

Is it Creutzfeldt-Jakob Disease?

Maybe it's not CJD

Because CJD affects many different areas in the brain, it can lead to a wide-variety of symptoms, some of which can mimic other neurological diseases. Your doctor's first mission is to rule out treatable illnesses that can present like CJD with rapidly progressive symptoms. In particular, the same symptoms and rapid progression can occur in association with brain inflammation, intermittent seizures, metabolic disorders (like vitamin deficiencies), vascular disease of the brain, Alzheimer's disease and dementias associated with Parkinson's disease.

Typically, your doctor will perform a standard neurological examination and request other diagnostic tests such as

- lumbar puncture to rule out more common causes of dementia and look for signs of an infection,
- electroencephalogram (EEG) to record the brain's electrical activity, which can be distinctive in CJD, and
- brain imaging either by computerized tomography (CT) to rule out stroke and tumor or magnetic resonance imaging (MRI) to exclude other forms of dementia or reveal characteristic patterns of CJD.

The information from these tests will help your doctor determine if you have something treatable or if you need further testing for CJD.

Confirming a CJD diagnosis

The major difficulty in diagnosing CJD is that there is no one typical pathway that the disease follows. CJD has a multitude of possible symptoms and what one patient experiences right away,

another patient may never experience. Diagnosis of CJD often happens through a process of elimination of other diseases. Currently there is no single diagnostic test for CJD but patterns seen on MRI are highly typical and can usually make or refute the diagnosis.

The main indications which can lead to a possible diagnosis of CJD are rapid dementia, unsteady gait and sudden jerky movements. Removing a tiny amount of brain tissue for biopsy and cerebral spinal fluid testing are often used to confirm a possible diagnosis, but both tests carry risk and can be inconclusive.

A definitive diagnosis of CJD can only be confirmed through autopsy. When seen under a microscope, the brain tissue from people with prion disease looks like a sponge due to many tiny holes. For this reason, prion diseases are also known as "spongiform encephalopathies".

UCSF scientists are working to improve MRI tests for CJD.