Sometimes Even a Well-Diagnosed Functional Movement Disorder is Not Functional

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CASE DESCRIPTION

CHIEF COMPLAINT

- "Spasms of left upper extremity"

HPI

- 65-year-old woman with an unremarkable neurological history came to Mayo Clinic for a second opinion on intermittent left arm jerking
- Intensive outside brain imaging and laboratory workup was inconclusive
- Acute exacerbation of symptoms resulted in family taking patient to our ED where a CT head was obtained and unremarkable
- Neurology consulted and on physical exam noted vertical nystagmus and slightly suppressible intermittent myoclonus
- Admitted to work up organic etiology underlying a perceived functional overlay
- Interpretation of outside imaging did not identify any obvious or concerning intracranial findings. CBC, electrolytes and TSH were within normal limits
- Working diagnosis: functional movement disorder
- Patient evaluated by PM&R, PT and OT and transferred to inpatient rehabilitation

REHABILITATION COURSE

- Patient made initial functional gains her first week on inpatient rehabilitation with a program aimed at muscle control and relaxation
- Developed progressive altered mental status and was unable to participate in therapies
- Psychiatry consult and infectious work up for delirium performed that revealed positive UA
- Antibiotics started for UTI without improvement in mental status
- Neurology re-consulted and patient ultimately transferred to their service for additional work-up

DISPOSITION

- EEG showed periodic sharp wave complexes
- Repeat brain MRI revealed new restricted diffusion at the basal ganglia- findings concerning for Creutzfeldt-Jakob disease (CJD)
- She dismissed to a facility closer to family and expired the following day
- Postmortem brain biopsy confirmed CJD diagnosis

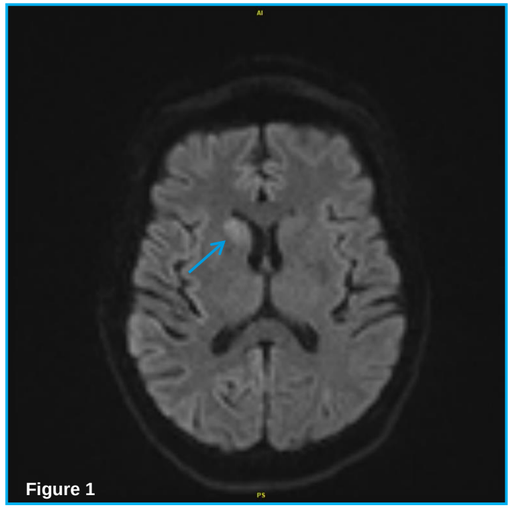
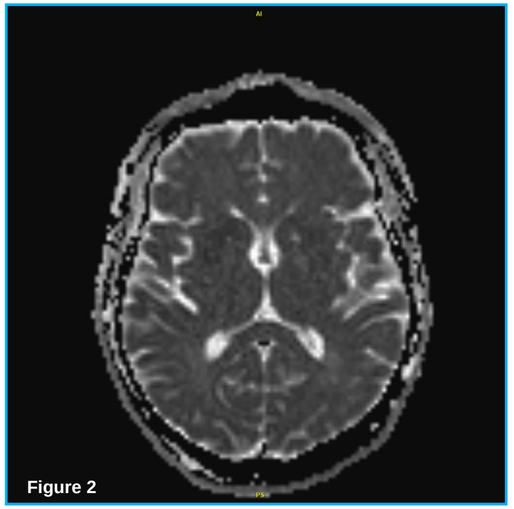


Figure 1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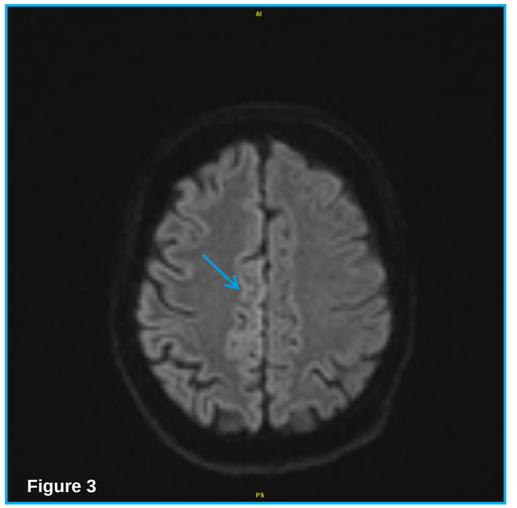
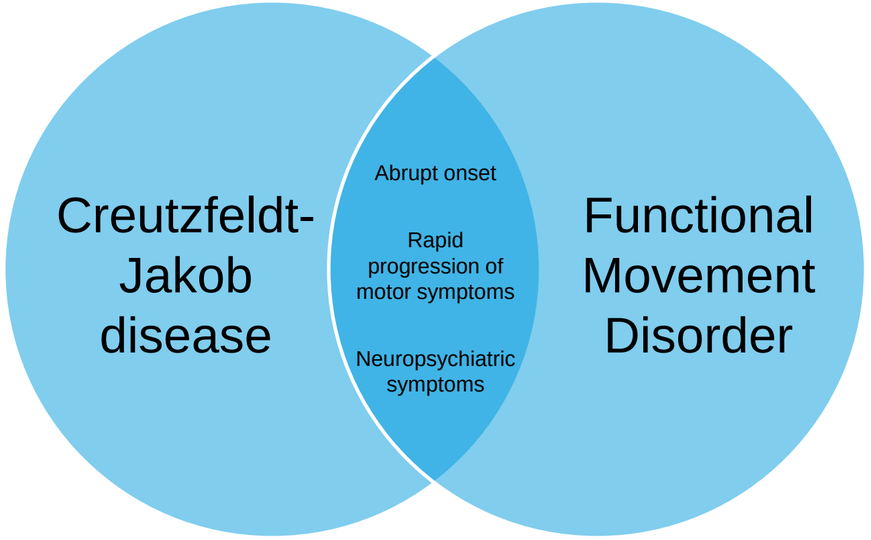


Figure 3

Figure 1,2, and 3: MRI Brain with and without IV contrast performed approximately 5 weeks after and compared to initial outside imaging which was read as negative. Interpretation of repeat imaging: there has been development of restricted diffusion at bilateral basal ganglia (Figure 1 DWI and Figure 2 corresponding ADC) and parasagittal frontal cortices (Figure 3), asymmetric and prominent on the right. Subtle restricted diffusion at the bilateral thalami cannot be excluded. These findings are concerning for Creutzfeldt-Jakob disease.

TABLE 1

COMMON CLINICAL FEATURES OF CJD	
Neuropsychiatric	Dementia, behavioral abnormalities, aphasia, apraxia and frontal lobe syndrome
Myoclonus	Provoked by startle (present in more than 90% of patient)
Cerebellar	Nystagmus, ataxia
Corticospinal Tract	Hyperreflexia, positive Babinski sign and spasticity
Extrapyramidal Signs	Hypokinesia, bradykinesia, dystonia and rigidity



DISCUSSION

- Creutzfeldt-Jakob disease is the most common prion disease, with the majority being the sporadic variant[1]
- Neuropsychiatric symptoms are followed by rapid decline and death[1]
- Similarly, functional movement disorders (FMDs) have an abrupt onset with rapid progression of motor symptoms in patients who often have underlying psychiatric disorders[3, 4]
- Unlike FMDs which have no organic abnormalities[3,4], brain MRI is critical to diagnosing CJD as reduced diffusivity on DWI images is the most sensitive finding[2]
- Periodic sharp wave complexes on EEG are also useful in the diagnosis[2]
- Although dementia and MRI abnormalities typically appear before motor symptoms[2], this patient had a reverse presentation
- While nothing could be done to halt her disease progression, all her treating team felt an earlier diagnosis would have permitted more effective end of life measures

CONCLUSIONS

- This case highlights that a healthy skepticism of even what appears to be a definite diagnosis is always warranted
- Repeat evaluation and testing ultimately allowed for an accurate diagnosis when this patient had persistent worsening motor symptoms, changes in behavior and a decline in functional status despite inpatient rehabilitation

REFERENCES

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