Prevalence of Epilepsy in Adults With Mental Retardation and Related Disabilities in Primary Care

Suzanne McDermott  
University of South Carolina  

Robert Moran, Tan Platt, Hope Wood, Terri Isaac, and Srikanth Dasari  
The University of South Carolina School of Medicine

Abstract

Two primary care practices were used to recruit adults with and without disability. Disability groups included autism, Down syndrome, cerebral palsy, and mental retardation. The patients without disability had an epilepsy prevalence rate of 1%. The prevalence of epilepsy within the disability groups was 13% for cerebral palsy, 13.6% for Down syndrome; 25.4% for autism, 25.5% for mental retardation, and 40% for adults with both cerebral palsy and mental retardation. During the decades of adulthood, the prevalence of epilepsy declined for those with cerebral palsy and mental retardation. The prevalence of epilepsy increased with advancing years for adults with Down syndrome, autism, and those without disability. Nonetheless, during each decade the prevalence of epilepsy was higher in all of the disability groups compared to those without disability.

Researchers have reported that 4% to 10% of all children experience a seizure at some time in their lives. By age 20, however, only 1% of the population has a diagnosis of seizures or epilepsy (Hauser, 1994). The distinction between seizures and epilepsy is based on a number of events. A seizure is defined as a behavioral, motor, sensory, or cognitive event that is due to abnormal neuronal activity characterized by excessive hypersynchronous neuronal discharges. Epilepsy is defined as recurrent seizures secondary to central nervous system dysfunction.

Investigators have studied the co-morbidity of epilepsy and numerous childhood onset conditions, including cerebral palsy, autism, Down syndrome, and mental retardation (Brodtkrøb, 1994; Johannsen & Christensen, 1996; Volkmar & Nelson, 1990). Recent reports about children with cerebral palsy indicate that 35% have a history of epilepsy, with the highest prevalence in children with spastic hemiplegia (66%), followed by those with quadriplegia (43%), and diplegia (16%) (Singh, Jagirdar, Khandelwal, & Malhi, 2003). The prevalence of epilepsy in children with autism is estimated to be 5% to 38%, with age, cognitive level, and type of language disorder having the greatest predictive power (Acardi, 1994; Deykin & MacMahon, 1979; Mouridsen, Rich, & Isager, 1999; Tuchman & Rapin, 2002; Volkmar & Nelson 1990). The prevalence of epilepsy in Down syndrome has been reported to be 8% to 9%, with a bimodal distribution; 40% start seizure activity before the age of 1 year and another 40% start seizures between 20 to 30 years of age (Pueschel, Louis, & McKnight, 1991). As is the case with autism, prevalence is age-related, and the proportion of adults with Down syndrome over the age of 50 years who have seizures is reported to be 46% (McVicker, Shanks, & McCleland, 1994). In a review article, Bowley and Kerr (2000) indicated that the prevalence of epilepsy in adults with intellectual disability ranged from 18.3% to 44%. In
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the most recent report, Morgan, Baxter, and Kerr (2003) reported that 16% of individuals with mental retardation have epilepsy.

The literature on the prevalence of seizures and epilepsy relies on data from specialty settings, institutions, and registries. In contrast, there are few investigators who describe the experience of adults with disabilities and epilepsy, who are receiving primary care in the community, alongside persons without disabilities.

We designed this study to contrast the age-specific prevalence and severity of epilepsy in adults with cerebral palsy, autism, Down syndrome, and mental retardation with adults who had no diagnosed disability, all of whom received primary health care in the same setting during adulthood. The hypothesis underlying this study was that the adults with lifelong disabilities would have higher prevalence rates for epilepsy during each decade of adult life compared to adults without disability.

Method

We used a retrospective cohort design to analyze the prevalence of epilepsy among adults with and without lifelong disabilities receiving primary health care in either a large urban family practice center or a small rural practice, before or during the 1990s through 2003. The two participating practices in this study were university teaching programs for medical students and residents. There was a focus on providing primary care to members of the community, with particular attention to persons with disabilities. The population in these practices were composed of 58% women and 42% men, of whom 35% were African American, 2% Spanish-speaking, and 3% Asian.

During the study period, the medical director of the urban practice was also the medical director for a large community service provider for individuals with mental retardation, autism, and related disabilities. He was responsible for the medical care for approximately 600 individuals living in Intermediate Care Facilities for the Mentally Retarded (ICFs/MR) and other supervised placements in two counties. Because of his expertise, high quality care for people with disabilities was emphasized in the instruction of medical students and residents. The rural practice also provided primary care services for many county residents with developmental disabilities. All family medicine residents, attending physicians, and nurse practitioners were assigned patients with disabilities and instruction on special needs was ongoing. As a result, a large number of people with moderate, severe, and profound mental retardation and related disabilities were part of these practices.

Participants

Patients with and without a primary disability were selected from a pool of over 58,000 individuals who received care during the study period (1990 to 2003). There were 51,146 individuals at the urban site and 7,851 individuals at the rural site. Patients without a disability were selected after all the patients with a disability were identified in order to use age-group stratification, which was done in 10-year increments based on age at entry into the clinics.

Case Definitions

Primary disabilities were defined as birth onset or acquired disabilities that are permanent and lifelong. The primary disabilities included in this analysis are mental retardation, Down syndrome, cerebral palsy, and autism. A secondary condition is defined as any condition to which a person with a primary diagnosis is more susceptible and may include medical, physical, emotional, family, or community problems (Lollar, 2001). Epilepsy is considered a secondary condition in this study. The case definitions used for selection of patients with a disability relied on identification of International Statistical Classification of Diseases (9th ed.)–ICD-9 codes and physician-identified conditions (World Health Organization, 2000).

Epilepsy. Epilepsy was identified in both progress notes and problem lists and/or by the presence of the ICD-9 Code 345. The severity of epilepsy was determined by the medications prescribed and the number, frequency, and type of seizures. Because the study subjects were established patients in the two primary care clinics, physicians had more than a history of epilepsy to make a diagnosis. Most patients had consultation notes from a neurologist, EEG reports, CAT scan, and other diagnostic studies. Two levels of epilepsy were used for coding: moderate and severe. If one or two drugs were required to control the epilepsy and there were few and infrequent breakthrough or partial seizures, the individual was considered a patient with moderate epilepsy. If the epilepsy activity required increasing doses
and/or at least three medications or use of medical device (e.g., vagal nerve stimulator) for control, the individual was coded as a patient with severe epilepsy.

**Cerebral palsy.** Cerebral palsy was identified in both the progress note and problem list and/or by the listing of the ICD-9 Code 343. Cerebral palsy severity was based upon physical limitations, impact on activities of daily living, and classification of cerebral palsy by anatomical distribution and neurological involvement. Mild cerebral palsy was characterized by mild motor impairment that required minimal assistance and did not prevent employment as well as independent functioning for most activities of daily living. A rating of moderate cerebral palsy was noted if an individual required a cane, walker, or intermittent wheelchair use and the assistance of others in performing many of the usual daily activities. If a person required a wheelchair at all times and a caretaker for constant assistance in daily activities, the level of cerebral palsy was determined to be severe. Spastic quadriplegia and dyskinetic cerebral palsy were usually rated as severe.

**Autism.** Autism was identified in both progress notes and problem lists and/or by the listing of the ICD-9 Code 299. Severity of autism was coded mild, moderate, and severe based on ability to perform activities of daily living (ADLs), level of social skills, employment status, and the most recent standardized score on a rating scale for autism. If the individual was identified as having mental retardation and autism, IQ was considered in the severity rating. Physician progress notes, psychiatric consultant reports, and other reports in the record provided information to make a determination of severity. Mild autism was coded if the individual worked, had an independent life, and needed minimal assistance in performing everyday activities. Moderate autism was coded if the individual was independent or with intermittent and needed limited assistance and did not prevent employment and recreation in the community. Severe autism was coded if the individual did not have any history of employment and required extensive supports. Severe autism was coded if the individual needed pervasive supports because of his or her autism and/or severe mental retardation. These individuals had minimal verbal abilities and social skills and severe aggressive behaviors.

**Mental retardation.** Mental retardation was identified in progress notes, problem lists, and consultation forms from service providers and other physicians. The ICD-9 Codes 317, 318, and 319 were listed in the medical record. Mental retardation severity was coded mild (ICD-317), moderate (ICD-318.0), or severe/profound (ICD-318.1, 318.2) based on the most up-to-date record of IQ and level of supports needed (intermittent, limited, extensive, or pervasive). Level of supports was based on descriptions in the record about the individual’s ability to perform ADLs and participate in employment and recreation in the community. Mild mental retardation included individuals who had IQs of 56 to 75, could perform ADLs, and worked and participated in the community independently or with intermittent supports. Moderate mental retardation included individuals who had IQs of 41 to 55, required some assistance with their routine ADLs, and needed limited or extensive support for community participation. In addition, many people with moderate mental retardation worked with supervision in a mobile work crew, enclave, or workshop. A diagnosis of severe mental retardation was given to individuals who required pervasive support and had IQs of 40 and below, performed only simple tasks with close supervision, and lived with substantial supports, primarily in a group home or with their family.

**Down syndrome.** Down syndrome was identified in both progress notes and problem lists and/or the ICD-9 Code 758.0 was listed. Severity was based on the individual’s level of mental retardation.

**Comparison patients without a disability.** Comparison patients were selected after all the patients with a primary disability were identified and removed from the pool of potential subjects. This was done to assure that comparison patients did not have a disability. The cases were stratified according to age when their first medical record was available. Using a random number generated list, we conducted random selection in each age strata from approximately 55,000 patients.

**Procedure**

The study, which was approved by the Institutional Review Board, included a medical record review and an interview for a subset of the patients with and without a disability. The record review relied on both computerized medical records for the decade of the 1990s to the present, and the companion paper records that included archived records from earlier medical care, often dating back to childhood. Thus, some patient medical records spanned over 30 years. In addition, a sample of patients consented to participate in an interview to validate onset dates for condi-
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Table 1. Characteristics of Patients by Disability Group

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Without disability (n = 1806)</th>
<th>With disability (n = 741)</th>
<th>Austim (n = 55)</th>
<th>Down syndrome (n = 59)</th>
<th>CP with MR (n = 165)</th>
<th>CP (n = 23)</th>
<th>MR (n = 439)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Entry age mean (SD)</td>
<td>39.4 (12.0)</td>
<td>36.5 (13.9)</td>
<td>25.5 (11.2)</td>
<td>38.0 (12.7)</td>
<td>32.3 (12.7)</td>
<td>32.5 (13.4)</td>
<td>36.9 (14.3)</td>
</tr>
<tr>
<td>Years follow-up (mean)</td>
<td>7.0</td>
<td>12.4</td>
<td>9.6</td>
<td>12.2</td>
<td>12.2</td>
<td>8.9</td>
<td>9.7</td>
</tr>
<tr>
<td>Gender (%)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>44.4</td>
<td>53.3</td>
<td>76.4</td>
<td>59.3</td>
<td>47.2</td>
<td>52.2</td>
<td>52.8</td>
</tr>
<tr>
<td>Female</td>
<td>55.6</td>
<td>46.7</td>
<td>23.6</td>
<td>40.7</td>
<td>52.8</td>
<td>47.8</td>
<td>47.2</td>
</tr>
<tr>
<td>Race (%)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>White</td>
<td>46.5</td>
<td>59.2</td>
<td>70.9</td>
<td>72.9</td>
<td>61.8</td>
<td>56.5</td>
<td>55.1</td>
</tr>
<tr>
<td>AA</td>
<td>49.8</td>
<td>39.5</td>
<td>26.8</td>
<td>25.4</td>
<td>37.6</td>
<td>39.1</td>
<td>43.7</td>
</tr>
<tr>
<td>Other</td>
<td>3.7</td>
<td>1.3</td>
<td>2.3</td>
<td>1.7</td>
<td>0.6</td>
<td>4.4</td>
<td>1.2</td>
</tr>
<tr>
<td>Severity of primary condition (%)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mild</td>
<td>n/a</td>
<td>34.9</td>
<td>18.2</td>
<td>10.2</td>
<td>24.9</td>
<td>47.8</td>
<td>43.5</td>
</tr>
<tr>
<td>Moderate</td>
<td>n/a</td>
<td>26.7</td>
<td>72.7</td>
<td>15.2</td>
<td>34.5</td>
<td>26.1</td>
<td>19.6</td>
</tr>
<tr>
<td>Severe</td>
<td>n/a</td>
<td>36.6</td>
<td>9.1</td>
<td>74.6</td>
<td>40.6</td>
<td>26.1</td>
<td>36.9</td>
</tr>
</tbody>
</table>

Note. CP = cerebral palsy, MR = mental retardation, AA = African American, n/a = not available.
The years of follow-up reveal how long each group had records available for review. Patients with a disability had records for 12.4 years; those without a disability, 7 years on average. There were five primary disability conditions: autism, Down syndrome, cerebral palsy with mental retardation, cerebral palsy, and mental retardation. These groupings were mutually exclusive and summed to 741 patients with a disability. Table 1 presents the severity of the mental retardation and related disability, ranging from only 10.2% with mild and 74.6% with severe mental retardation for patients with Down syndrome to 47.8% mild and 26.1% severe mental retardation for patients with cerebral palsy.

Table 2 shows that the overall prevalence of epilepsy is substantially higher for all the groups whose members had a disability, ranging from 13.6% for adults with Down syndrome to 40% for those with cerebral palsy and mental retardation compared to the patients without a disability (1.0%). The vast majority of epilepsy cases were classified as moderate, responding to one or two medications to achieve control. The group with cerebral palsy and mental retardation had the highest proportion of severe epilepsy (9.1%), requiring three or more medications, a vagal nerve stimulator, or other medical device or surgery. It is noteworthy that there were no patients with severe epilepsy in the groups with Down syndrome or cerebral palsy and only one patient with severe epilepsy in the group with autism. Table 2 also shows that the patients with cerebral palsy and mental retardation, cerebral palsy, or mental retardation had a declining prevalence of epilepsy over the decades compared to the increasing prevalence for patients without disabilities. We conducted a test of trend to compare each disability group’s tendency to increase or decrease prevalence of epilepsy as they age. The test statistic compared each group to the increasing prevalence of epilepsy observed in the patients without disability. Patients with autism, \( p = .405 \), and Down syndrome, \( p = .724 \), had an increasing prevalence of epilepsy over the decades and, thus, the test of trend was not significantly different from those without a primary disability. The trend toward decreasing prevalence with age was statistically significant for patients with both cerebral palsy and mental retardation, \( p = .001 \), cerebral palsy, \( p = .005 \), and mental retardation, \( p = .001 \).

Figure 1 shows the prevalence in the patients with and without disabilities, from age 20 to 60+.

The patients with cerebral palsy and mental retardation had the highest prevalence rates in 3 decades, 20 to 49 years. Patients with autism had the highest prevalence rates for the decade of 50 to 59 years, and patients with Down syndrome had the highest prevalence after age 60 years.

The relative risk for epilepsy for each of the groups was 14 to 61 times higher for individuals with mental retardation and related disabilities compared to patients without disability, after controlling for age. The group with the highest relative risk of having epilepsy was cerebral palsy with mental retardation, relative risk = 61.7, 95% confidence interval (CI) = 35.3 to 107.7. The relative risks for the other groups were as follows: autism, 39.7, 95% CI 18.7 to 84.3; mental retardation, 33.5, 95% CI 20.2 to 55.4; cerebral palsy, 14.7, 95% CI 4.0 to 54.0; and Down syndrome, 14.1, 95% CI 18.7 to 84.3. Although the CIs of the estimates are wide, their low ends are significantly higher than those of the control group. In all cases the probability of chance occurrence for these estimates was less than .0001.

All patients with moderate and severe epilepsy were taking at least one antiepileptic medication. For those with moderate epilepsy, the top three medications were Carbamazepine, Phenytoin, and Phenobarbital. For treatment of severe epilepsy, the top three medications were Valproic Acid, Phenobarbital, and Phenytoin (tied with Clonazepam). We note that a vagal nerve stimulator was also prescribed for patients with severe epilepsy.

**Discussion**

The use of a large urban and a rural primary care medical practice to estimate prevalence of epilepsy allowed us to compare the prevalence rate between primary disability groups and the comparison group of patients without disabilities. The medical practices used in this study have an over-representation of adults with disabilities because of the emphasis placed on providing care for patients with disabilities, although the practices are exclusively family medicine. Overall, the prevalence rate for epilepsy, in the groups with disability and the group without disability, are similar to reports in the literature (Hauser, 1994; Morgan et al., 2003).

One of the limitations of our approach was the variable observation time in each age group, as shown in Table 2. This occurred because people entered the practice at different ages, and they
departed at different ages. Some individuals left the practice for a time and returned, others died, and some were lost to follow-up. Thus, this retrospective cohort study has the limitation of incomplete data or censoring of data at different points in time. On the other hand, because we used carefully abstracted medical records, these data reflect the actual experience of patients in primary care. Further, one of the strengths of our approach was the average follow-up period of over a decade per patient, so physicians were able to distinguish between an isolated or precipitated seizure event and epilepsy. As a result, we were able to code epilepsy with seizure control (coded moderate) and epilepsy that was difficult to control (coded severe).

In previous research, investigators have grouped patients according to type of epilepsy and seizure frequency. Most clinicians and researchers use the International Classification of Epileptic Seizures (Commission on Classification and Terminology, 1981). This system, still the standard used in current textbooks, is based on the clinical features and electroencephalographic findings and is divided into partial seizures, primarily generalized seizures, and unclassified seizures (Lowenstein, 2001). Many researchers have distinguished the types of epilepsy (grand mal, psychomotor, infantile spasms) and reported on the epidemiology of the types, singly and in combination (Mouridsen et al., 1999). One investigator used a functional classification of epilepsy that included duration of time within which seizures occur, frequency of seizures within that time, use of medication and occurrence of side effects, and severity of the seizures. The severity measure was a general indicator of the degree to which the usual activities of a person are interfered with as a result of having epilepsy (Richardson, Koller, Katz, & McLaren, 1981). Other researchers have classified epilepsy based on frequency of seizures; grouping severe epilepsy as more frequent than once a month; intermediate epilepsy as more frequent than once a year, but less frequent than once a month; and well-controlled epilepsy as less frequent than once a year (Brodtkorb, 1994). Our simpler approach of categorizing epilepsy as moderate (controlled with medication) and severe (difficult to control with medications) conveys similar information with an explicit mention of the number of medications needed to maintain control. This categorization, although not replicated and validated, could prove a useful way of conveying information about the disabling potential of epilepsy.

Because we had an average of 12 years of follow-up time for patients with disabilities, we were able to explore prevalence of epilepsy during 4

### Table 2. Proportion of Patients With and Without Disability Who Have Epilepsy by Severity and Age

<table>
<thead>
<tr>
<th>Group</th>
<th>Without disability</th>
<th>With disability</th>
<th>Autism (n = 55)</th>
<th>DS (n = 59)</th>
<th>CP &amp; MR (n = 165)</th>
<th>CP (n = 23)</th>
<th>MR (n = 439)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Moderate epilepsy (%)</td>
<td>0.83</td>
<td>23.3</td>
<td>23.6</td>
<td>13.6</td>
<td>30.9</td>
<td>13.0</td>
<td>22.3</td>
</tr>
<tr>
<td>Severe epilepsy (%)</td>
<td>0.17</td>
<td>4.0</td>
<td>1.8</td>
<td>0.0</td>
<td>9.1</td>
<td>0.0</td>
<td>3.2</td>
</tr>
<tr>
<td>Epilepsy by age group</td>
<td>n (%)</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>20–29</td>
<td>4/357 (1.12)</td>
<td>75/265 (28.3)</td>
<td>8/29 (27.6)</td>
<td>2/25 (8.0)</td>
<td>28/70 (40.0)</td>
<td>1/8 (12.5)</td>
<td>36/133 (27.1)</td>
</tr>
<tr>
<td>30–39</td>
<td>7/626 (1.12)</td>
<td>89/297 (31.0)</td>
<td>1/16 (6.3)</td>
<td>3/29 (10.3)</td>
<td>35/69 (50.7)</td>
<td>1/7 (14.3)</td>
<td>49/176 (27.8)</td>
</tr>
<tr>
<td>40–49</td>
<td>12/711 (1.69)</td>
<td>83/267 (31.1)</td>
<td>2/11 (18.2)</td>
<td>5/21 (23.8)</td>
<td>30/60 (50.0)</td>
<td>0/7 (0.0)</td>
<td>46/167 (27.5)</td>
</tr>
<tr>
<td>50–59</td>
<td>11/512 (2.15)</td>
<td>38/150 (33.3)</td>
<td>1/3 (1.6)</td>
<td>4/16 (2.5)</td>
<td>7/25 (28.0)</td>
<td>0/2 (0.0)</td>
<td>26/104 (25.0)</td>
</tr>
<tr>
<td>60+</td>
<td>5/308 (1.62)</td>
<td>21/108 (7.0)</td>
<td>0/1 (0.0)</td>
<td>2/7 (28.6)</td>
<td>2/15 (33.3)</td>
<td>0/2 (0.0)</td>
<td>17/88 (19.3)</td>
</tr>
</tbody>
</table>

*Note. CP = cerebral palsy, MR = mental retardation, DS = Down syndrome.*
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Figure 1. Period prevalence by patients without and with disabilities and age group. Missing values indicate no patients in the age group. Black bars = 20–29 years old, gray = 30–39, diagonal = 40–49, white = 50–59, and striped bars = 60+ years.

decades of adult life. In a systematic review and meta-analysis of the incidence of epilepsy, Kotsoulos, van Merode, Kessels, de Krom, and Knottnerus (2002) found that the age-specific incidence of epilepsy was highest for the age group over 60 years, second only to the incidence in childhood. One interesting exception is for adults with Down syndrome, whose onset is distinct from onset of epilepsy for individuals with other causes of mental retardation. The overall mean age of onset was 37 years in one study in the United Kingdom, with a bimodal distribution. The first mode was before age 20 and the second, when individuals were in their 50s and 60s. This is important because researchers have found that late-onset epilepsy in Down syndrome is associated with Alzheimer’s disease, and early onset epilepsy is associated with an absence of dementia (Puri, Ho, & Singh, 2001). In the most recent published report, Morgan et al. (2003) found there was a decline in the relative risk of epilepsy among people with intellectual disability compared to the general population, resulting from increased mortality among individuals with co-existing epilepsy and intellectual disability. We found an increasing prevalence of epilepsy over the decades from age 20 to over 60 years for the comparison group, patients with Down syndrome, and those with autism. It is noteworthy that adults with mental retardation, cerebral palsy, and both mental retardation and cerebral palsy had declining prevalence over the decades, similar to the results reported in the literature (Bowley & Kerr, 2000; McVicker, Shanks, & McClelland, 1994; Morgan et al., 2003; Pueschel et al., 1991).

The use of medication to control seizures is central to our classification of moderate and severe epilepsy. Because this study included records of patients seen over decades, the treatment of epilepsy in our patient records reflected different prescription patterns and medication regimes. Many of the patients included in our study had complex neurological conditions, such as cerebral palsy and other developmental disabilities; therefore, multi-drug treatment was often necessary (Deckers, 2002). Further, because behavioral and psychiatric disorders are more prevalent in individuals with mental retardation and epilepsy, careful monitoring of medications is imperative (Devinsky, 2002). We compared the medication used by the study patients with a 1995 publication that contained data from 119 general practitioners. We found that the commonly used drugs were similar, although there was variation in the proportion of patients on each drug (Hart & Shorvon, 1995). In our study more Clonazepam, Valproic Acid, and...
Phenobarbital and less Carbamazepine and Phenytoin were used. The actual clinical management of our patient population is consistent with the International Association for the Scientific Study of Intellectual Disabilities guidelines for the treatment of epilepsy (Lennox et al., 2002).

Results of this study should provide some guidance to primary care physicians and other service providers about expected prevalence of epilepsy in adults with developmental disabilities. It is important for clinicians to know that the prevalence of epilepsy is not constant over the life-span. Patients with cerebral palsy and mental retardation had a declining prevalence of epilepsy over the decades compared to patients without disabilities; and patients with Down syndrome and autism had a steadily increasing prevalence of epilepsy over the decades.

References


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