Epilepsy Case Studies

Pearls for Patient Care

William O. Tatum
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This book is dedicated to patients with epilepsy and their families—thank you for your education about seizures and about life.

William O. Tatum IV

Preface

Sir William Osler said, "To study the phenomenon of disease without books is to sail an uncharted sea, while to study books without patients is to not go to sea at all."* This book is about 40 people whose lives took a different course after they were affected by seizures and epilepsy. The chapters in this book represent case histories drawn from "real-life" experiences in people with seizures. The intent of presenting these patient histories in a case-based format is designed to stimulate the same deductive reasoning that is commonly used when seeing epilepsy patients in the clinic. The use of neuroimaging and neurophysiology in the study of patients with epilepsy has become a staple with which the diagnosis and treatment of epilepsy has become inextricably intertwined. Therefore, the correct interpretation of these studies is essential to reach the correct diagnosis and treatment. Following the clinical scenario composed of a wide variety of epilepsy cases, questions are posed to organize the reader's thoughts in addressing each case. Questions that revolve around each patient include commonly asked questions such as, "How does this test help us with the diagnosis?" and "What is the precise relationship of the patient's seizures to their overall neurological condition?" The most poignant questions include, "How does this information help us to devise a treatment plan?" and "What do we know about the anticipated course and prognosis?" The questions raised in each section incorporate answers to these questions about diagnosis, treatment, and prognosis where the a knowledge base exists. They are addressed in a segment of the book that focuses on a discussion of the facts of the case. Where it is possible, these discussions rely upon the latest medical evidence to support the responses. At the end of each case a few salient citations are included. Our hope to provide an overview of the topic and search for an expanded bibliography, if they so desire.

We learn from every patient. Our "take-home" messages are encapsulated in the form of clinical pearls that shape the basis of our understanding. Furthermore, these pearls of wisdom guide our decision-making in the approach to treatment of future patients with similar case scenarios. There is simply no written text that can replace the knowledge that is derived from hearing and seeing our patient and what they tell us. Our overreliance and overuse of "tests" will never replace the clues that our patients give us when we perform the neurological history and examination.

The field of epileptology encompasses some of the most dynamic and dramatic conditions that a Neurologist will face. Little is more surprising in the field of Medicine than the spontaneity and unpredictability of seizures. Case Studies in Epilepsy will aid in selecting the approach to a clinically based problem list in a style that we hope stimulates reasoning in a style that is fun. From cases that include first onset seizure to drug-resistant epilepsy, from seizures stemming from unknown causes to those produced by a brain tumor, from infancy to the elderly, diagnostic dilemmas and treatment challenges exist and require an individualized approach. Standard and novel diagnostic associations with seizures including genetics and autoimmunity are addressed in addition to nonmedical treatment options including epilepsy surgery, neurostimulation, dietary control, and alternative medicine. These topics are well represented by 40 illustrative case studies contained in this book. An introduction to some of the emerbing treatments such as newer anti-seizure drugs, neurostimulators, and minimally-invasive brain surgeries for epilepsy are included. The cases themselves, serve as the platform to highlight and encompass the broad group of the epilepsies including those with genetic, structural-metabolic, and unknown causes. These cases were obtained from expert epilepsy clinicians at the Mayo Clinic. It is widely known that even in the most productive academic circles, even the most educated in epilepsy centers may be heard to say, “I remember that I once had a case of …”.

*Osler W. Books and men. Boston Med Surg J. 1901;144:61.

Jacksonville, FL, USA William O. Tatum IV, D.O.

Acknowledgment

I am grateful to my colleagues at the Mayo Clinics for both the opportunity to work with them and to learn from them. This multiauthored enterprise-wide work has been compiled by many outstanding epileptologists that have contributed freely and generously of their time. It serves as a testimony to their dedication to the field of epilepsy. Each author has presented a specific vignette that represents a patient who suffered from an individual affliction of epilepsy. It is not the symptom that is remembered, but rather it is the person who imprints the case on our minds. To a large degree, our collective experience in patient care has been shaped by the unique qualities of a single patient whose story has impacted our own lives. These individuals quickly come to mind when we need an example to serve as a prototype for a certain syndrome or situation.

One of my favorite mentors first taught me that it is the needs of the patients that come first in delivering the best neurological care possible. That people are behind the symptoms of their illness and that treatment begins with compassion and advocacy by a human touch. The stigma and painful lack of predictability with seizures that patients and their families endure is something that most of us will hopefully never know. The cases described in this book while presented in a didactic fashion lack the emotional coloring behind each case too shallow to be appreciated. I hope that the readers of this book will never experience a seizure. Instead, I hope that these 40 stories, which are drawn from real-life experiences, can portray the breadth and individuality of epilepsy and therein provide education and compassion that in some way is able to help at least 1 person with seizures.

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Chapter 1
Epileptic Spasms

Elaine Wirrell

Case Presentation

A 7-week-old female presented with a 2-week history of recurrent, brief spells that consist of bilateral arm and leg flexion (left more so than right). She also had head flexion to the left and leftward eye deviation. Each event lasted less than 1 s, but these occurred several times a day in clusters that lasted up to 10 min. Events were particularly prominent shortly after waking. She was diagnosed with a seizure disorder and started on Topiramate by her local pediatric neurologist. The events persisted without a change in event frequency, despite dose increases to 20 mg/kg/day.

She was the product of a healthy term pregnancy to a 31-year-old G1P0 mother. The delivery was a normal spontaneous vaginal delivery with a birth weight of 3,600 g. She was discharged from the hospital at 2 days of age and was well without incident until 5 weeks of age. Her previous family history was unremarkable.

The general examination was unremarkable. Her weight, height, and head circumference were all at the 25th percentile of growth for her age. A thorough examination of her skin was performed, including a normal evaluation with a Wood's lamp. There were no neurocutaneous lesions. She was alert and attentive at her neurological examination. Her cranial nerves were normal. Her motor examination demonstrated that she had mild hypotonia in her left upper extremity and tended to use it less than her right arm. Sensory examination revealed that she had symmetrical withdrawal to noxious stimulation. No pathological cerebellar functions or reflexes were evident. Brain MRI (Fig. 1.1) and EEG (Fig. 1.2) were also subsequently obtained.

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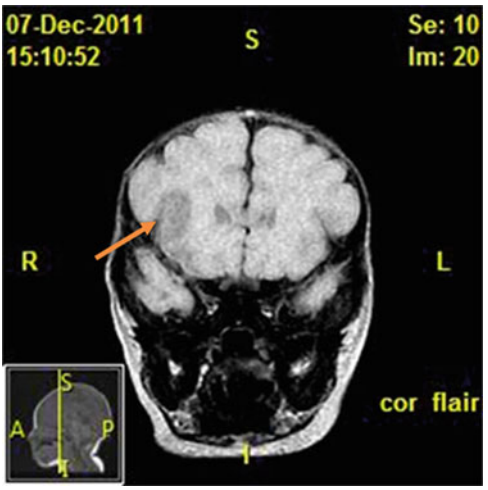

Fig. 1.1 Coronal T1 image of brain MRI at 6 weeks of age. Note the hypointensity in the right frontotemporal region involving insular cortex (*arrow*)

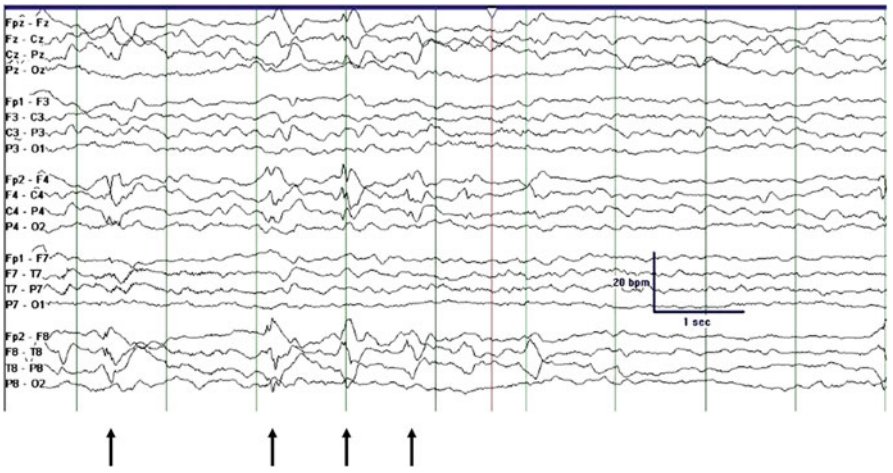

Fig. 1.2 Interictal EEG demonstrating right frontal-temporal epileptiform discharges (*arrows*). Sensitivity 10 μV/mm, filter settings 1–70 Hz, display speed 60 mm/s

Clinical Questions

1. What specific type of spell is she presenting with clinically?
2. What is the most likely etiology for these events?
3. What does her neuroimaging and EEG demonstrate?
4. How do you classify dysplastic cortical malformations?
5. How should she be managed?

Discussion

1. She is presenting with epileptic spasms (ES), which have a focal component. Epileptic spasms are seen most commonly in the first year of life and characteristically occur in clusters, as in this child's case. They are most commonly associated with West syndrome, though they may appear independent of a syndromic association. West syndrome is characterized by the triad of (a) spasms, (b) hypsarrhythmia on the EEG, and (c) intellectual disability, and is most commonly present between 2 and 24 months of age. ES may also be associated with Ohtahara syndrome (early infantile epileptic encephalopathy), which frequently occurs with focal seizures. In Ohtahara syndrome, onset of spasms typically occurs at a younger age than West syndrome, often in the first 2 months of life. Most infants with Ohtahara syndrome will be found to have a structural brain abnormality; however, in approximately 10 % of cases, a genetic etiology (particularly a mutation in *STXBP1*) is responsible. Children with Ohtahara syndrome are encephalopathic and show a burst-suppression pattern on EEG.
2. An underlying etiology can be identified in approximately 80 % of cases; however, the etiologies are diverse. They include structural abnormalities of the brain that include, but are not limited to, prior injury, tuberous sclerosis, and malformations of cortical development (MCD). In addition a genetic predisposition or chromosomal etiology (Trisomy 21, *CDKL5* mutation, *ARX* mutation, etc.) or metabolic disorders (mitochondrial cytopathies, pyridoxine dependency, etc.) may be involved.
3. The brain MRI scan that was done at 6 weeks of age showed a T1 hypointensity in the right anterior insular cortex. This is most likely due to a focal MCD. In early infancy, focal MCD are seen as T1 hypointensities without a corresponding T2 hyperintensity. Due to ongoing myelination, such malformations can be very challenging to visualize between 4 and 24 months. After 2 years, MCD can be detected by the more typical features of cortical thickening, blurring of the gray–white junction, abnormal gyral or sulcal patterns, or T2 hyperintensity.

   Her interictal EEG pattern showed sharp waves rising from the right frontotemporal region. Ictal EEG later confirmed seizure onset that arose from the same area. Her EEG at this time was not consistent with either a hypsarrhythmia (note the absence of high-voltage EEG) or a burst-suppression pattern. Because of the features on EEG, a more definitive diagnosis of West or Ohtahara syndrome could not be made.
4. A clinicopathological classification system has been proposed, which divides these lesions into the following groups:

   FCD type I: Abnormal cortical layering that either compromises the radial migration and maturation of neurons (FCD type Ia), the 6-layered tangential composition of the neocortex (FCD type Ib), or both (type Ic)

   FCD type II: A malformation that presents with disrupted cortical lamination and specific cytological abnormalities. FCD type IIa has dysmorphic neurons