

SEIZURES AND EPILEPSY: PATHOPHYSIOLOGY AND PRINCIPLES OF DIAGNOSIS

Thomas R. Henry, MD

[Scroll down for answers and explanations, which appear after question 5.]

QUESTIONS

1. A full-term male neonate has repeated tonic seizures and the electroencephalogram (EEG) shows a generalized burst-suppression pattern interictally. What is the likely diagnosis?
 - A. Benign familial neonatal epilepsy
 - B. Early myoclonic encephalopathy
 - C. Ohtsuka syndrome
 - D. West syndrome
 - E. Myoclonic epilepsy in infancy
2. A full-term female infant at 10 months has normal development, single or clustered focal or massive jerks, and a normal interictal EEG. What is the likely diagnosis?
 - A. Epilepsy of infancy with migrating focal seizures
 - B. West syndrome
 - C. Myoclonic epilepsy in infancy
 - D. Dravet syndrome
 - E. Progressive myoclonus epilepsy
3. A 5-year-old boy has new onset of generalized tonic-clonic seizures as well as seizures with emesis and unresponsiveness. The interictal EEG shows generalized 3-per-second spike-wave bursts and bilateral occipital spikes. What is the likely diagnosis?
 - A. Lennox-Gastaut syndrome
 - B. Childhood absence epilepsy
 - C. Benign epilepsy with centrotemporal spikes
 - D. Panayiotopoulos syndrome
 - E. Epilepsy with myoclonic absences
4. A 25-year-old graduate student has a first generalized tonic-clonic seizure after staying up all night to study, has a negative urine toxicology screen in the emergency department, and has a normal cranial computed tomography (CT) scan. Later in the neurology clinic, on further questioning she reports that for the last year she has had multifocal jerks on mornings after staying up late. The brain magnetic resonance imaging (MRI) study

shows subtle bilateral subcortical white matter signal changes. The interictal EEG is normal except for a single brief burst of diffuse, bilaterally predominant spike-wave complexes repeating at about 4 Hz. What is the likely diagnosis?

- A. Juvenile absence epilepsy
 - B. Juvenile myoclonic epilepsy
 - C. Progressive myoclonus epilepsy
 - D. Autosomal-dominant nocturnal frontal lobe epilepsy
 - E. Rasmussen syndrome
5. A 25-year-old cafeteria worker has a history of febrile convulsions in early childhood and poor school performance. No seizures have occurred for many years off antiseizure medications, until a generalized tonic-clonic seizure is observed at work. In retrospect, the patient has had intense "daydreams" at work or home for the last year, and others often have had to repeat his name to get him to answer. The interictal EEG shows frequent delta-theta transients over the right temporal lobe and occasional right temporal spike-wave complexes. On brain MRI the right hippocampus is smaller and brighter than the left hippocampus. What is the likely diagnosis?
 - A. Epilepsy with generalized tonic-clonic seizures alone
 - B. Autosomal dominant epilepsy with auditory features
 - C. Lennox-Gastaut syndrome
 - D. Gelastic seizures with hypothalamic hamartoma
 - E. Mesial temporal lobe epilepsy with hippocampal sclerosis

ANSWERS

1. The correct answer is (C), Ohtsuka syndrome. Only the syndromes in answers A, B, and C—benign familial neonatal epilepsy (BFNE), early myoclonic encephalopathy, and Ohtsuka syndrome—occur in neonates. Of these, only BFNE and Ohtsuka syndrome involve tonic seizures (although West syndrome involves tonic seizures as well). Finally, only the Ohtsuka

syndrome features burst-suppression on the EEG.¹⁻³

2. The correct answer is (C), Myoclonic epilepsy in infancy. Only the syndromes in answers C and E—myoclonic epilepsy in infancy and progressive myoclonus epilepsy—involve myoclonus without other seizures, and of the listed syndromes, only myoclonic epilepsy in infancy features normal development. In addition, myoclonic epilepsy in infancy, along with Dravet syndrome, often has normal interictal EEGs in infancy.⁴
3. The correct answer is (D), Panayiotopoulos syndrome. Nonconvulsive seizures with ictal emesis are rare except in the Panayiotopoulos syndrome. Interictal generalized 3-per-second spike-wave bursts and bilateral occipital spikes can occur in benign epilepsy with centrotemporal spikes (BECTS) and Panayiotopoulos syndrome, but the absence of centrotemporal spikes excludes BECTS.⁵
4. The correct answer is (B), Juvenile myoclonic epilepsy. Juvenile myoclonic epilepsy (JME) most often features generalized tonic-clonic seizures and myoclonus on awakening; each of the other listed syndromes requires at least one additional type of seizure that this patient does not have. Generalized fast spike-wave and polyspike-wave discharges are most characteristic of JME, and not of the other syndromes. Onset of myoclonus in JME occasionally occurs in early adulthood, although onset of myoclonus usually is earlier.⁶
5. The correct answer is (E), Mesial temporal lobe epilepsy with hippocampal sclerosis. The brain MRI is described as showing the findings of hippocampal sclerosis, and these findings together with the ipsilateral interictal temporal EEG spikes are compatible only with mesial temporal lobe epilepsy (TLE) with hippocampal sclerosis. Many patients with mesial TLE can provide a more definite description of auras and other ictal and postictal phenomena, but subjective symptoms of the partial seizures in mesial TLE also can be quite vague.⁷

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Table of Contents Epilepsy Board Review Manual

1. Understanding the eBook Epilepsy Board Review Manual
 - The Rise of Digital Reading Epilepsy Board Review Manual
 - Advantages of eBooks Over Traditional Books
2. Identifying Epilepsy Board Review Manual
 - Exploring Different Genres
 - Considering Fiction vs. Non-Fiction
 - Determining Your Reading Goals
3. Choosing the Right eBook Platform
 - Popular eBook Platforms
 - Features to Look for in an Epilepsy Board Review Manual
 - User-Friendly Interface
4. Exploring eBook Recommendations from Epilepsy Board Review Manual
 - Personalized Recommendations
 - Epilepsy Board Review Manual User Reviews and Ratings

- Epilepsy Board Review Manual and Bestseller Lists
- 5. Accessing Epilepsy Board Review Manual Free and Paid eBooks
 - Epilepsy Board Review Manual Public Domain eBooks
 - Epilepsy Board Review Manual eBook Subscription Services
 - Epilepsy Board Review Manual Budget-Friendly Options
- 6. Navigating Epilepsy Board Review Manual eBook Formats
 - ePub, PDF, MOBI, and More
 - Epilepsy Board Review Manual Compatibility with Devices
 - Epilepsy Board Review Manual Enhanced eBook Features
- 7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Epilepsy Board Review Manual
 - Highlighting and Note-Taking Epilepsy Board Review Manual
 - Interactive Elements Epilepsy Board Review Manual
- 8. Staying Engaged with Epilepsy Board Review Manual
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Epilepsy Board Review Manual
- 9. Balancing eBooks and Physical Books Epilepsy Board Review Manual
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Epilepsy Board Review Manual
- 10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
- 11. Cultivating a Reading Routine Epilepsy Board Review Manual
 - Setting Reading Goals Epilepsy Board Review Manual
 - Carving Out Dedicated Reading Time
- 12. Sourcing Reliable Information of Epilepsy Board Review Manual
 - Fact-Checking eBook Content of Epilepsy Board Review Manual
 - Distinguishing Credible Sources

13. Promoting Lifelong Learning
 - Utilizing eBooks for Skill Development
 - Exploring Educational eBooks
14. Embracing eBook Trends
 - Integration of Multimedia Elements
 - Interactive and Gamified eBooks

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