

Appendix C

Research & Policy Subcommittee Report

Introduction

The progression of HD has been divided into five stages defined by the person with HD's score on the Total Functional Capacity Scale, which defines level of function in the workplace, with finances and domestic chores, as well as activities of daily living and requirements for unskilled or skilled care. (1, 2)

	<i>Engagement in occupation</i>	<i>Capacity to handle financial affairs</i>	<i>Capacity to manage domestic responsibilities</i>	<i>Capacity to perform activities of daily living</i>	<i>Care can be provided at...</i>
Stage I (TFC 11-13) (0-8 y)	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

Course, Duration and Cause of Death in HD

The mean age at onset for HD ranges from 30 to 45. In many cases, the age at onset falls outside this range, and onset at the extremes of age has also been reported. In individual cases, onset may be difficult to pinpoint, particularly when behavioral and psychiatric changes are considered. In a review of the nature of onset of HD symptoms in 510 HD patients, Di Maio et al. found that neurological symptoms suggested the disease onset in

59 percent of subjects, while 23 percent had psychiatric and 18 percent had onset with both neurologic and psychiatric symptoms. (3) In people with HD who are closely followed, there appears to be a zone of onset spanning 3 to 5 years. Studies of motor onset in the Lake Maracaibo, Venezuela, where HD is endemic, showed that the first clinical development is subtle changes in volitional eye movement and clumsiness, before frank motor changes of the disease. Chorea develops, but then over the course of years, people become more dystonic and akinetic, eventually becoming bed bound. (4) Juvenile HD has a strikingly different phenotype, beginning with akinesia, rigidity and dystonia and often accompanied by myoclonus or seizures. The progression of HD occurs independent of any therapeutic intervention. It remains unclear whether progression rates relate to the CAG repeat length, thus limiting the prognostic value of genetic testing for disease severity and progression. (5-7)

In a survey of first-degree relatives of people with HD, respondents specified 19 physical, cognitive and emotional HD symptoms and when they occurred. The survey included 1,238 individuals with at least six years of HD symptoms. (8) Involuntary movements and mood and personality changes appeared most commonly in the first five years of the illness. Gait and balance disorders and cognitive dysfunction became more problematic around the five-year mark. Later disease was characterized by poor communication, weight loss and loss of control of bodily functions.

In the end-stages of HD, immobility is profound and death is generally due to complications including bronchopneumonia and heart disease. Nutritional deficiencies,

mental disorders, cerebrovascular disorders and accidents are also reported. Other rare causes of death include suicide and violence. (9, 10) An analysis of 452 deceased HD subjects and 831 living subjects from the HD National Research Roster indicated that although almost 30 percent of people with HD attempt suicide at least once, it is the cause of death in only 5.7 percent. Unlike other causes of death in HD, suicide attempts often occur during the early stages of the disease. (11) The average duration of HD at death is about 17 years; in the more severe juvenile form of the illness, survival averages 10 years.

Epidemiology of HD

The prevalence of HD has been estimated between 3 and 10/100,000 in people of European descent. (12, 13)

Geographic region	<i>Prevalence</i>
New South Wales (McCusker, 2000)	6.3/100,000
West of Scotland (Bolt, 1970)	5.2/100,000
Mauritius (Hayden, 1981)	4.6/100,000
Grampian (Simpson, 1989)	9.94/100,000
Ireland (Morrison, 1995)	6.4/100,000
Hong Kong (Chang, 1994)	3.7/1,000,000
South Africa (Hayden, 1980)	0.0/1,000,000

Most people with HD are identified by the presence of a suggestive neurologic course in a person with a positive family history. However, in a study of symptomatic subjects undergoing confirmatory genetic tests in British Columbia, nearly 25 percent had negative family histories. Although in many cases, the family history is obscured by misdiagnosis, obfuscation or competitive mortality, it has been estimated that as many as 8 percent of people with HD with negative family histories may reflect new genetic

mutations arising from borderline premutations. (14) Moreover, the study of HD likely suffers from under-reporting of cases. Studies of mutational flow suggest that verification is poor with late onset of HD. (15) Based on current epidemiologic data, it is estimated that there are 25,000 to 30,000 people with HD in the United States, and that there are on average five first-degree relatives at risk for each affected person.

Genetics of HD

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 huntingtin gene normally has fewer than 30 CAG repeats. When there are 40 or more CAG repeats, HD is fully penetrant. When there are 36 to 40 CAG repeats, the disease is less than fully penetrant in the carrier, but a full-length mutation may be passed to the next generation where HD will appear. When there are 30-35 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.

A direct DNA test for HD has been available since 1993, potentially allowing for almost total determination of those destined to develop the disease. However, the lack of an effective treatment for HD, the inability to predict the onset or course of the disorder in gene positive presymptomatic people and concerns about confidentiality and discrimination have conspired to limit its usefulness. (16-19)

Repercussions of HD in Society and Medicine

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.

Defining End of Life in HD and Areas of Subcommittee Focus

For the purposes of the work of the Research Subcommittee, the operational definition of *end of life* in HD includes two triggers: the request by a person with HD or family for end-of-life information or interventions, or the onset of loss of independence as defined by:

- Awareness of impending mortality by the person with HD or their family;
- Impaired ability to care for self;
- Inability to live in the community with support;
- Dysphagia, where nutrition is impaired or high risk for aspiration; and
- Impaired emotional ability to cope.

For the Research Subcommittee, the particular areas of focus are:

1. Search of current epidemiology to answer the following questions:

- What is the prevalence of manifest HD by Stage?
- What are the demographic features of people with HD by Stage?
- What are the demographic features of HD caregivers by Stage?
- What types of professionals care for people with HD by Stage?
- Where is care provided for people with HD by Stage?
- Where do people with HD die?
- Do people with HD have Advance Directives? What features of the illness predict the use of Advance Directives by Stage?

2. Search of current policy to answer the following questions:

- What is covered by private versus public funds by Stage?
- What assistance is available by Stage?
- How might policy impede the use of Advance Directives for people with HD and surrogates?
- How does the current definition of disability affect people with HD?
- How does public policy interfere with delivery of care by Stage?
- What special considerations affect juveniles with HD?

Search of Current Epidemiology Literature

A precise determination of the epidemiology of HD has been limited by the lack of pivotal studies. Epidemiological studies are by definition labor intensive and costly. The existing epidemiological literature is hampered by a number of methodological limitations and represents a major unmet research need. The largest available dataset regarding specific prevalence and demographic data in HD has been amassed by the Huntington Study Group (HSG).

The HSG was established in 1993 and is a non-profit group of physicians and other health care providers from medical centers in the United States, Canada, Europe and Australia experienced in the care of people with HD and dedicated to clinical research of Huntington's Disease. Since its inception, the HSG has focused on the following: systemic collection of clinical information on individuals with both manifest HD and presymptomatic people at-risk for HD; development and initiation of therapeutic trials aimed at slowing functional decline in HD; and development and initiation of observational trials in at-risk individuals to determine the earliest clinical and biological markers of disease onset.

A primary clinical tool of the HSG is the Unified Huntington's Disease Rating Scale (UHDRS), which was developed by the HSG as a research tool to provide a uniform assessment of the clinical features (motor, cognitive, behavioral and functional) and course of HD. The UHDRS has undergone extensive reliability and validity testing and has been used as a major outcome measure by the HSG in controlled clinical trials. The current UHDRS database numbers more than 6,000 people affected by or immediately at-risk for HD (out of a population of more than 200,000 manifest and at-risk individuals). A cross-sectional and prospective analysis of the database of the UHDRS instrument was published in *Movement Disorders*. (20) In a large cohort, using the UHDRS natural history database, the rate of functional decline was evaluated over a maximum of four years of observation. A full report was published in *Neurology*. (21) While the UHDRS natural history database represents a significant contribution to knowledge regarding the epidemiology of HD, its main limitation is a recruitment bias of those individuals who

present to HSG clinical sites. These sites may not capture the full population of individuals either with manifest HD or with an immediate genetic risk for HD.

In addition to the HSG database, several centers specializing in HD, notably at Indiana University and Johns Hopkins University, have been systematically investigating people with HD and families for several years. These latter databases have focused extensively on presymptomatic individuals although they have longitudinally followed individuals with manifest HD. The Indiana HD roster numbers over 2,500 individuals. (22)

What is the Prevalence of Manifest HD by Disease Stage?

Accurate estimates of the prevalence of manifest HD by stage have been limited by small sample sizes in various clinical studies. The UHDRS natural history database offers the largest available prevalence data to date. However, even the UHDRS database includes a disproportionate distribution of subjects by stage of disease. Based upon data supplied by Marder et al. (21) the current prevalence and duration of illness based upon the Total Functional Capacity (TFC) score in manifest HD is depicted below:

TFC Stage	Mean Disease Duration (yrs)	Percent of UHDRS Database Subjects
Stage I	4.7	23 %
Stage II	6.5	38 %
Stage III	9.1	29 %
Stage IV	10.9	8 %
Stage V	13.2	2 %

It is clear from this table that the majority of people with HD in this database represent individuals with early to mid-stage disease. There is a considerable drop off in this database for subjects with late- and end-stage HD. Several factors may explain this marked drop in the database. The most likely explanation is an access to care problem for

people with late-stage HD and families. Given the clinical severity of their HD in these stages, most people with HD have limited motor, cognitive and functional capacity, and are likely being cared for in long-term care facilities or institutions.

Nance and colleagues (23) conducted a retrospective review of 97 people with HD living in long-term care facilities in Minneapolis and St. Paul. The purpose of the study was to describe the demographic features, patterns of behavior, weight change, nursing issues, medication use and hospitalization in this population. On admission to the long-term care facility, the average person with HD was of either gender, 45 years old, previously employed, a high school graduate and not married. One third had severe behavior problems. Half gained and half lost weight but weight loss was not a predictor of death. Almost all were prescribed central nervous system-active drugs, most commonly neuroleptics. Eighty-four percent were ambulatory on admission, but 88 percent were non-ambulatory at the time of death. This study suggested the following: 1) HD residents of long-term care facilities are demographically different from other residents; 2) weight gain can occur in some people with late-stage HD; 3) negative behavior is a significant problem but is restricted to a subset; and 4) adapting creatively to increasing multifaceted disability is the greatest challenge to the staff in facilities caring for people with HD.

Given the lack of available treatment for HD in general, and late-stage HD in particular, it is not surprising that people with HD are often cared for by health care providers who work directly at long-term care facilities. A recent initiative of the Huntington's Disease Society of America (HDSA) has been the development of HDSA Centers of Excellence

within the United States. These Centers of Excellence and other Huntington's Study Group clinical sites are involved in the performance of outreach clinical care and education particularly at long-term care facilities, suggesting the possibility that a higher proportion of people with end-stage HD can be systematically evaluated with the UHDRS and entered into the HSG natural history database. A limitation of this effort is the funding available to these clinical sites to perform their clinical and research mandates.

What are the Demographic Features of People with HD by Stage?

To date, systematic analyses of demographic features of people with HD by stage of illness have not been published. A comprehensive Medline review failed to yield specific demographic information for HD by stage of illness. This clearly represents an unmet need in HD research. The published studies utilizing the UHDRS database have only presented group baseline demographic information. For example, data published by Marder and colleagues (21) in their sample of 960 subjects with HD report mean demographic information on age (48.1 ± 12.7 yrs), gender (48 percent male, 52 percent female), age at onset of HD (40.8 ± 12.4 yrs), education (12.9 ± 3.3 yrs), and race (95 percent white, 2.5 percent black, 1.8 percent Hispanic, 0.7 percent other). This study did not break down these demographic data by stage of illness.

Future research studies clearly need to address this deficiency in HD populations. In addition to the clinical variables yielded by the UHDRS, the HSG natural history database does include medical history and demographic information collected at the time

of the initial visit. To date, there has been a lack of funding to complete the analyses that would link key demographic features by stage of illness.

What are the Demographic Features of HD Caregivers by Stage?

An even larger unmet research need is the lack of information regarding caregiver burden in HD. There is extensive literature investigating caregiver burden in other neurodegenerative illnesses, particularly Alzheimer's-type dementia. However, review of the literature failed to identify specific studies investigating the demographic features of HD caregivers. Of the limited available studies, most have focused on relative costs to caregivers. Murman and colleagues (24) conducted a survey of health care utilization in three dementia syndromes to determine whether type of dementia influenced utilization or resulting direct costs. Subjects with Alzheimer's disease (n=131), dementia with parkinsonism (n=85), and Huntington's Disease (n=51) were identified from a registry and enrolled in the study. Caregivers completed the mailed survey and direct costs were estimated. Interestingly, the presence of dementia with parkinsonism resulted in significantly greater utilization of long-term care services and higher total direct costs. However, in all three groups, long-term care costs accounted for the majority of direct costs.

A presentation by Chesire and colleagues at the World Federation of Neurology/International Huntington Association meeting (August 2001) investigated caregiver burden in HD. Based upon the lack of any published studies on this subject, these authors researched these three areas: the degree of caregiver burden in HD; the relationships among caregiver burden and caregiving activities (e.g., time spent

delivering care); and 3) the relationships among the core clinical features of HD and caregiver burden in 45 people with manifest HD. Caregivers in this study were defined as individuals who provided unpaid assistance to a person with HD. Caregivers were asked to measure their burden by using various validated instruments and by a psychosocial assessment that was administered by a social worker.

Caregiver demographics of this sample included 28 females and 17 males with a mean age of 53 ± 14.5 years. The mean total burden, on a caregiver burden inventory, reflected a moderate level of burden. However, the participants in this sample consisted primarily of early to mid-stage HD. There were no Stage V subjects in this small study. No relationship was found between the total caregiver burden score and caregiver gender, age, education or number of years delivering care. However, the number of hours per week and the amount of caregiver-perceived stress significantly correlated with the level of caregiver burden. In addition, there were significant correlations between caregiver burden and the motor, cognitive and functional ability of the index HD cases. All correlations indicated a higher level of caregiver burden when greater motor, cognitive and functional disability were observed in the HD subject. This small pilot study suggests that caregiver burden in HD warrants further investigation using a much larger sample size across all stages of the disease. A possible hypothesis that warrants further exploration is the possibility that caregiver burden might decline by stage of illness possibly due to greater placement of people with late and end-stage HD in long-term care facilities.

What Types of Professionals Care for People with HD by Stage?

Information regarding the types of professionals who care for people with HD by stage of illness is largely anecdotal and principally based upon the expertise level of HD centers. The designated HDSA Centers of Excellence require an interdisciplinary team approach to HD care. Thus, all HDSA Centers of Excellence include neurologists, neuropsychologists or psychiatrists, social workers, nurse practitioners and nurses. Most Centers also have consulting agreements with a variety of other health care professionals, including speech, physical and occupational therapists. In rare cases, individuals with legal backgrounds are available to assist people with HD and their families. There is no research data regarding type(s) of health care professional by stage of illness. However, using the HDSA Center of Excellence model, some general assumptions can be inferred from the table below:

	Presymptomatic DNA Testing	Stage I	Stage II	Stage III	Stage IV	Stage V
Neurologist	X	X	X	X	X	
Neuropsychologist/ Psychiatrist	X	X	X	X	X	
Social Worker		X	X	X	X	X
Nurse Practitioner/Nurse		X	X	X	X	X
Speech/OT/PT, etc.				X	X	

Review of this model would suggest that the so-called “core” medical specialties of neurology and neuropsychology/psychiatry play a prominent role in the presymptomatic to beginning late-stage HD, and reflect the need for accurate diagnostic evaluation as well as the identification and treatment of symptoms that contribute to disability in HD (e.g., depression, anxiety, cognitive dysfunction).

As people with HD advance in stage of illness, greater emphasis on disability planning, long-term placement and financial assistance may predominate the clinical picture suggesting that social workers are a critical element of HD centers. The roles of neurology and neuropsychiatry tend to diminish with advancing illness as the physical progression of the disease (e.g., speech/swallowing problems, respiratory illness, vulnerability to infection, etc.) begins to predominate the clinical picture. Accordingly, specialists in speech, respiratory care and physical medicine are required. Many people with HD are placed in long-term facilities in the latter stages of illness and tend to be cared for by long-term care specialists employed by these facilities.

Where is Care Provided for People with HD by Stage?

A literature search failed to reveal data on care settings in the early to middle stages of Huntington's Disease. However, since it is a chronic illness with a slow onset and progression, the majority of people with early stage HD are cared for in the community, living independently or being cared for by family members. As the disease progresses, some people with HD may continue to live at home, being cared for by family, with or without community support, home care services, or rarely, hospice care. During the final stages of the illness, however, most are cared for in institutional settings such as nursing homes.

Studies mainly outside the U.S. have found that 16 to 30 percent of people with HD are in long-term care at any time and that 58 to 88 percent of people with HD die in long-term care facilities. (25, 26, 27-29)

People with HD are younger and less likely to ever return to community-based care than the average long-term care facility resident. In a review by Nance and Sanders of 97 HD residents in long-term care facilities from 1978 to 1993, 51 were men and 46 were women. Their average age was 44.6 years; institutionalized men were somewhat younger than women (41.7 versus 47.8). Twelve were younger than 30 and 14 were older than 60. The average disease duration was 10 years, similar to a study of institutionalized people with HD in the UK. (30) Due to the progressive nature of the illness, their care needs evolved over time. Although most were ambulatory (84 percent) at the time of admission, most became non-ambulatory prior to death as the disease advanced. For people with earlier stage HD, the main problems dictating care are behavioral, including smoking, unwillingness to bathe and sexual misconduct. Drugs are commonly used to control mood or behavior. Eighty-four percent took a regular dose of a neuroleptic at some time during their stay in the long-term care facility. Sixty-five percent used an anxiolytic and 46 percent were treated with antidepressants. People with more advanced HD are troubled by regurgitation, aspiration and infections. Eleven percent of the HD subjects had gastrostomy feeding tubes. Feeding tube placement was permanent in over 80 percent and all those with permanent feeding tube placements died over the course of the study. The main indications for acute hospitalization were psychiatric, but admissions for problems related to malnutrition and infection were also common. (23)

The reasons for admission from home to a long-term care facility were lack of a caregiver and the inability of the caregiver to continue to provide care. Among those transferred from other long-term care facilities, the primary reason for transfer was unmanageable

behavior. Once admitted, return to a less structured care environment or home did not occur. (23) These represent long-stay HD residents, with death occurring after two to eight years of institutionalization. (23, 25, 26, 30, 31) Unfortunately, more detailed data on the risk factors for the placement of people with HD in long-term care facilities are not available. Caregiver burden and depression have been identified as causes of early discontinuation of care for elderly disabled Alzheimer’s Disease (AD) patients by caregivers in Japan. (32, 33) The ability to generalize from AD data to HD is problematic, because people with HD are younger at placement in long-term care facilities than other demented patients. However, a better understanding of the risk factors for placement might allow more people with HD to spend the terminal phases of the illness in the community or with family.

Because of the tendency for problem behavior in HD, it is likely that at least a small number of people with HD reside in correctional institutions. However, no epidemiologic data are available on this population.

The table below summarizes the likely care settings for people with HD.

Stage	Home alone	Home, partial support	Home care or hospice	Nursing Home	Incarcerated
I	X	X			X
II	X	X			X
III		X	X	X	X
IV			X	X	X
V			X	X	

Where do People with HD Die?

The majority of people with HD die of illnesses secondary to the disability with impaired mobility. Thus, these deaths are likely to occur in people with HD in the later stages of the disease and the likely settings for these deaths are acute care hospitals, long-term care facilities or at home with full-time caregiving. About 5 to 7 percent commit suicide.

These deaths most likely occur in the earlier stages of HD and may occur in an independent living setting or in people with HD who reside with family or other caregivers. (34, 35) Accidental deaths through motor vehicle collisions or falls are likely to affect people with HD in Stages II through IV.

There are no published studies addressing the stage-related cause of death or the setting of death by illness stage. It is important to recognize that studies of death in HD are confounded by the low ascertainment rate of the disease based on death certificates. In a study published in 1986, only 66 percent of HD death certificates listed HD as the primary cause of death or as a contributing cause of death. While the availability of genetic testing since that time is likely to have improved diagnostic accuracy, omissions of the illness from death certificates likely occurs with some frequency. (10) An analysis of mortality in a group of 10 large kindreds with HD suggested a standardized mortality ratio of 1.5 (95 percent confidence interval 1.4-1.7). The excess mortality occurred between 40 and 70 years of age, as would be expected based on the typical age at onset and duration of HD. In the Nance study of nursing home residents in Minnesota, the average HD resident was unmarried. (23)

Although a study in Japan found unmarried people with HD had a much greater mortality rate than those who were married, it is not known if a similar phenomenon exists in this country. (36) It is also not known whether the care setting influences survival, mortality or cause of death in HD. Improving end-of-life care in HD would be facilitated by a better understanding of how the setting of death influences mortality and cause of death.

Do People with HD Have Advance Directives? What Features of the Illness Predict the use of Advance Directives by Stage?

No literature is available on the prevalence of Advance Directives in HD. The study of nursing home residents suggested that only 66 percent of those dying of HD had a Do Not Resuscitate (DNR) order and only 43 percent used a proxy for medical, legal or financial matters. (23) There are a number of pertinent studies in people with dementia of other causes. Systematic studies of elderly demented residents in long-term care facilities suggest that many demented residents have the capacity to execute a health care proxy. (37)

Optimal care would dictate that each person with HD have Advance Directives in place long before the terminal stages of the illness. This clearly represents an unmet need, the reasons for which remain unclear. However, it has been suggested that neurologists have a poor record of promoting the use of Advance Directives of all kinds. A recent review found few articles on Advance Directives and neurological diseases; the topic was not well covered in the primary neurologic literature. (38) Improving this deficiency will require educational efforts about Advance Directives directed at neurologists and other

health care providers as well as a greater presence in the primary neurologic literature of research about the end of life in neurologic illness.

Policy and Financial Aspects of Care for People with HD

If one were to design a condition with features that will lead it to fall between the cracks of the American health care systems and social safety net, one could hardly do better than Huntington's Disease. HD is a chronic degenerative disease that spans roughly the years of middle age, 30-50 years, and combines a wide variety of medical and social service needs during its course. Acute care medical services tend to be less important than access to prescription drugs, mental health services, rehabilitative services for mobility and gait, assistance with activities of daily living and custodial care and supervision regarding psychosocial symptoms of impaired judgment, inappropriate behavior, and dementia. Some periods of in-home assistance and eventually institutionalization in a long-term care facility are typical.

This combination of symptoms and needs presents a major challenge for the American health care system, which is currently designed as a hospital-based, high-tech-oriented, curative and acute care medical system. Its great weakness is in coordinating a range of social and medical services, continuity of care, financing expensive prescription drugs, mental health services and long-term care. The needs of people with HD clearly fall within the zone of the system's weakness rather than its strength.

Acute Care

The age of onset and progression of HD and its distribution in the population ensure that most people affected by HD fall outside the two main areas of public health insurance — Medicare for the elderly (65+) and Medicaid for children and for the poor with dependents. Eligibility standards differ by state but most are set below the federal poverty level, which is currently defined as an annual income of approximately \$18,400 for a family of four. Moreover, the U.S. public health insurance system does not provide well for those with long-term chronic illness and disability whether that disability be physical, cognitive or psychosocial. The worker's compensation and social security systems address income maintenance for those with chronic illness. Someone younger than 65 can qualify for supplemental income benefits under social security if they are totally and permanently disabled and unable to work. Therefore, disabling features of HD may ensure provision of health insurance through Medicaid. In some states, those benefits can be better than those provided to the elderly under Medicare and at times even better than some private insurance. But this support is based upon impoverishing the person with HD, if not the entire family. Even so, it is the rare family that will not be severely financially affected by the course of HD due to significant gaps in Medicaid coverage.

Private health insurance coverage is largely provided through group plans organized in the workplace, so this coverage is a primary source for people with HD until their condition progresses to the point where they cannot maintain employment. There is then a period when they can maintain their former insurance plan at group premium rates through an out-of-pocket payment, but eventually they must purchase an individual

private policy at much higher premium rates. Further complicating private insurance coverage is the genetic basis of HD. When the individual applies for private insurance, the required medical history calls upon them to disclose either their own at-risk status or their family history. Under some circumstances, HD can then become the prototypical pre-existing condition, to which insurance underwriting and premium rating will most likely be applied. This could make coverage unaffordable. Unusual among genetic disorders, HD can be predicted with virtual certainty long before the onset of symptoms through genetic testing. Therefore, depending upon state law and regulation, and depending upon how federal law concerning genetic privacy evolves in the near future, some people with HD in some states may find themselves unable to obtain affordable health insurance for the duration of their illness. Treatment for co-morbid conditions may continue to be covered by existing insurance, however. For the rare juvenile onset form of HD, they may still be covered under their parents' private insurance policy until they reach age 18. (Beyond that age, policies differ concerning how long and under what circumstances the child can be kept on the parents' policy.)

In recent years, the trend in the workplace and in the private group insurance market has moved away from offering family coverage without a substantial employee co-payment. As a result, increasing numbers of workers are not carrying family coverage.

Generalizations here are difficult because regulations differ by state, and the entire area of health insurance design and financing are in flux. But one thing is likely: The person with HD and their family will find themselves in a continuing struggle to obtain insurance reimbursement and financing for needed medical services, prescription drugs and access to specialists throughout the latter stages of the disease.

Long-Term Care

On the long-term care side, the picture looks even bleaker. Private insurance policies may only pay for very limited and short duration home care services. Often to qualify for home care coverage, the person must be completely housebound. Private insurance policies do not pay for custodial care in long-term care facilities or for skilled nursing care for prolonged periods; neither does Medicare. On the other hand, Medicaid will pay for care in a long-term care facility if the individual is medically indigent. Complex issues arise concerning the extent of the family's financial obligations during the individual's care or after their death (e.g., federal regulations stipulate that after a patient's death, attempts should be made to recoup Medicaid funds from the estate. Surviving family members may find themselves faced with liens being placed on inherited property and the like).

Due to the complexity of care involved and low reimbursement rates for people with mainly custodial needs, long-term care facilities may be reluctant to admit people with HD, since many are on Medicaid coverage from the time of admission rather than following a period of private out-of-pocket pay (at higher rates). Even with insurance coverage, placement in a long-term care facility may be difficult, if not impossible, to arrange for HD families. In summary, careful financial and estate planning are essential in the early stages of HD before the person loses decision-making capacity and legal competence, so that assets can be divided among the person with HD and spouse in order to limit the spouse's financial liability for long-term care services.

Medication

The behavioral and psychological symptoms of HD can be treated with expensive medications but coverage of prescription drugs is an ongoing problem in the American health care system. Some private insurance policies cover prescription drugs and some do not, depending upon the employer and the policy. For indemnity type insurance coverage, a substantial co-payment is required out of pocket for the insured. For managed care, the co-payment may also be quite large for some types of new or very expensive drugs. Some brand name drugs may not be covered at all. Some managed care plans make it difficult to obtain the psychiatric care that some people with HD need or that HD families might prefer. Standard Medicare does not yet cover prescription drugs although Congress is currently considering adding such coverage. Medicare Managed Care plans (Medicare Plus Choice) do offer drug benefits but many providers have been pulling out of this system precisely because of the drug costs and other expenses associated with the care of the elderly and the chronically ill. Medicaid does cover drugs, but subject to variation among states and among the managed care plans that contract with state Medicaid offices to actually provide the coverage. At later stages, the prescription drug needs of the person with HD are covered by the long-term care facility. Although here again controversies can arise in individual care plans over whether a particular drug is available and whether it should be provided in generic or branded form.

Rehabilitation

Rehabilitation services may prolong the mobility and independence of a person living with HD and enhance their quality of life. Coverage for rehabilitative services has long

been handled, as a policy area, under the rubric of the worker's compensation system. The goal of that system is essentially restoration of the ability to return to work. Total and permanent disability status, which triggers other Social Security and Medicaid benefits, tends to undermine the rationale for extensive rehabilitative services. The system is fraught with problems and restrictions. Only under very unusual circumstances would public coverage for inpatient rehabilitation be available for people with HD. Out-patient rehabilitative services may be covered, though not for a long duration, and subject to periodic assessment and certification of "progress," which people with HD may not be able to demonstrate.

Some payment for rehabilitation services is available under Medicare, Medicaid (as a mandated benefit for children and an optional benefit for adults) and some private insurers. Even when these third-party payers provide benefits, actual reimbursement amounts depend on both policies and interpretations by different fiscal intermediaries that can vary widely even within the same program.

As the result of the Balanced Budget Act of 1997, Medicare Part B (outpatient) benefits for occupational therapy are limited to \$1,500. Effective July 1, 2003, physical therapy and speech-language pathology services combined are limited to \$1,500. In the case of Medicaid, limited reimbursement for provider services can result in difficulty in finding a provider who accepts the Medicaid rate.

For people with HD, reimbursement for a swallowing evaluation and treatment is common, since this ability can be improved. (39) The length of treatment is relatively short and restoring the ability to take nutrition orally can eliminate the need for more

costly medical procedures with higher, more expensive levels of care. Reimbursement for other rehabilitation services is less routine since medical necessity and expectation of improved function must be demonstrated. Even then, only a limited number of sessions are typically authorized. Treatment prior to demonstrating impaired function is seldom, if ever, allowed. Neither are clinical models that emphasize family intervention without significant direct patient contact.

These treatment policies do not contribute to the quality of life of people with HD. The number of sessions allowed arbitrarily limits progress. Earlier intervention, when the person with HD is not yet impaired and can benefit from treatment, is not reimbursable, although it may enable the patient to more easily learn skills that are needed as the disease progresses. While teaching the family more effective communication is more beneficial than direct patient intervention in the later stages of the disease, private insurers will typically not pay for it.

Medicare will pay for a limited number of sessions to develop a program for maintaining the current functional level for people with progressive diseases, including family training (*Medicare Program Integrity Manual* [2002], Chapter 6, Section 6.5.2B, Centers for Medicare and Medicaid Services). Development of a new maintenance program may be authorized if functioning declines with progression of the disease. Additional services are not considered reasonable and necessary after instructions to the patient and supportive personnel have been completed and they can safely and effectively carry them out.

End-of-Life Decision Making

The main public policy issue to be addressed has to do with medical decision making. Statutes exist in every state that provide a mechanism for an individual to designate another person to make health care decisions when and if the person loses their decision-making capacity. Such a loss of capacity is a predictable occurrence at some point during the course of HD. Advance decision-making planning is best done in the early stages involving the person with HD, the agent to be designated, other family and friends and the person's physician(s). A durable power of attorney for health care must be executed in accordance with state law. An additional treatment directive (living will) may be included to further state the person's wishes and treatment preferences, especially regarding life-sustaining treatment modalities. Special mention should be made of their preferences regarding the use of artificial nutrition and hydration because in some states, the health care agent must be explicitly authorized to make those decisions (see Advance Directives in Addendum A of Appendix A, Care Subcommittee report).

Under the heading of health care decision making, two additional areas of public policy have potentially significant implications for the care of people with Huntington's Disease and their families. The first has to do with the use of medical records as regulated by the Health Insurance Portability and Accountability Act of 2000 (HIPAA), and the second area involves regulations for surrogate consent for participation in medical research.

HIPAA

Enacted by Congress in 1996 with implementing regulations issued by the Department of Health and Human Services in August 2002, this complex law contains significant new rules designed to provide continuation of health insurance under certain circumstances; to protect the privacy of individual medical records through appropriate administrative, technical and physical safeguards; and to control the electronic storage and transmission of those records. The basic point of the law is to place the individual in a position to control what is done with medical records that identify that individual, and to be aware of who has access to those records and how they are being used. Prior to HIPAA, the issue of who owned medical information and how it could be shared, disseminated, transmitted and used was less clear. Individuals could be subject to discrimination or embarrassment if their medical condition was disclosed to third parties such as employers, landlords, neighbors, friends or professional marketers. Under HIPAA, identifiable medical information can be used for treatment, payment and health care operations, but other uses, e.g., medical research, require written authorization of the individual. The definition of medical information within HIPAA includes genetic information. Also, as with any new law as complex as HIPAA, it may take several years and much litigation before regulations that are applied to specific and particular cases become entirely clear.

At this time, one concern regarding the possible implications of HIPAA for people with HD and families is worth noting. In general, HIPAA may be seen as beneficial to the HD community because it strengthens privacy protections concerning information that can be socially stigmatizing or could subject the individual to discrimination. On the other hand,

improving the continuity and coordination of services for people affected by HD would be one of the most important health care reforms that could improve the quality of care throughout the course of the disease (and also at the end of life). As previously mentioned, HD is a complex condition that requires multiple forms of care and assistance, some medical, some psychological, rehabilitative, social, residential and occupational. Increased concern for the protection of medical confidentiality and privacy may make developing well-integrated networks of various kinds and levels of assistance more difficult and more expensive. At the very least, it may require the person with HD to be called upon numerous times to provide (usually written) consent for information transfer if the information is not clearly and directly related to treatment. At times during the course of the disease, when the person's executive functions are compromised or when the person may be struggling with changes of mood and affect, being called upon for routine consent may prove complicated and burdensome.

Moreover, the requirement for consent to release and share medical records underscores the importance of an Advance Directive in the form of a durable power of attorney for health care. With such legal authorization, a family member or friend would be able to fulfill HIPAA requirements if the patient lacks decision-making capacity. Without such authorization, it is not clear how consent under HIPAA will be given. Until this is clarified, families may encounter more red tape in the course of managing care for their loved ones.

Medical Research

The second area of policy addresses participation in clinical trials on new drugs and diagnostic tests that may be developed in the future. Many individuals and families may wish to enroll in these trials for a variety of reasons. In most cases, the research cannot promise direct therapeutic benefit to subjects enrolled in the study but they can feel nonetheless that they are doing something to help people with HD and their families in the future. This altruism may extend to unknown strangers but in the case of a familial disease such as HD, the concern for a younger generation in one's own family is obviously a factor.

All medical research involving human participants is subject to review by Institutional Review Boards (IRBs), which are committees based in hospitals, universities or research centers. These committees look at experimental designs carefully to ensure that the participants have given truly informed consent and that the risk-benefit ratio in the experiment is justified. For the most part, experiments that raise the most serious ethical questions are those in which someone else must provide consent because the individual cannot, where there is no direct medical benefit to the individual and there is more than minimal risk for the individual. This is precisely the set of circumstances that confront people with HD in later stages of disease. Do Advance Directives for health care apply to medical research as well? In a study now underway, interviews with the Attorneys General in all fifty states indicate that the answer would be "no" in most states, or at least it would be legally unclear. Until state legislatures amend and clarify the existing Advance Directive laws, it may not be possible for people with HD without capacity to

participate in clinical trials. This may not have a large impact on the HD community at present since so few promising drugs are now under investigation. As that situation changes, however, the lack of a clear public policy regarding participation of vulnerable individuals with neurological or psychiatric disorders in clinical trials may become a serious problem.

The following chart summarizes the main point of this review, using the five-stage Total Functional Capacity (TFC) Scale.

	Acute Coverage	Long-Term Care Coverage	Medication	Rehabilitation	Decision Making
Stage I	Private Group Private individual Medicaid	None	Variable by policy	None	Execute Advance Directives before loss of capacity Estate planning
Stage II	Coverage will continue as long as employment	None	Variable by policy	None	
Stage III	Conversion to Supplemental Security Income (SSI) and Medicaid Private insurance unlikely to be affordable/ obtainable	Included with Medicaid if institutional; Home care will mainly be volunteer or private pay	Coverage by Medicaid	Limited	Need surrogate or agent to provide consent for sharing medical information; Possible opportunities to take part in medical experimentation
Stage IV	Medicaid	Private pay or spend down to Medicaid nursing home coverage	Coverage by Medicaid	Limited	Need surrogate for consent to share medical information; Clinical research may be an issue
Stage V	Medicaid Medicare depending on age	Medicaid	Coverage by Medicaid Hospice Medicare Benefit	None	Health care agent will probably have to make some difficult decisions, including eventually a DNR decision in this stage

Legislative Influence on Care for People with HD

Disability

Congress has established major programs for people with disabilities. The Americans with Disabilities Act (ADA) prohibits discrimination in employment, state and local government programs, services, and activities; public accommodations; and telecommunications. Section 504 of the Rehabilitation Act prohibits discrimination in federally-assisted programs and activities. The Individuals with Disabilities Education Act (IDEA) provides a free and appropriate public education. Title II of the Social Security Act (Federal Old Age, Survivors and Disability Insurance) authorizes federal monthly cash benefits to workers with disability who have paid Social Security taxes for roughly half the years since they were 21 and to certain of their family members. Individuals entitled to disability benefits for 24 consecutive months become eligible for Medicare as well. Title XVI of the Social Security Act provides Supplemental Security Income to establish a minimum income level for people with disabilities who meet a test of financial need. Unlike the Title II program, family members must qualify on their own behalf. For all of these programs, states may pass their own laws but cannot limit the rights or benefits provided by federal statute.

To qualify for any of these programs, a person must have a disability as defined by the law that created the program or the regulations that implement that law. Disability for each of these programs is defined as follows:

- Americans with Disabilities Act (ADA)
Disability means, with respect to an individual –

- a physical or mental impairment that *substantially limits* one or more of the *major life activities* of such individual;
- a record of such an impairment; or
- being regarded as having such an impairment [29 CFR 1630.2(g)].

Substantially limits means unable to perform a major life activity that the average person in the general population can perform, or significantly restricted as to the condition, manner or duration under which an individual can perform a particular major life activity [29 CFR 1630.2(j)].

Major life activities means ability to care for oneself, perform manual tasks, walk, see, hear, speak, breathe, learn and work [29 CFR 1630.2(i)].

People currently engaging in the illegal use of drugs are specifically excluded.

- Section 504 of the Rehabilitation Act
Handicapped person means any person who has a physical or mental impairment which substantially limits one or more major life activities, has a record of such an impairment, or is regarded as having such an impairment [7 CFR 15b.3(i)] (Note: ADA definition modeled after this definition).
- Individuals with Disabilities Education Act (IDEA)
Child with a disability means a child evaluated as having mental retardation, a hearing impairment including deafness, a speech or language impairment, a visual impairment including blindness, serious emotional disturbance (hereafter referred to as emotional disturbance), an orthopedic impairment, autism, traumatic brain injury, and other health impairments, a specific learning disability, deaf-blindness, or multiple disabilities, and who, by reason thereof, needs special education and related services [34 CFR 300.7 (a)(1)].
- Federal Old Age, Survivors and Disability Insurance (Social Security Disability Income, or SSDI)
Disability is the inability to do any substantial gainful activity by reason of any medically determinable physical or mental impairment that can be expected to result in death or that has lasted or can be expected to last for a continuous period of not less than 12 months. To meet this definition, one must have a severe impairment, which makes it impossible to do previous work or any other substantial gainful activity that exists in the national economy [20 CFR 404.1505(a)].

An appendix of more than 73,000 words details requirements for medical and other evidence of the existence of disability for 15 body systems.

- Supplemental Security Income (SSI)
The definition of disability for *adults* parallels that for SSDI [20 CFR 416.905 (a)]. The definition for *children* (under 18 years of age and who are entitled to benefits on their own behalf) is a medically determinable physical or mental impairment or

combination of impairments that causes marked and severe functional limitations, and that can be expected to cause death or that has lasted or can be expected to last for a continuous period of not less than 12 months [20 CFR 416.906].

The definition of disability is program-specific, with the SSDI and SSI definitions being the most restrictive. The ADA and Section 504 definitions seem to apply, in general, to people with HD at Stage III or later. The IDEA definition may apply as early as Stage II if educational performance is affected. The SSDI and SSI definitions apply to a person with Stage IV to V HD.

The definitions of disability under the ADA, Section 504, and IDEA do not seem to particularly disadvantage people with HD. However, since they leave the workforce early, it is unfortunate that the SSDI and SSI definitions are so restrictive, particularly with respect to inability to perform any gainful activity. At least when a patient does qualify for SSDI, benefits extend to his or her children. Under both SSDI and SSI, documentation requirements are extensive and benefits are subject to continuing disability review, potential trial work periods, other administrative requirements and extensive paper work, which families with HD may not have the physical or emotional energy to complete.

Overview of State Laws Specific to HD

As of the end of 2002, a few states have passed legislation that specifically cites Huntington's Disease. At least three states have laws establishing medical care and social support programs for people with hereditary disorders, including HD (note that citations

refer to the annotated laws of individual states; these laws are often available on individual state Web sites):

- California (Cal Health & Saf Code § 125130),
- Massachusetts (Mass. Ann. Laws ch. 111, § 60), and
- New Jersey (N.J. Stat § 26:5b-2).

Connecticut's law (Conn. Gen. Stat. § 17b-25b) creates a program specific to HD.

Kentucky (KRS § 304.17B) classifies HD as a high cost condition and allows people with HD to participate in Kentucky Access, a health insurance program for the uninsured.

Maryland (Md. Insurance Code Ann. §14-501ff.) created a state-run comprehensive health benefits plan for certain individuals with pre-existing conditions for which people with HD can qualify. Illinois (20 ILCS 2305/5.5) charged its Department of Public Health to establish specialized training and experience criteria for people who provide health or home care to people with Alzheimer's disease or other dementia-related disorders, including HD. California passed laws for HD research grants and a statewide consultant to enhance quality of care and treatment of "brain-injured" adults (Cal Wel and Inst Code § 4362).

These laws were identified from a key word search of a proprietary database of state laws using "Huntington's Disease" or "Huntington's chorea." A total of 23 citations were generated and read to identify substantive actions and to eliminate multiple references to HD in different parts of the same law.

For comparison, a similar search was done for "Parkinson*" (where * provides a wild card search so that references to "Parkinson's," "parkinsonism," and similar variants would be captured) and "Alzheimer's Parkinson*" generated 77 citations and

“Alzheimer’s” 448 citations, both more than the 23 citations for HD. Although the last two searches were not analyzed in-depth as was done for HD, the greater number of citations for “Parkinson*” and “Alzheimer’s” seems to indicate a greater legislative interest in these diseases than in HD. One may speculate that greater legislative interest in a disease improves quality of care for that disease, but no empirical data were found to support or refute that hypothesis.

Specific Laws Related to Certain Clinical Decisions

A similar key word search was done to identify state laws with respect to certain clinical decisions: use of restraints (physical or chemical), provision of artificial nutrition, use of psychopharmacological drugs and cessation of active treatment. Federal laws referenced in citations from the state search were also analyzed.

Use of Restraints

Many states have statutes that protect a patient’s right to be free from physical or chemical restraints when used for purposes of discipline or staff convenience. Restraints can only be imposed as part of medical treatment or to ensure the physical safety of the patient or others. Even then, such use requires the written order of a physician that specifies the duration and circumstances under which the restraints are to be used.

Typically, restraints without order may be used under emergency circumstances, but written orders must follow promptly. Skilled nursing facilities in all states, as part of conditions for participation in Medicare and Medicaid programs, must assure the patient’s right to be free from restraints as stipulated in 42 U.S.C.S. § 1395i-3(c)(1)(A)(ii) and 42 U.S.C.S. § 1396r(c)(1)(A)(ii), respectively.

Specific states may impose additional conditions on the use of restraints such as:

- informed consent;
- notification of a person or organization of the patient's choosing when restraints are used;
- patient monitoring at specified intervals (e.g., 10-15 minutes) while under restraint;
- periodic release from hand restraints for people whose primary mode of communication is sign language;
- detailed requirements for documentation of need;
- required trial use of less restrictive measures;
- authorization of only specific personnel who can impose the restraints; and
- written facility policies and procedures on the use of restraints.

These conditions are not meant to be restrictive but rather to preserve patients' rights.

Public policy per se does not seem to prohibit the use of restraints for people with HD more than it would for other patient populations. Professionals in states that have a long list of conditions for use of restraints may consciously or subconsciously refrain from authorizing their use due to facility resistance to compliance with all of the conditions.

People with HD who use alternative and augmentative forms of communication, e.g., word/picture books or electronic devices, may need use of their hands while under physical restraint. Orders for use and circumstances of physical restraint should consider this need.

Artificial Nutrition

State laws are consistent in viewing the provision of artificial nutrition and hydration as life-sustaining. They also give patients the right to make decisions regarding the provision, withholding or withdrawal of such treatment as long as they have the decision-making capacity to do so. States typically require that loss of capacity to make health care decisions be determined by the attending physician or by the attending physician with corroboration of another physician or psychologist. Traditionally, general mental status

instruments are used to make this determination although specific instruments to evaluate skills necessary to provide informed consent for treatment or to execute a health care proxy are currently under development and testing. (37, 40)

What happens when a patient loses capacity varies considerably from state to state. States typically require a properly executed health care Advance Directive that designates an authorized representative to make decisions about life-sustaining treatment. Some of these states do not permit withholding or withdrawal of artificial nutrition or hydration unless the patient has specifically given the authorized representative the power to make such decisions (e.g., New Hampshire in RSA § 137-H:6). A state (e.g., South Carolina in S.C. Code Ann. § 44-77-65) may even require that such a declaration made in another state be supplemented with a document specific to the current state of residence. A state (e.g., Kentucky in KRS § 311.629(3)(d)) may also restrict a representative's ability to withhold or withdraw artificial nutrition or hydration if needed for comfort or the relief of pain.

When an Advance Directive is absent, some states do not permit the withholding or withdrawal of artificial nutrition or hydration. Other states (e.g., California in Cal Prob Code § 3208) allow a court, under certain circumstances, to appoint a representative to make such a decision. Still other states have developed mechanisms for appointing surrogates for this decision making. Hawaii (HRS § 327E-5) allows the primary physician, or the physician's designee, to locate people interested in the patient and to have them reach a consensus as to who among them should make health care decisions

for the patient. The surrogate can then make decisions on artificial nutrition and hydration when the primary physician and a second independent physician certify that provision or continuation of artificial nutrition or hydration is merely prolonging the act of dying. Nevada (Nev. Rev. Stat. Ann § 449.624) allows the authorized representative or a family member to make the decision. North Carolina (N.C. Gen. Stat. § 90-322 (b)) has established a hierarchy for this decision making: (from first to last) a health care agent, a guardian, the person's spouse, a majority of first degree relatives and finally the attending physician. Other states have a different hierarchy.

Advance decision-making planning is vital for all people with HD and must adhere to the requirements of a specific state. Without specific Advance Directives, a patient may be over-treated by providers concerned about liability or under-treated because of a health agent's lack of knowledge of patient preferences.

Requirements for Advance Directives are not barriers to quality of care as long as they are included as part of the early stages of case management for HD. Such planning is especially critical in states that do not provide the option of surrogate decision-makers and for patients for whom a health care agent may be difficult to identify. Some 40 states offer public guardianship programs, but they are usually unfunded or underfunded.

Moreover, only a few states have established certification or training for such guardians.

Psychopharmacological Drugs

As with artificial nutrition, patients retain the right to make decisions regarding the provision, withholding or withdrawal of psychopharmacological drug treatment as long

as they have the decision-making capacity to do so. Following an order by a physician, patients may be required to provide written consent for administration of these drugs after first receiving information on the consequences of the medication and alternatives. These rights are often asserted in the *Patients' Bill of Rights*, especially for residents of skilled nursing facilities.

When the patient loses capacity, states then require a health care Advance Directive and, again, as for artificial nutrition, may require specific authority or a court order to make decisions about psychotropic medication (e.g., Alaska in AS 47.30.836). States that allow surrogates to make health care decisions typically allow them to make decisions about the administration of these drugs.

A state may have additional requirements for the use of psychopharmacological drugs. Connecticut (Conn. Gen. Stat. § 19a-550[b][21]) requires a written plan of care that is reviewed at least annually by an external consultant for appropriateness. When antipsychotic medication is used in long-term health facilities, California (Cal Health & Saf Code § 1418.9) requires notification of an interested family member. A comparison of the utilization rate of these drugs in states with and without additional requirements would provide evidence to influence public policy.

Without Advance Directives, the same risks of under-treatment and over-treatment occur as with artificial nutrition. The authority for the health care agent to make decisions about

the use of psychopharmacological drugs must be stated specifically when Advance Directives are developed.

Cessation of Active Treatment

Besides the situations noted above, it is not readily apparent that public policy restrictions exist that are specific to the decision to move a patient from active treatment toward a palliative care model. When patient rights are enumerated in statute, they discuss broad principles such as:

- The right to receive adequate and appropriate medical care;
- The right to be fully informed of condition, treatment options and expectation for improvement;
- The opportunity to participate in the planning of medical treatment;
- The right to receive services with reasonable accommodation of individual needs and preferences; and
- The right to reasonable continuity of care.

If a patient does not have the capacity to participate in decision making, these rights typically devolve to a guardian or other representative without need for specific authority to make this decision.

Codes of ethics and state licensing acts for individual professions provide further guidance to providers. However, such a search was beyond the scope of the present investigation.

Juveniles with HD

Special considerations for juveniles with HD have been discussed above. The problem of private health insurance coverage of children with pre-existing conditions or children beyond age 18 (somewhat longer if the dependent is a student) when coverage under a parent's policy lapses, was discussed under the previous section, "Policy and Financial

Aspects of Care for People with HD.” An HD child’s entitlement to educational services was discussed under “Disability,” a subsection of “Legislative Influence on Care for People with HD.” Until educational performance becomes affected, children with HD still have access to certain accommodations under Section 504, since almost all schools are recipients of some form of federal funding.

Conclusions

The following represent gaps in our knowledge base or gaps in epidemiologic research, financing and policy pertinent to improving end-of-life care in HD:

- The exact epidemiology of Huntington’s Disease (HD) remains unknown as no formal epidemiological studies have ever been conducted. The available large databases of HD, located at Indiana University and Johns Hopkins University have a primary focus on genetic testing and presymptomatic HD individuals. The largest database of individuals with manifest HD is maintained by the Huntington Study Group, which has been systematically collecting demographic and clinical information on HD patients since 1993.
- The lack of formal epidemiological studies of HD make it difficult to identify and characterize specific demographic features of HD patients by stage of illness. Despite the availability of robust and reliable clinical markers of stage of illness, little information is available regarding demographic and occupational features of HD patients by stage of illness, most notably early to mid-stage, when patients are more likely to be functional and employed.
- Accurate estimates of HD prevalence by stage of illness have not been studied. The existing published literature on HD prevalence provides group data without breakdown by stage of illness.
- Little information is known regarding caregiver burden in HD. There have been no published studies of caregiver burden in HD. Demographic characteristics of HD caregivers are largely unknown. Moreover, the relationship between caregiver demographic features as well as caregiver burden and the clinical features of HD has not been systematically investigated.
- While it is known that HD clinical care requires a multidisciplinary effort involving individuals with backgrounds in neurology, neuropsychology, psychiatry, social work

and ancillary therapies (speech, occupational and physical therapy), little information is available regarding types of professionals caring for people with HD by stage of illness.

- What seems clear from the literature is that many, if not most, people with HD spend a significant part of their disabled lives in institutional settings, particularly in long-term care facilities. Once admitted, discharge does not occur. A better understanding of the risk factors for placement in a long-term care facility and for subsequent death would likely lead to strategies that keep people with HD at home longer and give more patients access to hospice care or other home care services.
- Limited study suggests that many people with HD, even those with advanced disease, have not executed Advance Directives. The reasons for this are unclear since HD is a progressive illness with a uniformly fatal outcome. Reluctance on the part of neurologists may play a role in this shortfall. Because of progressive cognitive changes, Advance Directives are best executed early in the course of the illness. Strategies to increase the use of Advance Directives will allow patients more control in the terminal stages of the illness.
- The definition of disability varies by federal statute or program. Definitions do not seem to negatively impact people with HD, except for Social Security Disability Income (SSDI) and Supplemental Security Income (SSI). Because of stringent definitions under these programs, eligibility for these cash benefits does not occur until the later stages of HD.
- Unlike other neurological diseases, such as Parkinson's or Alzheimer's, few states have passed laws that establish special programs for care of people with HD. Clinical decisions like the use of restraints, provision of artificial nutrition, use of psychopharmacological drugs and cessation of active treatment are governed by the state.
- Laws concerning health care decision making vary by state. States may require that any Advance Directive include specific authority to make certain decisions. If a patient does not have decision-making capacity and a specific authority has not been granted in an Advance Directive, a patient may be over-treated by providers concerned about liability or under-treated because of a health agent's lack of knowledge of patient preferences.
- Health insurance benefits for people with juvenile onset HD is problematic after the age of 18.

Research Resources

National Huntington Disease Research Roster Indiana University Medical Center

975 West Walnut Street
Indianapolis, IN 46202

The National Huntington Disease Research Roster was founded in 1979 at Indiana University in response to a recommendation from the Commission for the Control of Huntington's Disease and its Consequences. The Roster is funded by the National Institute of Neurological Diseases and Stroke. The Roster, which contains family trees and demographic data, provides a link between researchers and patients and family members.

Huntington Study Group

<http://www.Huntington-Study-Group.org>

The Huntington Study Group (HSG) maintains a database of clinical information on patients followed by HSG clinicians. Demographic data and serial ratings with the Unified Huntington Disease Rating Scale are available in this database. Within the HSG, the Clinical Correlates Working Group analyzes the UHDRS database, including factors associated with disease onset and progression as well as relationships between various components of the UHDRS.

The National Nursing Home Survey: 1999 Summary

The National Nursing Home Survey is a series of national surveys of nursing home residents and staff. Surveys have been conducted in 1973-1974, 1977, 1985, 1995, 1997, and most recently in 1999. Nursing homes surveyed had at least three beds and were certified by Medicare, Medicaid or a state licensing board. Data about the facilities include characteristics such as size, ownership, Medicare/Medicaid certification, occupancy rate, number of days of care provided and expenses. For recipients, data are obtained on demographic characteristics, health status and services received. Although demographic data on diagnosis does not specifically address HD, these patients are included under "other diseases of the nervous system and sense organs" (includes ICD-9-CM 320-330, 331.3-331.9, 333-337, 341, 345-389). The 1999 National Nursing Home Survey resident questionnaire may be downloaded from the CDC web site at www.cdc.gov/nchs/data/series/sr_13/sr13_152.pdf. The survey begins on page 91.

Minimum Data Set

Regulatory guidelines demand the collection of the Minimum Data Set (MDS) in every nursing home participating in Medicare or Medicaid programs (Omnibus Budget Reconciliation Act of 1987). The revised version (MDS) was implemented in 1996 and contains more than 400 items addressing common problems such as delirium, cognitive impairment, falls, mood, etc. Full assessments are required at least annually or when there is a significant change in patient population. Information collected that is directly relevant to HD research includes: prior residential history, marital status, Advanced Directives (do not resuscitate, do not hospitalize), memory and other cognitive skills,

ability to communicate, indicators of depression and anxiety, behavioral symptoms (wandering, verbally abusive, socially inappropriate behaviors), activities of daily living, mobility, nutrition and weight and continence. Information on co-morbid diseases such as diabetes and heart disease is collected as is information on pneumonia. There are data on the number of medications administered and data on antipsychotic, anxiolytic, antidepressant and hypnotic medications is collected. Utilization of rehabilitative services is recorded, as is use of physical restraints.