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You are a flight surgeon at a geographically separated medical clinic. Your patient is a 40-yr-old pilot complaining of weakness for the past month during a routine appointment. He has not been into the clinic since his last annual flight physical. The patient states he has been working out and would suddenly lose his balance while walking. He states the symptom presents itself intermittently and has no predictive pattern; he denies the weakness is any different during the morning vs. the evening. He denies any intercurrent illness. He denies any change in diet or intake of supplements other than post-workout protein. He denies generalized fatigue, difficulty swallowing, difficulty breathing, and pain. He is very muscular and his examination is grossly normal with +2/4 reflexes symmetrically and 5/5 motor strength in upper and lower extremities. Upon examination, you notice he is unable to sit still and his extremities portray jerky, fidgety movements.

1. If you suspect myopathy as the pathology in this case, which of the following labs would be of LEAST use to you?

A. Creatine kinase (CK).
B. Lactate dehydrogenase.
C. Aspartate aminotransferase.
D. Erythrocyte sedimentation rate.

ANSWER/DISCUSSION

1. D. Erythrocyte sedimentation rate is indicative of inflammation but is not specific to muscle disease, as are all of the other labs. CK would be leaked into the blood stream when muscle fibers are damaged. Lactate dehydrogenase is also indicative of acute muscle/tissue damage. Aspartate aminotransferase is usually indicative of liver disease but can also be elevated in cases of muscle damage. These levels may be elevated when a person strenuously works out. It is advisable to tell the patient not to vigorously work out prior to the lab being drawn. This is especially true if the person is deconditioned.

These labs as well as a comprehensive metabolic panel and thyroid panel are drawn from the patient, and they are within normal limits. CK is in the high range of normal, but the patient is fairly muscular and had been working out prior to the lab being drawn.

2. What symptoms in his history would relatively exclude this patient from the diagnosis of myasthenia gravis?

A. Lack of fluctuating weakness.
B. Lack of ptosis.
C. Lack of difficulty swallowing.
D. Lack of difficulty breathing.
E. A and B.

ANSWER/DISCUSSION

2. E. Myasthenia gravis is a disease that affects the neuromuscular junction. Symptoms are fluctuating weakness, ptosis, diplopia, difficulty swallowing, and difficulty breathing. The fluctuating weakness would show that the patient is weaker at the end of the day compared to the morning. The patient doesn't portray ptosis nor does he portray fluctuating weakness, which would be more indicative of myasthenia gravis. He does not present with difficulty swallowing nor does he present with difficulty breathing, which may be signs of this disease. Aside from history, this would be diagnosed by finding antibodies against acetylcholine receptor or muscle-specific tyrosine kinase.[4] Although the pathology can show similar symptoms, the lab test for myasthenia gravis was negative.

On subsequent visits, you notice the spastic, jerky involuntary movement in his extremities has noticeably increased. You also notice on examination today that the patient's upper extremity has become hypotonic and then hyper-reflexive. His hands and fingers are also twisting in an unusual fashion. You order a magnetic resonance imaging (MRI) of the brain.

3. What do you expect to find?

A. Normal findings – unremarkable.
B. Basal ganglia abnormalities.
C. White matter lesions in the brain.
D. Overall decrease in brain size and mass.

ANSWER/DISCUSSION

3. B. MRI of the brain in a patient with movement disorder will show abnormalities in the basal ganglia. However, these findings are not very definitive. An unremarkable brain scan can be seen in many neurological disorders, notably those that do not involve the central nervous system. White matter lesions are often a nonspecific finding on brain MRI scans, but can be seen with conditions such as trauma,

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migraines, infection, stroke, hypoxia, and demyelinating diseases. Overall decrease in brain mass would be suggestive of degenerative conditions such as Alzheimer's disease.

The patient's brain MRI showed atrophy in the caudate and putamen, which is suggestive of Huntington's disease (HD.) Shortly thereafter, when he did not show up for work that morning, his wingmen found him having a psychotic break.

4. What confirmatory test would diagnose his underlying condition?

A. Serum ceruloplasmin.
B. Anti-Smith antibody.
C. Cytoplasmic antineutrophil cytoplasmic antibody.
D. Huntington gene (HTT) test with cytosine-adenine-guanine (CAG) mutations.

ANSWER/DISCUSSION

4. D. HD is a hereditary disease that is caused by a mutation in HTT. HD causes the breakdown of nerve cells in the basal ganglia, specifically the caudate and the putamen. The symptoms include chorea, dystonia, personality changes, and impaired judgment.[7] The hallmark trait of the disease is chorea, which are jerky, nonrhythmic, involuntary movements. HD patients also portray athetosis, which are slow, purposeless movements that flow. These are typically seen in the hands and fingers. Unfortunately, the prognosis in HD is poor, with most patients dying within 10 yr of onset. Due to the lack of any effective treatment and the prognosis of this terminal disease, HD patients are at increased risk for suicide.[3,8]

HD is inherited in an autosomal dominant pattern. HTT parents pass on the gene, and each successive child has a 50% chance of getting the gene. The gene is associated with an increased expansion of the CAG repeats. People with regular HTT gene alleles will present with less than 26 CAG units. People who are symptomatic will present with over 36 CAG repeats.[2,6] People who had increased repeats would be symptomatic at an earlier age. This condition is subject to genetic anticipation. Genetic anticipation is when the disease presents itself earlier in successive generations. Affected children can show signs and symptoms at an earlier age than their affected parent. This is seen with trinucleotide repeat disorder diseases such as HD and fragile X syndrome.

Anti-Smith antibody tests for systemic lupus erythematosus, serum ceruloplasmin tests for Wilson's disease, and cytoplasmic antineutrophil cytoplasmic antibody tests for vasculitis.[3]

5. What would have made this easier to diagnose and would have given definitive diagnosis?

A. History of exposure to person with human immunodeficiency virus.
B. History of exposure to Fukushima reactor during tsunami.
C. History of intake of workout supplements.
D. Admission of family history of HD.

ANSWER/DISCUSSION

5. D. As stated above, HD is a genetic disease, with each successive generation having a 50% chance of getting the disease. The pilot was thoroughly questioned and admitted he intentionally omitted a family history of HD to keep his job. One of the main teaching points in this case is to get a thorough family history, which would have precluded most of the tests.

Exposure to somebody with human immunodeficiency virus would not cause HD. While exposure to the Fukushima reactor would increase radiation exposure, it would not change genetic material to increase CAG repeats. There are no workout supplements that would cause CAG repeats.

6. What should you do as a flight surgeon?

A. Apply to the major command for an indefinite waiver to keep him flying.
B. Wait 1–5 yr to see if he is stable, then apply.
C. See if the Federal Aviation Administration (FAA) would approve a waiver for this individual as a civilian.
D. Send him to a Medical Evaluation Board (MEB) for permanent disqualification from flying duties.

ANSWER/DISCUSSION

6. D. The member has to have an MEB, since this condition is considered unfitting for military duty according to the Medical Standards Directory.*

Waiting for 1–5 yr is incorrect, as the patient is already symptomatic and the condition will continue to progress. The major command would not grant an aeromedical waiver for this condition for the reason cited in the Medical Standards Directory. The U.S. Navy Aeromedical Reference and Waiver Guide and the U.S. Army Flight Surgeon's Aeromedical Checklists do not have this condition listed as an allowable condition.[5,8] Although the FAA is not as strict as the military in waivers, this condition requires an FAA decision.[1] This would be the incorrect choice since the FAA does not grant waivers for military aviation operators.

The member underwent an MEB and was medically discharged from military service due to Huntington's disease. He was also permanently disqualified from flying duty.

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* U.S. Air Force. Section L: neurologic USAF medical standards, L9 & L10. In: Medical standards directory. 2016:41. [Accessed 1 Nov. 2016]. Available from https://kx2.afms.mil/kj/kx4/FlightMedicine/Documents/Medical%20Standards%20Directory%20(MSD)/Correction%20MSD%2029%20November%202016.pdf to those with access.

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