

# Headaches, Mood Swings, and Dropping Things: Differential Diagnosis of a Nervous System Disorder

by

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## Part I – Signs and Symptoms (June 15, 11 am)

“Dr. Gorga, I think you’re going to want to take this case,” said Dr. Boutross, chief of medicine at City Central Hospital.

“What makes you say that?” questioned Dr. Gorga, head of neurology at City Central.

Dr. Boutross proceeded to brief Dr. Gorga about an interesting patient, Carmen Moreno. Dr. Gorga questioned Dr. Boutross throughout his presentation.

“Mrs. Carmen Moreno, age 62, has been complaining about continuous headaches for about one month. Along with her headaches, her husband has noted changes in her personality.”

“What kind of changes?”

“Her husband says she becomes very easily annoyed. He also admitted that she appears to be happy one moment, but changes to an extreme solemn state shortly after.”

“Other than psychological changes, has he noted any physical changes?”

“Locomotion and moving her upper extremities efficiently has become extremely difficult.”

“What is her family history regarding neurological illnesses?”

“Unknown. She was adopted at the age of two from Italy.”

“I’ll gather my team together,” stated Dr. Gorga.

## Questions

1. From the information above, what do you think could be causing Mrs. Moreno’s headaches, mood swings and her motor deficits?
2. What tests do you think the doctors should perform to gather more information about the patient?

## Part II – Pre-Diagnoses (June 15, 1 pm)

Dr. Gorga relayed the patient information he had received to his team of highly intelligent and qualified neurologists: Dr. Perez, Dr. Morelli and Dr. Stevenson.

*Dr. Gorga:* So, talk to me; what could this be?

*Dr. Perez:* Based on these preliminary symptoms, Parkinson’s disease. We should run more tests. If they indicate Parkinson’s, then get her started on L-DOPA injections immediately.

*Dr. Morelli:* Difficulty with walking and performing motor functions does not necessarily mean she has Parkinson’s. It could very well be Huntington’s disease.

*Dr. Perez:* Huntington’s? That’s the exact opposite of Parkinson’s.

*Dr. Stevenson:* Well, according to your collective pre-diagnoses, Carmen either has too much or too little dopamine. Let’s wait until the test results to make a pre-diagnosis.

*Dr. Gorga:* Before we run tests, don’t you all want to know a little bit more about our female patient? Carmen is a 62-year-old woman who lives in Brooklyn, New York, with her husband. She is the mother of four children. Her symptoms started about a month ago, immediately after she went on vacation to Lake Okeechobee, Florida, with her husband.

*Dr. Stevenson:* There it is! The curve ball. A warm, freshwater lake.

*Dr. Gorga:* Don’t jump to conclusions just yet, Dr. Stevenson. Please inquire more about this trip with our patient, especially if she went swimming in the lake.

### Questions

- Based on the general description of symptoms, what specific symptoms of Huntington’s disease/chorea does Dr. Morelli suspect in Mrs. Moreno? (Diagnostic Table: disease/condition and history/signs/symptoms)
- Based on the general description of symptoms, what specific symptoms of Parkinson’s disease does Dr. Perez suspect in Mrs. Moreno? (Diagnostic Table: disease/condition and history/signs/symptoms)
- How can the symptoms indicate that it could be both Parkinson’s or Huntington’s disease?

6. What types of tests are needed to properly diagnose Parkinson's disease or Huntington's disease? (Diagnostic Table: testing)
7. Why could swimming in the lakes (exposure to lake water) provide insight on the underlying disease? (Diagnostic Table: disease/condition)

### *Recommended Resources*

#### *Parkinson's Disease*

- Heyn, S., and C.P. Davis. *n.d.* Parkinson's disease symptoms, causes, stages, treatment, and life expectancy. *MedicineNet*. <[http://www.medicinenet.com/parkinsons\\_disease/article.htm](http://www.medicinenet.com/parkinsons_disease/article.htm)>.
- Mayo Clinic. *n.d.* Parkinson's disease. <<https://www.mayoclinic.org/diseases-conditions/parkinsons-disease/symptoms-causes/syc-20376055>>.
- WebMD. *n.d.* What is Parkinson's disease? <<http://www.webmd.com/parkinsons-disease/tc/parkinsons-disease-topic-overview#1>>.

#### *Huntington's Disease*

- Alzheimer's Association. *n.d.* Huntington's disease. <<http://www.alz.org/dementia/huntingtons-disease-symptoms.asp>>.
- Mayo Clinic. 2018. Huntington's disease. <<http://www.mayoclinic.org/diseases-conditions/huntingtons-disease/symptoms-causes/dxc-20321436>>.
- Huntington's Disease Society of America. *n.d.* What is Huntington's disease? <<http://hdsa.org/what-is-hd>>.

\*\*\* Internet resources accessible as of February 14, 2019 \*\*\*

## Part III – Evaluation, Observations and Testing (June 17, 9am)

*Dr. Stevenson:* Rule out primary amebic meningoencephalitis..

*Dr. Morelli:* Why is that?

*Dr. Stevenson:* She never stepped a foot into any lake during her vacation. In fact, she hates swimming.

*Dr. Gorga:* Her tests revealed slightly abnormal levels of dopamine. We need more ideas.

*Dr. Perez:* Hydrocephalus?

*Dr. Gorga:* There is no history of head injuries, stroke or other cerebral vascular disorders. What else can cause cognitive and motor problems without dopamine being the culprit?

*Dr. Morelli:* A demyelinating disease?

*Dr. Perez:* Could be multiple sclerosis or Guillain-Barre syndrome?

*Dr. Gorga:* Very possible. Let's run the appropriate tests. Has the patient been complaining about anything else since her admittance?

*Dr. Morelli:* She's exhibiting problems with her vision.

*Dr. Stevenson:* Double vision? Double vision, the unbalanced gait and jerky movements observed during her evaluation could indicate something rare like Creutzfeldt-Jakob disease.

*Dr. Perez:* Unsteady gait? Jerky movements? Visual deficits? Sounds like Parkinson's to me. Let's re-run the dopamine test. In the meantime, I'm starting her on L-DOPA for Parkinson's. As for Huntington's, since there's no family history to confirm, I'm running blood tests and sending her in for a CT scan.

### Questions

8. Which symptoms or observations are consistent with multiple sclerosis or Guillain-Barre syndrome? Which are not? (Diagnostic Table: disease/condition and history/signs/symptoms)
  
9. List the symptoms that are associated with Creutzfeldt-Jakob disease that Carmen is presenting. (Diagnostic Table: disease/condition and history/signs/symptoms)
  
10. What abnormalities are the doctors looking for in the blood tests for a Huntington's diagnosis? What abnormalities are they looking for in the CT scan? How do these abnormalities manifest physically in the affected patient? (Diagnostic Table: testing and results of testing)
  
11. What is your diagnosis at this stage of the case study? Why?

## Recommended Resources

### Primary Amebic Meningoencephalitis

Grace, E., S. Asbill, and K. Virga. 2015. *Naegleria fowleri*: pathogenesis, diagnosis, and treatment options. *Antimicrobial Agents and Chemotherapy* 59(11): 6677–81. <<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4604384/>>.

Centers for Disease Control and Prevention. *n.d.* Amebic encephalitis. <<https://www.cdc.gov/parasites/naegleria/general.html>>.

### Hydrocephalus

Delgado, A. and V. Higuera. *n.d.* Hydrocephalus (water on the brain). *Healthline*. <<https://www.healthline.com/health/hydrocephalus>>.

National Hydrocephalus Foundation. 2018. Facts about hydrocephalus. <<http://nhfonline.org/facts-about-hydrocephalus.htm>>.

### Multiple Sclerosis

Dangond, F. 2002. Multiple sclerosis: a prototypical human demyelinating disease. In: Dangond, F., ed., *Disorders of Myelin in the Central and Peripheral Nervous Systems*. Boston: Butterworth-Heinemann, 2002, pp. 103–38.

Mayo Clinic. *n.d.* Multiple sclerosis: symptoms and causes. <<http://www.mayoclinic.org/diseases-conditions/multiple-sclerosis/symptoms-causes/dxc-20131884>>.

### Guillain-Barre Syndrome

U.S. National Library of Medicine. *n.d.* Guillain-Barré syndrome. *Genetics Home Reference*. <<https://ghr.nlm.nih.gov/condition/guillain-barre-syndrome>>.

Mayo Clinic. *n.d.* Guillain-Barre syndrome symptoms. <<http://www.mayoclinic.org/diseases-conditions/guillain-barre-syndrome/basics/symptoms/con-20025832>>.

### Creutzfeldt-Jakob Disease

Centers for Disease Control and Prevention (CDC). *n.d.* Prion diseases. <<https://www.cdc.gov/prions/index.html>>.

MRC Prion Unit at UCL. *n.d.* Prions and prion disease. University College London. <<http://www.prion.ucl.ac.uk/clinic-services/information/prion-disease>>.

\*\*\* Internet resources accessible as of February 14, 2019 \*\*\*

## Part IV – Testing Results and Observations (June 24, 12pm)

*Dr. Morelli:* The patient reports that she has blurry vision, but there is no double vision. CJD is ruled out.

*Dr. Perez:* And we can also rule out Huntington's; her blood test results showed her CAG repeats are way below 40 for the HTT complement gene on chromosome 4. Also, her CT was clean. No shrinkage around the caudate or putamen. Ventricles also don't appear to be enlarged in size.

*Dr. Gorga:* How is her body reacting to the L-DOPA injections?

*Dr. Perez:* There's no change in her condition as of this time.

*Dr. Gorga:* Dr. Perez, perform one last thorough physical exam. We need more ideas. This disorder is winning and we're losing.

*Dr. Stevenson:* I think we should reconsider Dr. Perez's earlier idea: given her motor dysfunction and her gender, we need to check for MS.

*Dr. Gorga:* Then get to it.

*[Three hours later, the team is still in Dr. Gorga's office.]*

*Dr. Gorga:* What about measles?

*Dr. Morelli:* You're, right, what about measles?.

*Dr. Gorga:* Dr. Morelli, ask the patient if she was diagnosed with measles recently.

That night, Dr. Stevenson performed two tests on Mrs. Moreno for the presence of MS: a lumbar puncture and a MRI on her brain.

### Questions

12. Why do you think Dr. Gorga suggested the idea that Mrs. Moreno might have measles? (Diagnostic Table: disease/condition and history/signs/symptoms)
  
13. With respect to the testing for MS, what results would indicate the MS diagnosis? (Diagnostic Table: testing and results of testing)

### Recommended Resources

#### *Subacute Sclerosing Panencephalitis (Measles Virus in the Brain)*

Jayamaha, J. 2013. Measles and SSPE: occurrence and pathogenesis. *Microbiology Australia* 34(3). <<http://microbiology.publish.csiro.au/paper/MA13044.htm>>.

U.S. National Library of Medicine. *n.d.* Subacute sclerosing panencephalitis. *MedlinePlus*. <<https://medlineplus.gov/ency/article/001419.htm>>.

\*\*\* Internet resources accessible as of February 14, 2019 \*\*\*

## Part V – Narrowing the Differential Diagnosis (June 25, 9am)

The lumbar puncture on Mrs. Moreno revealed that her CSF was normal and the results of her MRI showed no water retention in her brain; demyelination had not occurred in Mrs. Moreno's brain.

*Dr. Morelli:* Just an update on our patient: she's showing extreme muscle rigidity, her muscles are tense and in a contracted state at all times.

*Dr. Stevenson:* It's got to be Parkinson's!

*[At that moment Dr. Perez walks into Dr. Gorga's office.]*

*Dr. Perez:* Sorry, it's not Parkinson's. Not only was there no improvement when I put her on L-DOPA, her locomotion deficits have never included a shuffling gait, she didn't present with a hand tremor at rest and her facial expressions seem to be intact. Those are all the hallmark signs of Parkinson's and she still has not presented with those deficits.

*Dr. Gorga:* Well then, it looks like Mrs. Moreno either has Guillain-Barre or measles in the brain," concluded Dr. Gorga.

*Dr. Perez:* Measles in the brain? That would mean she would have to have had a measles infection in the last 15 years, as an adult, right?

*Dr. Gorga:* I asked her about recent illnesses during the initial patient history. She couldn't recall any particular illness but I get the distinct impression that this family does not seek medical attention until something is catastrophic. Her vaccination records are incomplete because she was adopted.

*Dr. Morelli:* Dr. Perez, how's her breathing since you conducted the latest examination on Mrs. Moreno?

*Dr. Perez:* She presented with interesting results. Her initial blood pressure reading was elevated and respirations were a tad higher than usual, about 28 per minute.

*Dr. Gorga:* Well, twenty breaths per minute is normal.

*Dr. Perez:* Right. I then took a second blood pressure reading before I left her room just to confirm my first reading and it was extremely low.

*Dr. Gorga:* Fluctuating blood pressure and difficulty breathing... we're finally on the right track. Hopefully, it's not too late for Mrs. Moreno. These symptoms correspond with late stages for Guillain-Barre or subacute sclerosing panencephalitis.

*Dr. Morelli:* I'll test for SSPE.

*Dr. Stevenson:* Dr. Perez and I will test for Guillain-Barre.

Dr. Perez and Dr. Stevenson performed nerve conduction studies and an electromyography test while Dr. Morelli performed a serum antibody titer test.

### Questions

14. What are all of the symptoms associated with subacute sclerosing panencephalitis? Guillain-Barre? (Diagnostic Table: disease/condition and history/signs/symptoms)
15. How rare is a case of subacute sclerosing panencephalitis? Guillain-Barre?
16. What is your diagnosis at this stage of the case study: SSPE or Guillain-Barre? Why? (Diagnostic Table: elimination of disease/condition)

## Part VI – Final Diagnosis (June 25, 12pm)

The three doctors were waiting for Dr. Gorga in his office. When he entered, Dr. Stevenson began the discussion of Mrs. Carmen Moreno's results.

*Dr. Stevenson:* Looks like we found our 1-in-10 cases a year.

*Dr. Morelli:* Serologic testing confirms the presence of measles antibodies.

*Dr. Perez:* And nerve conduction and electromyography tests were negative; she definitely doesn't have Guillain-Barre syndrome.

*Dr. Gorga:* Dr. Morelli, tell the patient and her family what she's suffering from. Let's start treatment immediately.

