Specialty Clinic Perspectives

In response to a request by the local parent group, a clinic was developed to provide for the medical and psychosocial needs of adults with Down syndrome. One of the concerns of the parents was that their sons and daughters were not receiving an adequate evaluation when they had a decline in function and were being given a diagnosis of Alzheimer's disease. The thorough evaluation that each patient receives at the clinic is described. The majority of the adults seen showed no decline in function. Of those that showed a decline in function, a small percentage were diagnosed with Alzheimer's disease, but most of those with a decline had a potentially reversible problem and, with treatment, most returned to their premorbid level of function.

The occurrence of Alzheimer's disease among persons with Down syndrome has caused a great deal of concern for families and care providers of adults with Down syndrome. As discussed by Holland (this volume), Dalton and Janicki (this volume), and others, research had found that all adults with Down syndrome over the age of 40 develop plaques and tangles in their brains similar to those seen in persons and professionals often conclude that all persons with Down Syndrome will eventually develop Alzheimer's disease. However, this conclusion is primarily based on autopsy data without a similar evaluation of premorbid clinical information. Many parent organizations are concerned that such conclusions may have deleterious effects if it is believed that all adults with Down syndrome eventually will be effected by Alzheimer's disease. This was the case in the metropolitan Chicago, Illinois, area. Parents were concerned that, based on such conclusions, adults with Down syndrome were being given the clinical diagnosis of Alzheimer's disease whenever they had a decline in function and that these diagnosis
were being made often without any substantive clinical evaluation. Their concern was strong enough to seek assistance in developing a center with sufficient expertise to offer periodic health and neurological reviews and to the able to conduct differential diagnoses when presented with the symptoms of dementia. Thus, at the request of the National Association for Down Syndrome (NADS, a parent group serving the Chicago Metropolitan area), the Adult Down Syndrome Center of Lutheran General Hospital was developed. This chapter discusses the concerns of the parents, describes the development of a multidisciplinary clinic to address their concerns, and offers information on how clinic staff evaluates persons with Down syndrome who are discussing in function and potentially developing Alzheimer's disease. Recommendations for the prescriptive care of persons who have been diagnosed with Alzheimer's disease are also discussed.

The Down Syndrome Clinic Model

In the metropolitan Chicago area, the NADS has been providing advocacy services for persons with Down syndrome since 1961. This voluntary association was started by parents of young children and, therefore, for a number of years their focus was on children and adolescents with Down syndrome. However, as their sons and daughters grew beyond childhood, new concerns became evident. Parents felt that their children had received good care from their pediatricians and family physicians, but that as their children reached adulthood, they recognized that the dearth of practitioners experienced with adult development and intellectual disabilities could impede good health care. To address these concerns, NADS initially provided funds to help pay a social worker to meet with persons with Down syndrome, their families, and careers and provide counseling services, advocate for services, and support the parents when their sons and daughters were having problems.

During the early 1990s, NADS also surveyed its members regarding the psychosocial and medical concerns they had and confirmed the need to address the medical and psychosocial issues associated with aging of their sons and daughters with Down syndrome (Chicoine, McGuire, Hebein & Gilly, 1994). A major concern expressed was a decline in function experienced by some persons with Down syndrome and whether the decline could be secondary to Alzheimer's disease. Parents expressed particular concern for how their sons and daughters were being evaluated when they presented with a decline in function. Numerous stories were shared of persons with Down syndrome presenting with a decline in function and being given a diagnosis of Alzheimer's disease and the need to exclude other diseases that present similarly but are reversible. The parents sensed that many physicians had little training with regard to adults with an intellectual disability and were making a diagnosis without an adequate evaluation. They expressed interest in the development of a clinic that would specifically address the needs of adults with Down syndrome and their families and care providers.

These concerns led to a collaborative effort being undertaken by the NADS, the Lutheran General Hospital, and the University of Illinois at Chicago to develop a
multidisciplinary clinic to serve the needs and concerns of persons with Down syndrome and their families and care providers. Using a community-oriented primary care model as a foundation, a new clinic was designed based on discussions with the NADS staff and other parents (Chicoine, McGuire, Hebein, & Gilly, 1995). Since so many of the concerns involved both medical and psychosocial issues, it was felt that a team approach would be the best strategy. Thus, the initial staff included a physician and a social worker in a tandem. As the clinic began seeing adults, several other team members were added. One, a specialist on hearing assessment, was added because it was felt that hearing impairment was a common problem in persons with Down syndrome (Evenhuis, van Zanten, Brocaar, & Roerdinkholder, 1992). Audiology screening services were now included to help determine whether hearing was a cause in notable behavioral decline. In addition, a nutritionist was added because obesity was also common in persons with Down syndrome (Cronk, Chumlea, & Roche, 1985). Parents expressed significant concern about obesity and a need for sound nutrition.

As part of the development process, core staff also visited other local clinics and reviewed what services were being offered. Soon additional disciplines and services, such as medical consultants, physical therapists, and occupational therapists, as well as vision screening were included as part of the clinic's offerings. However, due to cost and inconsistent need, these were provided as referral services and not provided on site. For example, it was observed that most adults had their own optometrist or ophthalmologist and, thus, providing these services would duplicate what the family was already accessing.

As now constructed, the Down syndrome clinic model used at the Adult Down Syndrome Center calls for each adult to be seen by a family practice physician, a social worker, an audiologist, and a nutritionist. Several assessments are carried out as part of normal data gathering. For example, a health maintenance checklist (based on The Down syndrome Medical Checklist, Ohio/Western PA Down Syndrome Network, 1992) is completed. This checklist outlines appropriate health screening guidelines and is used to guide the health care provided. Questionnaires are used that focus on health problems that are seen more commonly in persons with Down Syndrome. A social services review is carried out using the Checklist for Psycho-social Concerns, which is administered in a structured interview with a family member or care provider or both and the adult. This interview screen is an adaptation of the diagnostic criteria for depression and other potential problem areas taken from the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV; American Psychiatric Association[APA], 1994). An adaptation of a standardized behavioral assessment instrument, the Developmental Disabilities Profile (Brown, Hanley, Nemeth, Epple, Bird, & Bontempo, 1986), is also used to assess adaptive and living skills. In addition to the annual evaluation by the physician, social worker, nutritionist, and audiologist, thyroid blood tests are ordered annually. Neck X rays are used to evaluate atlanto-axial instability and are administered on all new patients seen as needed. Hepatitis B screening and hepatitis immunizations are performed if indicated. Hepatitis eye exams are carried out once every one two years. Other routine
preventive medical services are performed on a schedule indicated for the general population.

Personal well-being and an emphasis on health and wellness are foundations of the clinic's activities. Since hypothyroidism, atlanto-axial instability, chronic hepatitis, and other health problems are common in people with Down syndrome and can lead to physical or mental Deterioration of the health status reviews. We have observed that the majority of the adults seen at the clinic are living healthy lives. In addition to the benefiting from the availability of medical and psychosocial services, we have found that social opportunities afforded adults with Down Syndrome are correlated with the being healthy (Fujiura, Fitzsimons, Marks & Chicoine, 1997).

The clinic opened in 1992 with at least two adults being seen each of two mornings a month. The demand for the services has grown steadily, and the clinic staff now sees referees five days a week. The clinic is operated by Advocate Health Care, a not-for-profit health system, with additional funding from Advocate Medical Group, a large multispecialty medical group, and the Advocate Foundation. In addition to providing the services of the physicians, the office staff, the audiologist, and nutritionist, Advocate Health Care provides for the office and other office expenses. NADS provides funding for a parent who serves as an advocate who advises the adults, families, and care providers and helps them obtain services. NADS also provides partial funding for the services of the social worker. The University of Illinois at Chicago, another partner in the clinic, provides additional funds for the services of the social worker, a consulting psychiatrist, and a postdoctoral research fellow.

All of the collaborating organizations work together to provide several other services, a resource center, and educational programs for parents, care providers and professionals. The exercise programs have been beneficial in helping adults lose weight and improve cardiopulmonary fitness and providing an opportunity to socialize. The group sessions, during which the families meet in one room and the adults with Down syndrome meet in another, each led by a social work student, have been very helpful for both groups to discuss common concerns, support each other, and learn more about solving problems that they have encountered. The staff of the clinic has seen more than 400 adults with Down syndrome and have become familiar with the similarities and differences of persons with Down syndrome to persons in the general population with regard to general health care and, specifically, decline in function and Alzheimer's disease.

The clinic was developed and continues to function through a cooperative effort of a variety of organizations. The input of the parents is extremely important to the approach used to provide care as well as to the success of the clinic. The majority of the referrals have to come through NADS. The staff and parents of NADS continue to advise on the functioning of the clinic and in developing additional programs to meet the needs expressed by adults, families, and careers, and clinic personnel.
Rationale for Thorough Screening and Evaluation

When a patient declines in function, a thorough evaluation is necessary to look for reversible causes or, if no reversible cause is found, to confirm that the decline is consistent with Alzheimer's disease. Alzheimer's disease is a diagnosis of exclusion in persons with Down syndrome, just as it is in the general population. Traditional neuropsychological testing, used for persons in the general population who are suspected of having Alzheimer's disease, is less helpful for assessing persons with an intellectual disability (Janicki, 1994) and, therefore, often is not helpful in making the diagnosis. Therefore, careful clinical evaluation is necessary to avoid missing potentially reversible causes of the decline. The medical history and physical exam provide insight into the decline, evaluate for associated symptoms, and initiate the evaluation for etiology. Vision and hearing evaluations are important because loss of a sense can be very problematic for a person with an intellectual disability who has limited reserve (i.e., limited cognitive function) with which to compensate. Blood work should include thyroid function, vitamin B12, folic acid, CBC, and chemistry profile. A computerized tomography scan or magnetic resonance image scan can evaluate for intracranial lesions or atrophy. A lateral neck X-ray in neutral, flexion, and extension is indicated to evaluate for atlanto-axial instability.

In addition, careful screening for psychological or social problems is a critical part of the evaluation. Mental health disorders often present differently in persons with Down syndrome and other disabilities because of cognitive and expressive language limitations in this population (McGuire & Chicoine, 1996; Sovner, 1986; Vitello, Spreat, & Behar, 1989; Wetzler & Katz, 1989; Zajecka & Ross, 1995). As a result, mental health disorders may be difficult to distinguish clinically from Alzheimer's dementia. For example, the following symptoms are common to both depression and Alzheimer's dementia in this population: loss of adaptive skills, disruption of sleep cycle and appetite changes, apathy, moodiness, irritation, Aggressiveness, psychomotor agitation or retardation, memory loss, and the presence of psychotic features (such as extreme withdrawal, delusions, and an increase in hallucinatorylike self-talk) (McGuire & Chicoine, 1996: Pary, 1992). Alzheimer's dementia is particularly difficult to rule out because there is no definitive test for this disorder (Burt & Aylward, this volume; Dalton, Seltzer, Adlin, & Wisniewski, 1994). To further complicate matters, depression may coexist with Alzheimer's dementia (Burt, Loveland, & Lewis, 1992; also see Burt, this volume). In this case, prompt treatment of depression will preserve functioning for some time, even though a downhill course may be inevitable (Evenhuis, 1990; storm, 1990; Warren, Holroyd, & Folstein, 1989.

Despite these difficulties, depression and other mental health disorders have been diagnosed in previous case studies (Collacott, Cooper, & Mcgrother, 1992; Harris, 1988; McGuire & Chicoine, 1996; Myers & Pueschel, 1991; Szymanski & Biederman, 1984; Warren et al., 1989). These reports demonstrate that diagnosis of mental health disorders is enhanced when behaviors, rather than subjective feelings, are emphasized as criteria.
and when care is taken to rule out all other sensory deficits and medical conditions such as Alzheimer's dementia or hypothyroidism. Additionally, results from previous case studies suggest that the differential diagnosis of depression and Alzheimer's dementia is enhanced if close attention is paid to the symptom course. Depression tends to show an up-and-down pattern of decline, which will show improvement with time and an eventual return to premorbid levels of functioning with time and treatment. Symptoms of Alzheimer's dementia tend to fluctuate up and down in the early stages, but over time will show a progressive and nonreversible pattern of decline (Evenhuis, 1990; Pary, 1992; Warren et al., 1989).

**A Review of the Cases Seen at the Clinic**

A review of diagnosed disorders from the current clinical sample Adult Down Syndrome Center supports previous case reports showing reversible disorders, such as depression, to be a predominant cause of loss of functioning of this sample. Of the 443 adults with Down Syndrome seen at the Adult Down Syndrome Center, 148 (33%) have presented with a decline in function. Of the 148 with a decline in function, only 11 individuals (2.5% of the 443 seen have shown a progressive and nonreversible decline and deterioration over time which would merit the diagnosis Alzheimer's dementia. Several of these 11 initially showed a reduction in depressive symptoms in response to treatment, but the decline in function continued and all were later diagnosed with Alzheimer's dementia. For the remaining 137 of the 148 who were also presented with a decline in function, all have shown significant improvement, or a return in premorbid state of functioning in response to treatments, which would not suggest Alzheimer's dementia.

Many of the 148 individuals with a decline in function had more than one health problem that caused or contributed or both to the decline. A mood disorder was the most commonly diagnosed problem. Seventy-six individuals, representing 51% of the 148 with a decline in function, were diagnosed with mood disorder, which most often anxiety or obsessive compulsive disorder. Including those with a mood disorder, 82 people (55%) had loss of function from a mental health disorder alone. Of the rest of the 148 who presented with a decline in function, 42 (28.5%) had a mental health disorder and a medical disorder 13(9%) had only a medical disorder (other than Alzheimer's disease. The 148 adults received a total of 247 diagnoses (see table 15.1). Treatment of multiple problems was necessary in some individuals to achieve improvement or a return to their premorbid state.

There are a number of additional findings from the Adult Down Syndrome Center sample that have a bearing on the issue of Alzheimer's dementia in Down Syndrome. As shown in Table 15.2, the Adult Down Syndrome Center sample shows a large number of individuals who are over the age of 30, who many professionals and careers believe to be at greater risk for Alzheimer's dementia. Of these, 307 (69%) adults were over the age of
30, while 171 (39%) were over the age of 40. Table 15.3 shows the diagnosis category of all the adults and for those over the age of 40 who presented with a loss of function. In our sample, the incidence of Alzheimer's dementia ranged from seven (6%) for individuals in their 40s, to two (13%) for individuals over the age of 60 (See table 15.4). In the general population, the incidence of Alzheimer's disease ranges from 10% for persons in their mid 60s to up to 40% for persons over the age of 80 years (Evans et al., 1990). Our sample may not be representative of all persons with Down syndrome; therefore, we cannot make a statement about comparing our population with the general population, it is However, in interesting to note that our population has a similar rate of Alzheimer's dementia, with an increase with age, except that it seems to occur 20 twenty years earlier. Our observations also suggest that some people with Down syndrome seem to age, more rapidly as they reach their middle 30s. In many of our adults who are in their middle 30s, we have observed graying hair, physical slowness, and other changes associated with aging. In addition, the estimated life expectancy of persons with Down syndrome is approximately 20 years less than the life expectancy for the general population (Baird & Sadovnick, 1988; Chicoine & McGuire, 1997; Janicki, Dalton, Henderson, & Davidson, In press). Therefore, the physical age of individuals with Down syndrome who are 40 or 50 years of age may be equivalent to individuals in the general population who are chronologically 60 to 70 years of age. Comparing the prevalence of Alzheimer's dementia by age of our sample with the prevalence in the general population suggests that the rates may be similar except that the higher rates with age in our sample occur approximately 20 years earlier. If the prevalence of our sample is comparable with the prevalence in all persons with Down syndrome at any given age may be explainable by accelerated aging with an earlier onset of Alzheimer's disease. Therefore, the rate of Alzheimer's dementia in persons with Down syndrome should be compared with the rate for the cohort 20 years older in the general population.

What may be a more compelling finding from the Adult Down Syndrome Center sample is the incidence of decline for older adults with Down syndrome that is not attributable to Alzheimer's dementia but to other reversible disorders. For example, for individuals more than 40 years of age in the sample, 53 out of 171 (30.9%) presented with a decline in function (see table 15.4). Of this group, only 11 individuals out of 53 (21%) showed a pattern of continued decline and deterioration in functioning suggestive of Alzheimer's disease. The remaining 42 individuals (79%) with a decline in function were found to have reversible disorders that were responsive to treatment. In our sample, if the diagnosis of Alzheimer's disease was given based on an assumption that all persons with Down syndrome develop Alzheimer's disease rather than on a thorough evaluation, then more than three-quarters of those that presented with a decline in function would have received an inappropriate diagnosis of Alzheimer's dementia. As Szymanski (1988) warned, the result of this misdiagnosis of an "untreatable disease" may be to offer no treatment at all or to offer ineffective medications (such as an antipsychotic medication). In either case, this strategy could result in an increasing loss of functioning, which is then viewed by the practitioner as further evidence of Alzheimer's disease.
Some case studies are presented to highlight the importance of the complete evaluation. For example, a 35-year-old man was seen at his house because he would not leave his home and rarely left his bed. He would eat only beans and had a dramatic decline in function. His thyroid stimulating hormone (TSH) level was high, and he was diagnosed with hypothyroidism with depression. He was started on levothyroxine, and an occupational therapist was consulted to help him become ambulatory again. Over the next year he responded to the treatment, became ambulatory again, and started going outside.

Another example is a 38-year-old woman who was seen for a decline in function. The history revealed that she was very independent and came home from work each day on public transportation. She admitted (and the staff was able to confirm) that she disembarked from the bus one stop early each day and went into the local pub for multiple alcoholic drinks. She was diagnosed with depression probably secondary to her alcohol consumption. With the consent of her family, the staff was able to change her transportation and she stopped drinking. Interestingly, she later forgot or denied that she had ever drank alcohol, and she is doing well without any evidence of craving alcohol.

In yet another example, we encountered a 43-year old man who presented with withdrawal, frequent crying, decline in function, and loss of memory. Within two or three months his depressive symptoms responded to antidepressant medications. However, his memory and decline in function continued on a downhill course and he developed seizures, gait apraxia, and incontinence and he was eventually given the diagnosis of probable Alzheimer's disease.

In this last example, a 52-year-old woman was brought by the staff of her residential facility for evaluation of a decline in function and a concern that she had Alzheimer's disease. She was losing money, bills that she previously paid without problem, and other items. The work-up revealed no clear etiology; however she did not clearly fit the criteria for Alzheimer's disease. No diagnosis was given to her, and over the next 18 months there was no further significant decline. During this time, the carpeting in her room was changed and in the process the furniture was moved. Staff discovered the missing money, bills, and other objects neatly placed in small plastic bags and hidden in many places in her room. She was given the diagnosis of obsessive compulsive disorder and started on fluvoxamine (Luvox). Within a few weeks the staff stated that she was much better at attending to her finances and overall seemed much improved in her daily functions.

The criteria we use to determine a probable diagnosis of Alzheimer's disease include progressive decline in function, progressive memory loss, gait apraxia, incontinence of urine or stool or both, and seizures. While psychological symptoms often accompany Alzheimer's disease, we avoid making the diagnosis on the basis of these alone because of the difficulty differentiating between their etiology and the diagnosis of psychological and behavioral problems.
Care management

When an adult with Down syndrome is diagnosed with Alzheimer's disease, we focus on "The four S's": safety, stability (of the environment), social (emotional) issues, and symptoms. As the adult with Down syndrome experiences greater decline, family or carers will often have difficulty maintaining a safe environment. Falling because of gait apraxia, wandering from home, and touching or using object in the home that can be harmful if used inappropriately (e.g., the stove) can all be problems that the family or carer cannot prevent. In addition, when the adults become bed bound, frequent turning or changing the position to prevent bed sores is necessary and may be more than the family can provide. Safety is frequently the concern that requires a change in living arrangement and, if no specialty care programs are available, may require admission to a nursing facility.

As the person declines, stability of the environment helps reduce confusion. Persons with Down syndrome often have difficulty with change in environment including changes in career and location. This is no less true for persons with Down syndrome who develop Alzheimer's dementia. Negotiating a new environment is more difficult and increases confusion and limits the remaining skills. This frequently presents a dilemma because the present environment may no longer be safe, but a new environment may increase confusion. We recommend maintaining the individual in the present environment as long as it is safely possible, making only minor changes as necessary and using canes, walkers, handrails, or other devices to prolong the length of time that the adult remains ambulatory and is able to remain in his or her present environment.

Emotional supports are also important. These include supporting the family and carers who are dealing with their own grieving issues of loss of the individual with Down syndrome. The rate of decline for a person with Down syndrome who develops Alzheimer's disease may be more rapid than in the general population, and family or carers may require a great deal of emotional support to deal with the change.

Another important social issue involves keeping the individual with Down syndrome as much involved in activities as possible. Keeping the adult involved in activities reflects the goal of maximizing function by stimulating the person at a level that is challenging but not overwhelming. Regular assessment is needed to determine the level of stimulation of activities to meet the ever-changing level of skill. Regular small changes seem to be beneficial so as not to confuse the patient but at the same time meeting the changing needs.

Incontinence is a problem and is even seen in some adults with Down syndrome who have no other decline in function. Therefore, evaluation for other causes can help reduce or delay this problem. Obstruction from benign prostatic hypertrophy or other causes, detrusor dysfunction, infections, and urinary dysfunction secondary to atlanto-
axial subluxation should also be considered. For those whom no reversible cause is found toletting regimens may be helpful.

Seizures will generally respond to standard medications. Tonic-clonic seizures are frequently seen but adults also may have petit mal seizures as well as less pronounced "jerking" type movements. These abnormal movements have also tended to respond to antiseizure medications. One woman had jerking or twitching movements of her arms and her body that actually caused her to fall or flip out of bed. These momentary abnormal movements responded nicely to phenytoin and helped prevent further injuries.

Many adults will also have psychological symptoms such as depression, psychotic features, and anxiety. Sleep disturbance and changes in eating are common. An evaluation for other causes will sometimes find a problem in addition to the Alzheimer's disease. Medical problems such as hypothyroidism, infections, peptic ulcers, and others may alter sleep, appetite, or behavior. Environmental issues may also contribute to these symptoms. Again, a careful evaluation of the environment is necessary to maximize function without overwhelming the individual's declining abilities. Antidepressants or antipsychotic medications are sometimes necessary to improve the function or reduce the symptoms. Using the adverse-effect profile of each to guide the selection of medication reduces the symptoms while minimizing additional problems. For example, for adults who develop depression and have problems with sleep, use of amitriptyline or other sedating antidepressants at night may benefit the sleep disturbance directly as well as treat the depression. However, another factor in the selection of the drug is that adults with Down syndrome do seem to be particularly sensitive to the anticholinergic side effects of medications, and many of the antidepressants and antipsychotics have adverse effects that could further reduce skills.

**Commentary**

Careful evaluation of a person with Down syndrome who has a decline in function has broader public health implications. Many of the adults we have seen for a decline in function have either previously been given the diagnosis of Alzheimer's disease or there was a strong suspicion by the family or carers that the person had Alzheimer's disease. In discussing the concern with the family or carer, it is clear that many people believe that all persons with Down syndrome will develop Alzheimer's disease. Our experience and the research of others would suggest that not all persons with Down syndrome will develop clinical dementia stemming from Alzheimer's disease. The exact prevalence of Alzheimer's disease in persons with Down syndrome is still being investigated.

Unfortunately, the belief that all persons with Down syndrome get Alzheimer's disease appears to guide the evaluation of some people with Down syndrome who are declining in function. The misdiagnosis of Alzheimer's disease tends to increase the
misperception that all people with Down syndrome will develop Alzheimer's disease. The use of this cyclical logic leads to more diagnoses of Alzheimer's disease. We have heard repeated stories of persons with Down syndrome who were declining in function, were given a brief evaluation, and were then given a diagnosis of Alzheimer's disease. The potentially reversible causes were not ruled out. Much of the decline in function that we have seen experienced by persons with Down syndrome is reversible, and the quality of the person's life can often be improved. Even if some of these people later develop Alzheimer's disease, they often receive benefit in the interim from treatment of reversible disease processes. The diagnosis of Alzheimer's disease is still a diagnosis of exclusion, and a thorough evaluation for potentially reversible causes for a decline in function is indicated.

A close working relationship with the families and carers of the adults with Down syndrome has provided insight into their needs and leads us to argue for a multidisciplinary approach to diagnosis and care management. We have found that such a multidisciplinary approach can be instrumental in providing for the diagnosis of adults with Down syndrome who present with a decline in function. In our sample, there were a variety of medical and psychosocial problems causing the decline, and the team approach helped provide insight into these problems. Both a thorough medical and psychosocial evaluation are important to avoid missing reversible causes and overdiagnosing Alzheimer's dementia. Further evaluation of this and other approaches will aid in developing improved services to adults with Down syndrome.

References


