Tonsillar Biopsy as Diagnostic Choice in Variant Creutzfeldt Jakob Disease

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Introduction
There are four human forms of prion diseases, which compose the four forms of Creutzfeldt-Jakob disease: sporadic or classical (sCJD), variant (vCJD), iatrogenic and genetic (or familial). Variant CJD was first described in 1996 and differs from sCJD by having different presenting clinical features with neuropsychiatric symptoms and a slower progression to death over 6–40 months. It also tends to affect younger adults, with the median age at presentation of 26 years (1). There have been 224 reported cases as of Dec 2012: 173 in UK, and 27 in France (2).

There have been 224 reported cases of vCJD, with the median age at presentation of 26 years. There have been 224 reported cases of vCJD, the most number of reported cases of this disease in the UK. This has not been shown to be present in lymphoreticular tissue, specifically tonsillar tissue. Evaluation of tonsillar tissue for PrP\textsubscript{Sc} can be a first-line choice for diagnostic biopsy when investigating vCJD. Disposable instruments should be used when considering biopsy of tonsillar tissue for vCJD.

Case Report
A twenty-three year old male presented to the Emergency Room with a weeklong history of worsening headache. He was found to have altered mental status upon further evaluation. MRI imaging of the brain revealed signs consistent with encephalitis while electroencephalogram ruled out seizure activity. A spinal fluid specimen was negative for meningitis. The Infectious Disease team was consulted and recommended tonsillar biopsy by Otolaryngology to rule out a variant Creutzfeldt-Jakob disease. The patient was taken to the Operating Room after consent was obtained. With the use of a limited number of instruments including a McVor mouth gag, Herd elevator, Bovie electrocautery (Covidien \textsuperscript{R}), and a suction bovie electrocautery, the right tonsil was removed in a subcapsular plane.

The tonsil was sent for permanent pathological confirmation as a fresh specimen. All instruments used during the operative case were disposed of.

The patient we investigated revealed 1+ tonsillar tissue on the right during examination. Pathological specimen did not reveal vCJD.

Discussion
We are providing the first reported case in the US of an attempt to diagnose vCJD based on tonsillar biopsy. The conversion of the protease-sensitive cellular form of the prion protein, PrP\textsubscript{C}, into a pathological isoform, PrP\textsubscript{Sc}, appears to be a key process in pathogenesis (3). Presence of PrP\textsubscript{Sc} in the tonsil is specific for vCJD in comparison to other forms of prion disease. A distinctive PrP\textsubscript{Sc} type, designated type 4t, is consistently seen in tonsil from patients with vCJD.

As Wadsworth et al. have shown, concentrations of PrP\textsubscript{Sc} are not uniform throughout the lymphoreticular system, but are consistently higher in tonsil (4). Tonsil remains the tissue of choice for diagnostic biopsy in the investigation of possible vCJD.

There has been a push for the UK Department of Health to recommend use of disposable instrument kits for tonsillolithectomy as a first step in risk reduction strategies to limit any secondary transmission of vCJD given the most number of reported cases of this disease in the UK. This has not translated into the U.S. given the low case numbers present.

Conclusions
vCJD is a rare but devastating disease with death occurring of 6-40 months after contraction of disease. vCJD has been shown to be present in lymphoreticular tissue, specifically tonsillar tissue. Evaluation of tonsillar tissue for PrP\textsubscript{Sc} can be a first-line choice for diagnostic biopsy when investigating vCJD. Disposable instruments should be used when considering biopsy of tonsillar tissue for vCJD.

References