ROCHESTER REGIONAL HEALTH

Towards Earlier Diagnosis of Transmissible Spongiform Encephalopathies (TSEs): A Case Series, Including One Associated with Squirrel Brain Consumption

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Background

TSEs present diagnostic and infection control (IC) challenges.

Creutzfeldt-Jakob Disease (CJD) is the most common human TSE, occurring in 1-2 per million per year in the US.

Other zoonotic factors or transmissions remain incompletely understood.

TSEs have a long incubation period, but once manifested, prognosis is poor, with death usually within 1 year of symptom onset.

When 4 suspected cases occurred from 11/2017-04/2018, we sought to illustrate its variable presentation and the need for more rapid identification to enhance disease-specific disinfection, sterilization, and quarantine measures.

Methods

Retrospective review was conducted between 2013 to 2018 to identify patients who underwent investigation for probable CJD.

We defined a case as any patient with a rapidly progressive dementing or neurologic illness and lab tests for CJD.

Results

5 patients met case definition. The average time to confirming a diagnosis was 15.4 days respectively.

Table 1. Summary of Patient Characteristics and Laboratory Findings in Suspected CJD Cases

Case	1	2	3	4	5
Age/Sex	61 M	65 F	51 F	61 F	80 M
Cognitive symptoms	Psychosis, schizophrenia, cognitive decline	Dysphasia, depression, psychosis	Vertigo, progressive encephalopathy	Memory loss, aphasia	Aphasia, dysarthria, dysphagia
Motor symptoms	Impaired gait	Impaired gait	Bilateral ataxia	Impaired gait incontinence, abnormal muscle tone with paratonia	Unilateral weakness, jerking movements
EEG	Triphasic pattern	Abundant generalized discharges	Occasional bi-frontal sharp wave discharges	Generalized encephalopathy	NSC
MRI	Increased T2 signal in the pulvinar of the thalamus and cortex (especially frontal lobes)	NSC	NSC	NSC / small vessel infarcts	NSC / small vessel infarcts
RT-QuIC	+	+	+	-	-
14-3-3	+	+	+	-	-
T-tau	8750	>4000	>4000	390	194
Epidemiology	Intake of squirrel brains 	Concurrent apheresis and GYN surgery	Hotel Housekeeping	Industrial Chemist Residence in UK, intake of dog food	Janitor
CJD	V	S	S	No	No
Days to suspecting diagnosis	1	13	2	4	6
Days to confirmation	16	12	18	12	19
Months of Illness	5	3	>12	>12	>12
Outcome	Deceased	Deceased	Alive	Alive	Alive

NSC=Nonspecific changes; S= Sporadic; V=Variant; RT-QuIC = Realtime Quaking Induced Conversion

Results

3 of 5 patients were diagnosed with CJD.

All patients underwent the current proposed testing for CJD, including EEG, MRI, and protein marker investigation.

Variant CJD was diagnosed based on MRI features of increased T2 signal in the pulvinar of the thalamus, which is absent in classic CJD and present in >75% of variant CJD.

The deceased patients' clinical course was consistent w/ CJD.

Discussion

CJD diagnosis is often delayed.

Variant CJD should be considered in diagnostic & infection control measures.

Policies should be modified accordingly.

Improved empiric classification algorithms, and tests with faster turnaround times are needed.