Chapter 5
The Differential Diagnosis of Epilepsy

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Introduction

Most children and adults seen in a first-seizure clinic or paroxysmal disorders clinic will not have epilepsy. There are as many, and likely many more, imitators and mimickers of epilepsy as there are different types of epileptic seizure. Several studies in adult and pediatric populations have shown high levels of misdiagnosis of epilepsy, consistently on the order of 25–50%\(^1,2\). Unfortunately, the principal reason why many individuals are wrongly diagnosed is a lack of time and effort spent on the difficult art of history taking. In a first-seizure clinic the history takes clear precedence over the neurological examination, which in this context is often of limited value. Physicians must have an equally expert knowledge of the imitators of epilepsy as of the myriad clinical expressions of epileptic seizures in order to better refine and guide the history taking process.

The paramount importance of history taking is confirmed by the observation that misdiagnosis of epilepsy is high throughout the world, irrespective of the wealth and resources available to the health care system. Although the principal error is diagnosing epilepsy in an individual who does not have epileptic seizures, there are certain epileptic seizures, particularly those arising from the frontal lobe at night-time, which may mimic nonepileptic events.\(^3\)

The differential diagnosis of epilepsy encompasses the same spectrum of disorders and normal behaviors in individuals with and without intellectual disabilities (ID). In this context we must acknowledge that frequently any paroxysmal event in an individual with ID is often wrongly assumed to have an epileptic basis until proved otherwise. The physician must recognize that stereotypes and other behavioral events are more frequent and often more atypical in this population. A child or adult with a brain injury will have a higher risk of epileptic seizures than the general population. but they will also have a higher risk of nonepileptic myoclonus, sleep disturbance, and movement disorders. There are rare special situations leading to nonepileptic seizures, such as compulsive Valsalvas in children with autism resulting in anoxic seizures, which are rarely seen in the non-ID population. There are also many situations in which epileptic and nonepileptic disorders may coexist.
The diagnosis of epilepsy must be positive and not based on exclusion of other possibilities. Evidence of an epileptic seizure from a detailed history should ideally be supported by additional investigations. If the physician is honest with the patients and caregivers concerning high levels of misdiagnosis, then it is unlikely they will begrudge the time taken to reach diagnostic certainty. There is often a perception that delaying the diagnosis of epilepsy will be dangerous and that doubt or uncertainty is a sign of a lack of the physician’s skill. However, there is little evidence that reasonable delay in diagnosis is harmful. This compares to the mounting evidence that misdiagnosis leads to inappropriate medical treatments and limitation of educational and employment opportunities. If it is epilepsy, then continued review, an open mind, and the use of videotape recordings with or without electroencephalography (EEG) studies or ambulatory EEG may allow early diagnosis. For the individual with ID who requires respite or other community support, the diagnosis of an additional medical condition may result in withdrawal or limitation of support or an increased financial cost of that support. The impact of epilepsy misdiagnosis can span more than one generation; if a woman is inappropriately treated with medication through her pregnancy, her child is at risk from the teratogenic effects of antiepileptics, and the physician is at risk of a lawsuit.

In this chapter the term seizure is not synonymous with an event with an epileptic basis. Many terms such as fit, seizure, and convulsion are used interchangeably and can sometimes lead to confusion when ascribed to a particular individual. I believe it clarifies matters if we use the term epileptic seizure for a change in behavior caused by a hypersynchronous discharge of cortical neurons and use specific prefixes such as pseudo- or psychogenic or nonepileptic or anoxic when describing seizures that do not have an epileptic basis.

The diversity of nonepileptic events is considerable. Some of these nonepileptic events fully justify the term nonepileptic seizure. The most common type of nonepileptic seizure leading to diagnostic confusion in clinical practice is the anoxic seizure, or syncopal convulsion.

When children suspected of having epilepsy are studied for diagnostic purposes in tertiary monitoring units, nonepileptic events predominate. In the study of Bye and colleagues, which included children with ID, psychological and sleep phenomena were most common, and the EEG frequently showed misleading “epileptiform” discharges. Kotagal and colleagues reported on 134 children and adolescents referred to a pediatric epilepsy monitoring unit at the Cleveland Clinic over a 6-year period. They divided their results into three age groups. In the preschool, two-month to five-year group, the most common diagnoses were stereotypies, sleep jerks, parasomnias, and Sandifer syndrome. In the school-age 5- to 12-year group the most common diagnoses were conversion disorder (psychogenic pseudoepileptic seizures), inattention or day dreaming, stereotypies, sleep jerks, and paroxysmal movement disorders. In the adolescent 12- to 18-year group, over 80% had a diagnosis of conversion disorder (hysteria, psychogenic pseudoepileptic seizures). A significant proportion, 19–46%, of the children studied had concomitant epilepsy.