Ataxia in Children: Review Questions

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QUESTIONS

Choose the single best answer for each question.

1. **A 6-year-old boy awakens in his normal state, but by noon becomes so unsteady that he cannot walk. On examination, he looks scared but is well-spoken and cooperative. He has mild nystagmus on lateral gaze bilaterally, past-pointing on finger-nose-finger test, and normal reflexes. Strength is intact, but he cannot balance while standing, and his gait is wildly lurching. He developed chicken pox 10 days ago, but he is otherwise healthy. Developmental milestones are normal. What is this child's most likely diagnosis?**
   (A) Acute postinfectious cerebellitis
   (B) Friedreich ataxia (FRDA)
   (C) Medulloblastoma
   (D) Miller Fisher syndrome
   (E) Multiple sclerosis

2. **A 2-year-old girl has sudden attacks of falling multiple times a day. During these attacks, she "walks like she's drunk," looks scared, and turns pale, but there is no loss of consciousness. Her parents have noticed that sometimes with an attack, her eyes will appear to "wobble." She will grab at the walls, and, when on the floor, grab at the floor. If she falls asleep after onset of an attack, she wakes up normal. Vomiting also helps relieve symptoms transiently. She has these episodes about once every few weeks to few months, in between which she is normal. Physical examination is normal. What is this child's most likely diagnosis?**
   (A) Benign paroxysmal positional vertigo
   (B) Benign paroxysmal vertigo (BPV) of childhood
   (C) Brainstem encephalitis
   (D) Ependymoma
   (E) Frontal lobe epilepsy

3. **An 11-year-old boy still cannot ride a bike and has become increasingly clumsy over the past 4 to 5 years. A distant uncle was also clumsy and died in young adulthood of heart disease. Physical examination shows dysarthric speech, head thrusts with visual tracking, past-pointing on finger-nose-finger test, disorganization on rapid pronation-supination of the hand, jerky and illegible handwriting, and lurching gait. Deep tendon reflexes are not elicitable. What test is most likely to be diagnostic in this child?**
   (A) Magnetic resonance imaging (MRI) of the brain and lumbar puncture
   (B) Electromyogram (EMG)/neurologic deficit score (NCS)
   (C) Lipid panel, apolipoproteins A and B
   (D) Test for trinucleotide repeat of FRDA gene
   (E) Plasma amino acids and urine organic acids

4. **A 4-year-old girl presents for involuntary writhing movements in her arms with small, random, superimposed twitches. She has a history of asthma and frequent pneumonia and sinusitis. Physical examination shows hypometric saccades, forced blinking to initiate saccades, truncal ataxia, and choreoathetosis. Skin examination shows telangiectasias on the bulbar conjunctivae, on the tops of the ears, along the malar prominences of the face, and within the flexor creases of the elbows and knees. What test is most likely to be diagnostic?**
   (A) Alpha-fetoprotein (AFP) and carcinoembryonal antigen (CEA)
   (B) Chest radiograph
   (C) EMG/NCS
   (D) MRI of the brain and lumbar puncture
   (E) Urine for catecholamines

(turn page for answers)

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ANSWERS AND EXPLANATIONS

1. **(A) Acute postinfectious cerebellitis.** When following a viral infection by 10 to 14 days, onset of cerebellar symptoms (maximal within a few hours) is typical of acute postinfectious cerebellitis.[1,2] In this condition, the sensorium should remain clear. MRI of the brain may show increased T2 intensities in the cerebellum. Improvement should begin within a few days of onset, and gait should normalize within 1 to 5 months. The rapidity of symptom onset distinguishes this condition from medulloblastoma and FRDA. This child's preserved reflexes and absence of ophthalmoplegia distinguish this condition from the Miller Fisher variant of Guillain-Barré syndrome. Multiple sclerosis involves recurrent attacks with evidence of central demyelination and would not be diagnosed with a first episode.
2. **(B) BPV of childhood.** This child's recurrent attacks of vertigo relieved by sleep and vomiting most likely represent BPV of childhood.[3] It commonly affects infants and preschoolers and may be a migraine variant, but there is usually no headache. About 1 in 5 children will later develop migraines, and about 2 in 5 have a family history of migraine. The outcome is benign, with resolution of attacks over months to years. The time course of symptoms in this child is not consistent with ependymoma or brainstem encephalitis. Frontal lobe seizures (when not convulsive) are characterized by brief episodes of posturing, spinning, or strange behaviors that occur in clusters, especially at night. Benign paroxysmal positional vertigo occurs in adults and involves the dislodging of otoliths in the vestibular apparatus.
3. **(D) Test for trinucleotide repeat of FRDA gene.** FRDA is the most common of the hereditary ataxias, affecting 1 in 50,000 children.[4] It is an autosomal recessive GAA trinucleotide repeat disorder affecting the FRDA gene on chromosome 9q13 (which codes for frataxin protein). Triplet repeats greater than 100 result in accumulation of iron in mitochondria with gradually progressive dysarthria, ataxia, and posterior column signs. Presentation usually occurs between ages 2 and 16 years. Upper extremities are affected more than lower extremities. Deep tendon reflexes are usually absent on presentation; when reflexes are retained, the course may be milder. Plantar response may be upgoing. Most patients will have an axonal neuropathy, with small or absent sensory nerve action potentials, and relative sparing of motor nerve conduction. Brain MRI may show atrophic cerebellum, brainstem, and posterior columns, and the corticospinal tract may also be affected. A lipid panel with apolipoprotein levels would be helpful when hypobetalipoproteinemia or abetalipoproteinemia is suspected; however, in both of these conditions, there is generally some element of failure to thrive. Screening for metabolic disorders with amino and organic acids is more helpful when there is a history of lethargy, vomiting, or decompensations with minor illnesses. Most spinocerebellar ataxias are also trinucleotide repeat disorders, but most present in adulthood and are autosomal dominant. Management of FRDA includes screening for cardiomyopathy, scoliosis, and diabetes mellitus.
4. **(A) AFP and CEA.** This child has the classic triad of ataxia, telangiectasia, and frequent sinopulmonary infections, which suggests a diagnosis of ataxia-telangiectasia (AT).[5] AT is an autosomal recessive disorder affecting the ATM gene on chromosome 11q22-23, which is a gene involved in cell-cycle progression and DNA repair. Ataxia begins proximally and spreads distally, and choreoathetosis is frequent. Telangiectasias appear between age 2 and 10 years, and mild mental retardation occurs in 1 in 3 persons. There is an increased tendency to develop malignancies, especially lymphomas and leukemias. The immune system is also impaired, accounting for recurrent infections. AFP is often elevated, CEA may be elevated, and IgG (especially subclasses 2 and 4), IgA, and IgE are depressed. Together these tests have up to 90% sensitivity. Specific testing for the ATM gene is also available. Brain MRI may show cerebellar atrophy. Management of AT includes aggressive treatment of infections and avoiding radiation, including chest radiography and sunbathing. In the past, early death was most commonly the result of infection but is now caused by malignancies. Heterozygotes are also at risk for malignancy, especially breast cancer. Urine for catecholamines is a test for neuroblastoma (opsoclonus-myoclonus) and pheochromocytoma.

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