Chief, Today's Caregiver
Caregiver Media Group
Hollywood, Fla.

H. Taylor Butler, LCSW

HDSA Center of Excellence
Emory University School of Medicine
Wesley Woods Health Center
Atlanta, Ga.

Ira Byock, M.D.

Director
Promoting Excellence in End-of-Life Care
Missoula, Mont.

Carol Maier Clerico, O.T./L.

Therapy Education Coordinator
University of Virginia Health System
Huntington's Disease Center of Excellence
Charlottesville, Va.

Peter G. Como, Ph.D.

Co-Director, HDSA Center of Excellence
University of Rochester Medical Center
Rochester, N.Y.

D. Brookes Cowan, Ph.D., M.S.W.

Department of Sociology
University of Vermont
Burlington, Vt.

Charles Diggs, Ph.D. (ASHA)

Director, State and Consumer Advocacy
American Speech-Language-Hearing Association
Rockville, Md.

Barbara Heiman, M.S.W./LISW

HDSA Center of Excellence
The Ohio State University
Columbus, Ohio

Bruce Jennings, M.A.

Senior Research Scholar
The Hastings Center
Garrison, N.Y.

Marilee Monnot, Ph.D.

Assistant Professor
Department of Neurology
Oklahoma University
Health Sciences Center
Oklahoma City, Okla.

Martha Nance, M.D.

Director of HDSA Center of Excellence
Hennepin County Medical Center
Minneapolis, Minn.

Frances Diaz Saldaña

Caregiver
Fountain Valley, Calif.

Kathleen Shannon, M.D.

Associate Professor
Department of Neurological Sciences
Rush Medical College
Senior Attending Physician
Rush-St. Luke's Presbyterian Medical Center
Chicago, Ill.

Teresa Tempkin R.N.C., M.S.N., A.N.P.

HDSA Center of Excellence
University of California,
Davis Medical Center
Department of Neurology
Sacramento, Calif.

Susan Jo Bumagin, M.Ed.

Peer Workgroup Director

The case scenarios provided in this report are based on real life stories of people with Huntington's Disease.

Visit the comprehensive Web site of *Promoting Excellence in End-of-Life Care* for more information on innovative demonstration projects dedicated to long-term changes to improve health care for dying people and their families: <http://www.promotingexcellence.org> or contact:

Ira Byock, M.D., Director
Promoting Excellence in End-of-Life Care
The University of Montana
1000 E. Beckwith, Missoula, MT 59812
Phone: 406/243-6601
Email: ibyock@aol.com

Jeanne Sheils Twohig, M.P.A., Deputy Director
Promoting Excellence in End-of-Life Care
The University of Montana
1000 E. Beckwith, Missoula, MT 59812
Phone: 406/243-6602
Email: TwohigJS@mso.umt.edu

APPENDIX CD-ROM TABLE OF CONTENTS

1	APPENDIX A: Care Subcommittee Report <i>This subcommittee report discusses 10 domains in three different settings: home, long-term care facility and hospital. The report defines the domain of care, presents ideal and worst case scenarios and suggests solutions and resources. Vignettes illustrate key points in each domain.</i>
3	Autonomy
8	Dignity
16	Meaningful Social Interaction
21	Communication
28	Comfort
33	Safety and Order
39	Spirituality
42	Enjoyment, Entertainment and Well-Being
46	Nutrition
52	Functional Competence
59	ADDENDUM A: Advance Directives
61	ADDENDUM B: Euthanasia and Physician-Assisted Suicide
62	ADDENDUM C: Hospice Care in Relation to the HD Patient
66	ADDENDUM D: Policy
67	ADDENDUM E: Values History Form
71	APPENDIX B: Education Subcommittee Report
84	APPENDIX C: Research and Policy Subcommittee Report
127	Research Resources
129	APPENDIX BIBLIOGRAPHIES AND RESOURCES