



Essential tremor

..... a syndrome
or a disease?

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'Essential Tremor' or 'the Essential Tremors': Is This One Disease or a Family of Diseases?

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There is accumulating evidence that the entity referred to as 'essential tremor' (ET) is not a single disease. It may be a family of diseases better referred to as 'the ETs'. This review will



absence of other neurological signs.³⁹ Clinicians ultimately recognized a common isolated tremor syndrome of bilateral upper limb postural or kinetic tremor, with or without head tremor or tremor in other locations, and called this ET. However, the diag-

Consensus Statement on the Classification of Tremors. From the Task Force on Tremor of the International Parkinson and Movement Disorder Society

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“ Essential tremor

- 1) *isolated tremor syndrome of bilateral upper limb action tremor*
- 2) *at least 3 years' duration*
- 3) *with or without tremor in other locations (e.g., head, voice, or lower limbs)*
- 4) *absence of other neurological signs, such as dystonia, ataxia, or parkinsonism.*

”



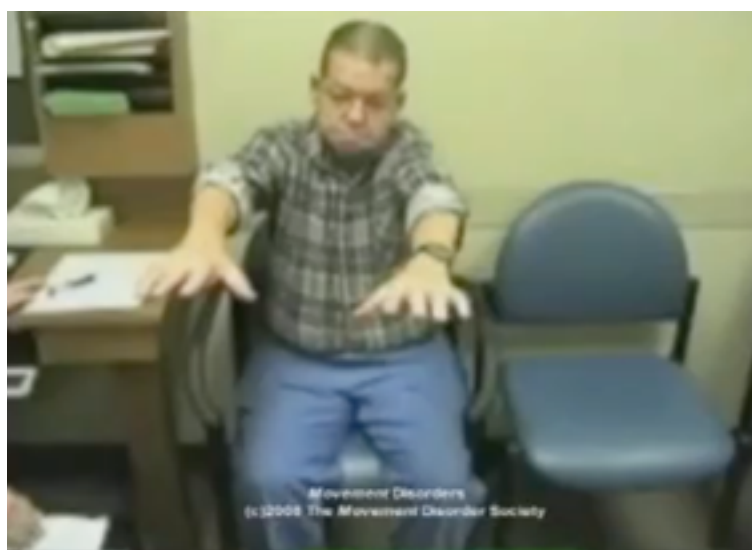
“ **Essential tremor plus:** *Tremor with the characteristics of ET and additional neurological signs of uncertain significance such as impaired tandem gait, questionable dystonic posturing, memory impairment, or other mild neurologic signs of unknown significance that do not suffice to make an additional syndrome classification or diagnosis. ET with tremor at rest should be classified here.* ”



Essential tremor

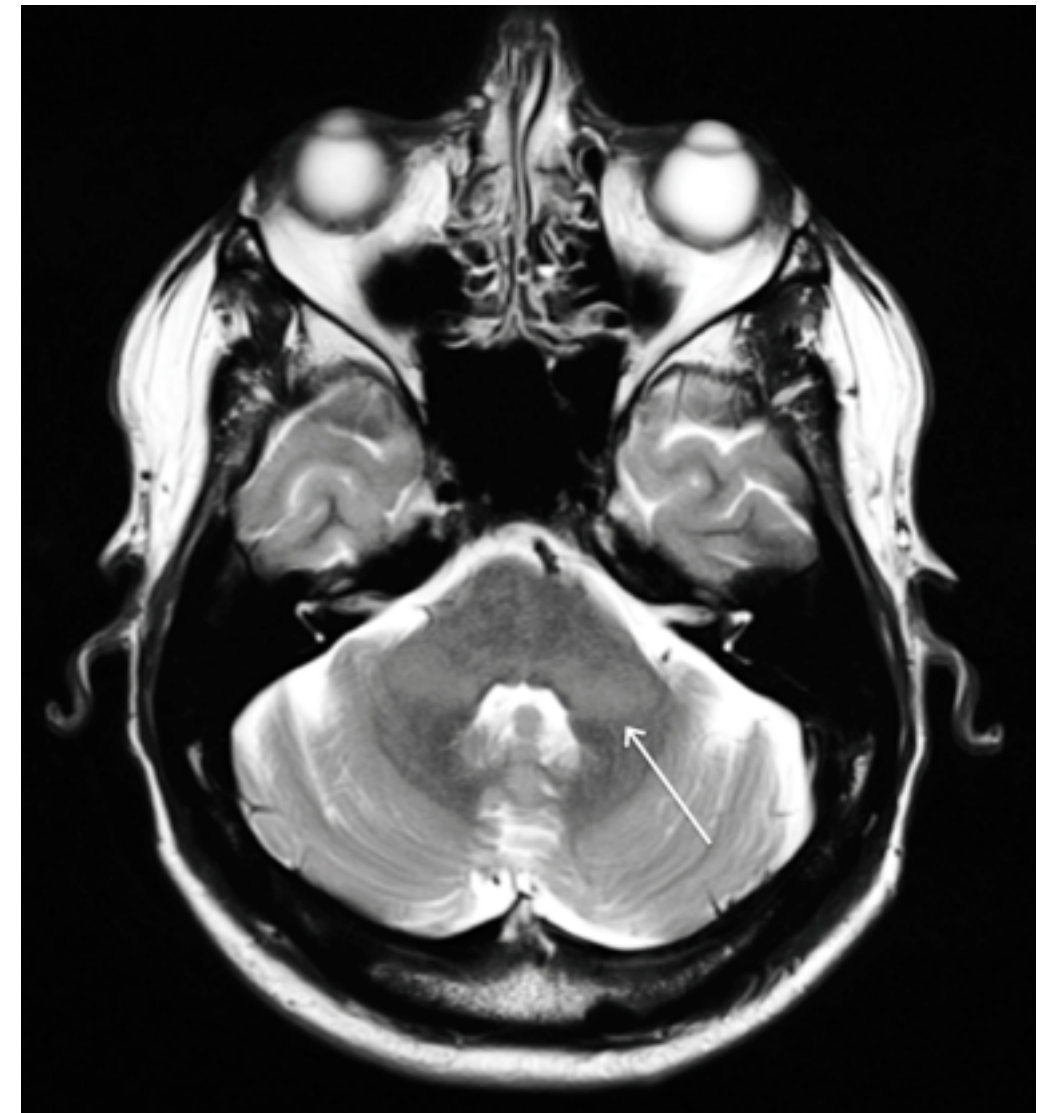
• • • • • or the essential
tremors?

That is the question





Progressive Ataxia, Tremor, Autonomic Dysfunction, and Cognitive Impairment: FXTAS

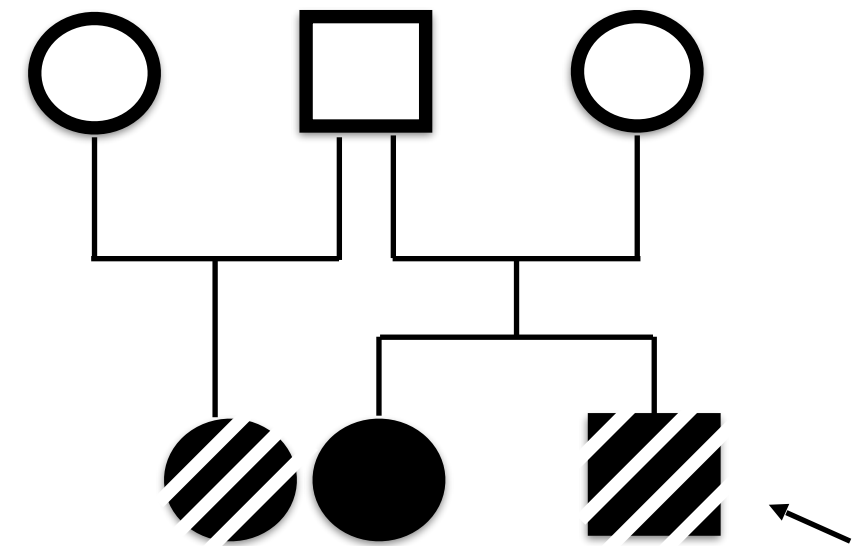


Axial MRI T2-weighted sequence showing the “MCP sign” bilaterally (white arrow).

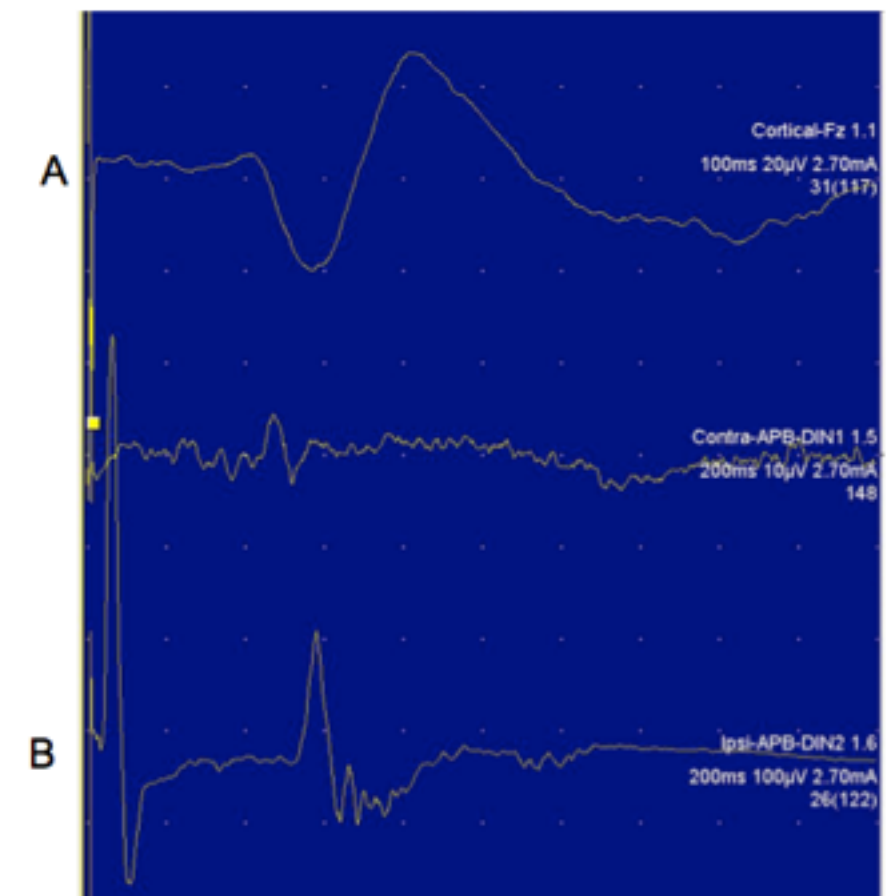


From essential tremor to Klinefelter through Fragile X, an unexpected journey

Tanya L. Harlow et al, 2008

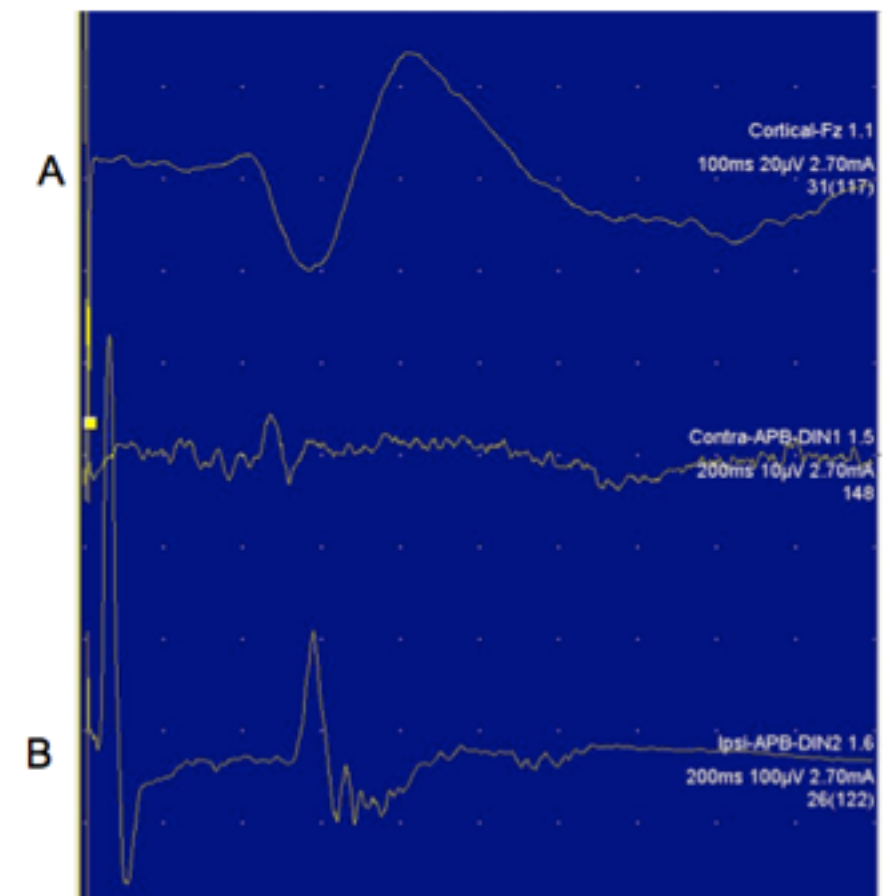
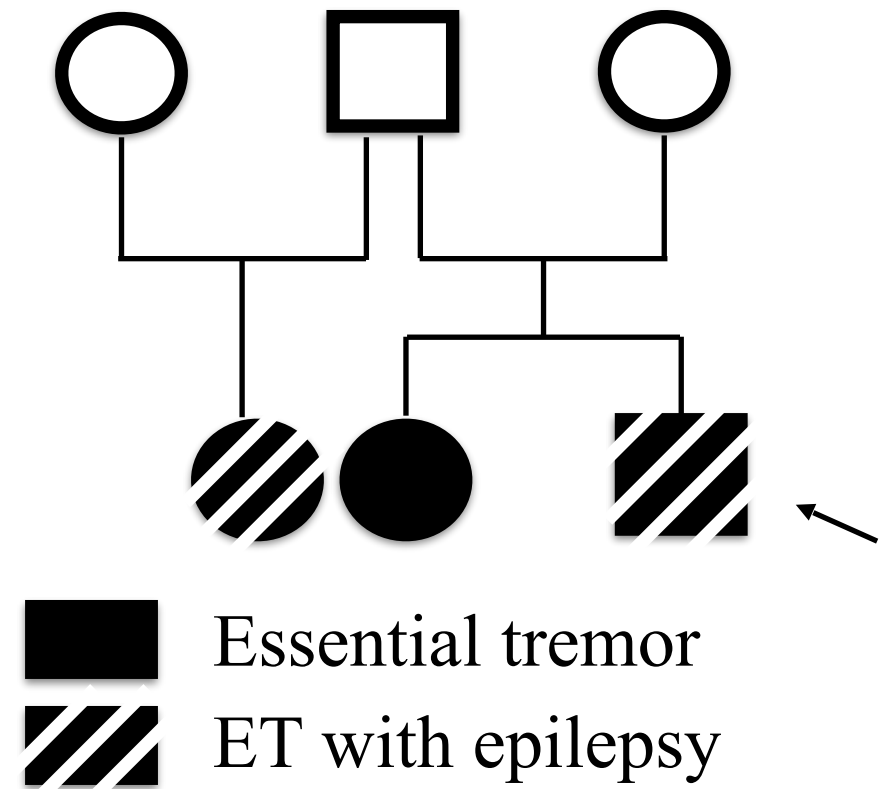


■ Essential tremor
▨ ET with epilepsy





Familial cortical myoclonic tremor with epilepsy







Bimodal distribution of age of onset

Peaks occurring during:

➤ Second or third decades

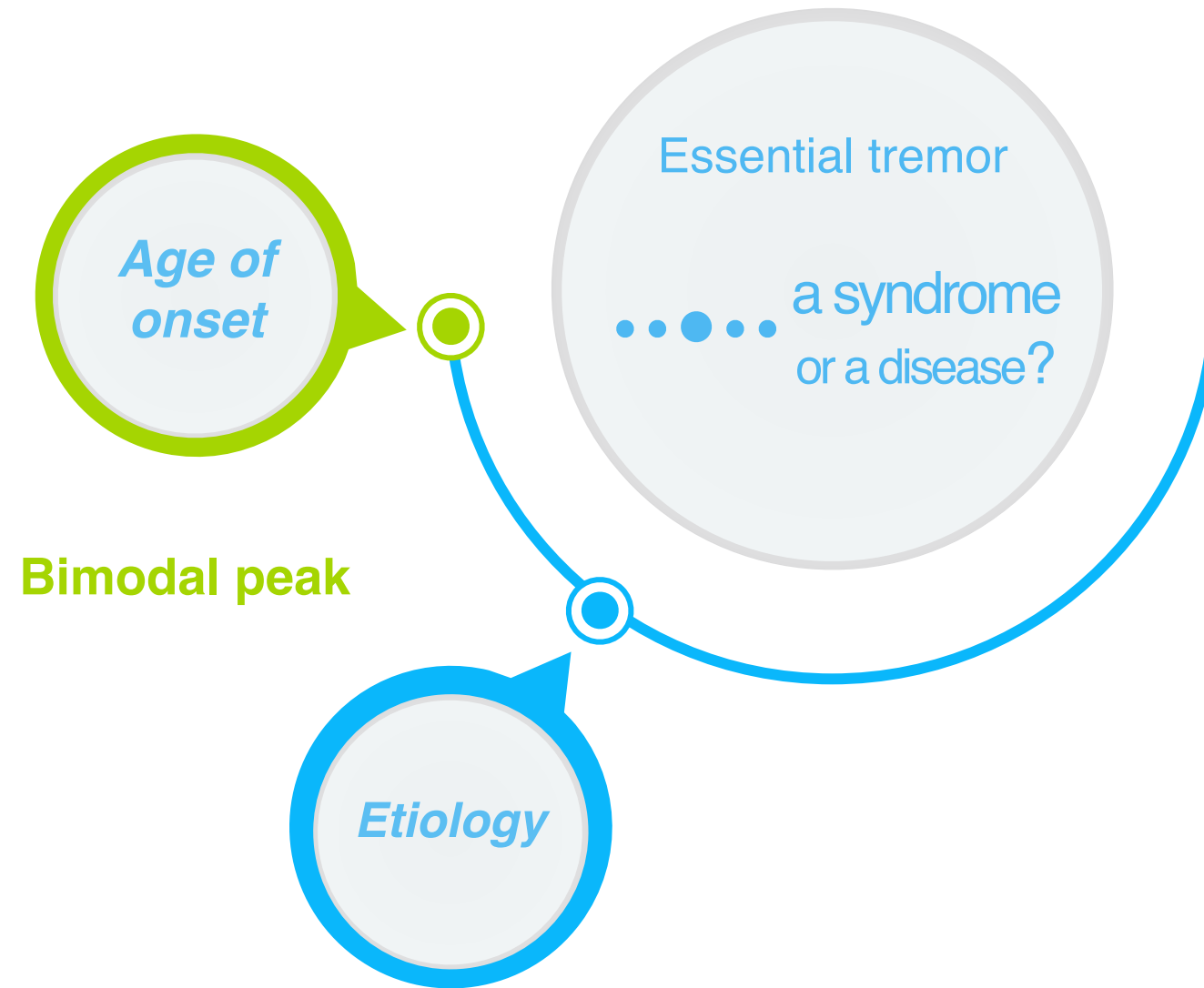


associated with greater
response to alcohol;
more likely to be familial

➤ Seventh or eighth decades



associated with greater
probability of cognitive
impairment





Lack of a defined etiology

Genetic factors:

- Linkage studies have demonstrated three loci (2p24, 13q13, and 6p23) that are of possible significance in ET. However, all candidate regions remained unconfirmed and no causative genetic variant was detected.
- More recently, genome-wide association studies have identified a number of common variants that are associated with a modest elevation in risk of ET

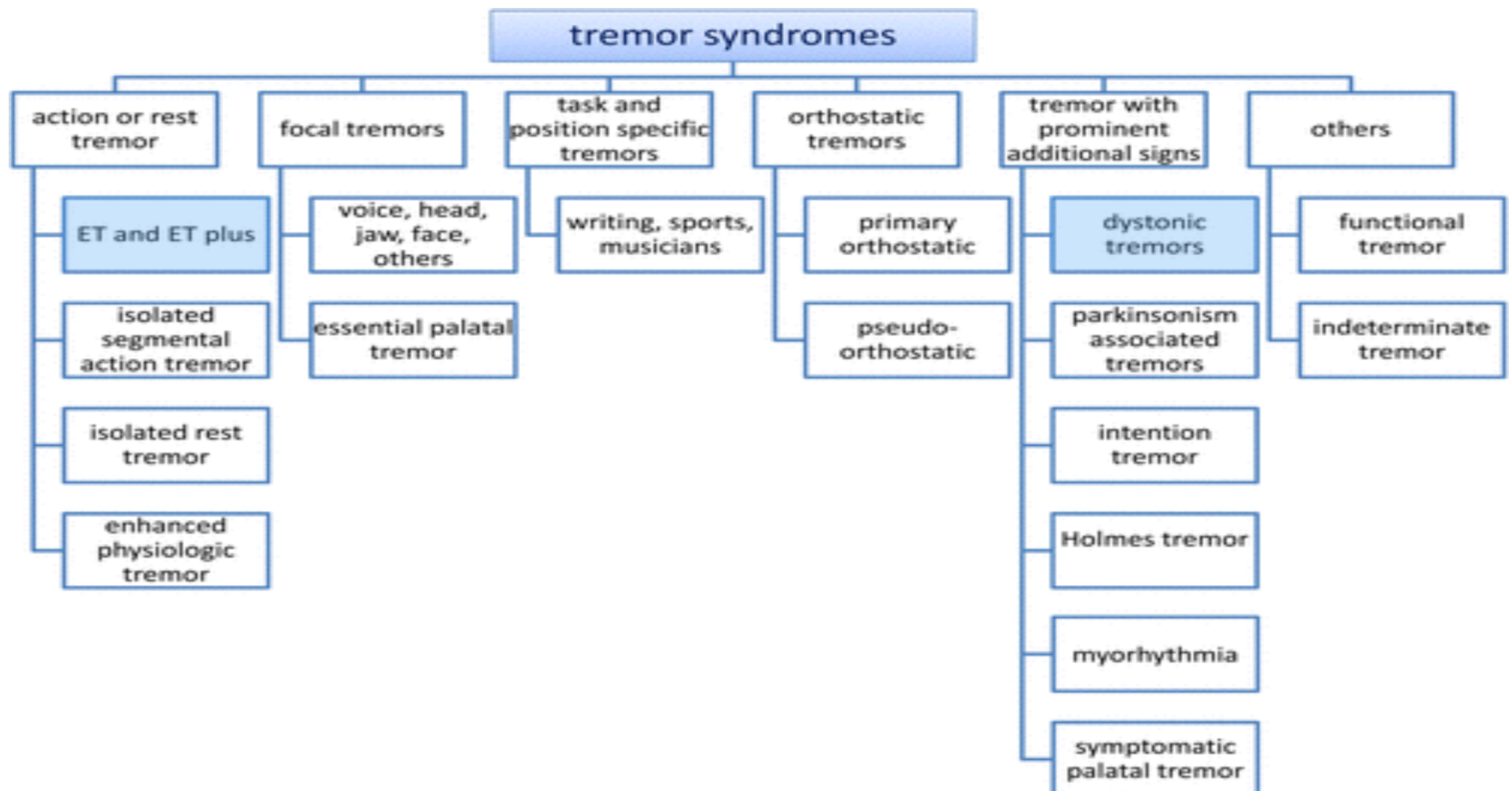
It is important to emphasize that the definition of ET in Axis 1 allows for the existence of multiple etiologies for this common syndrome. Patients frequently

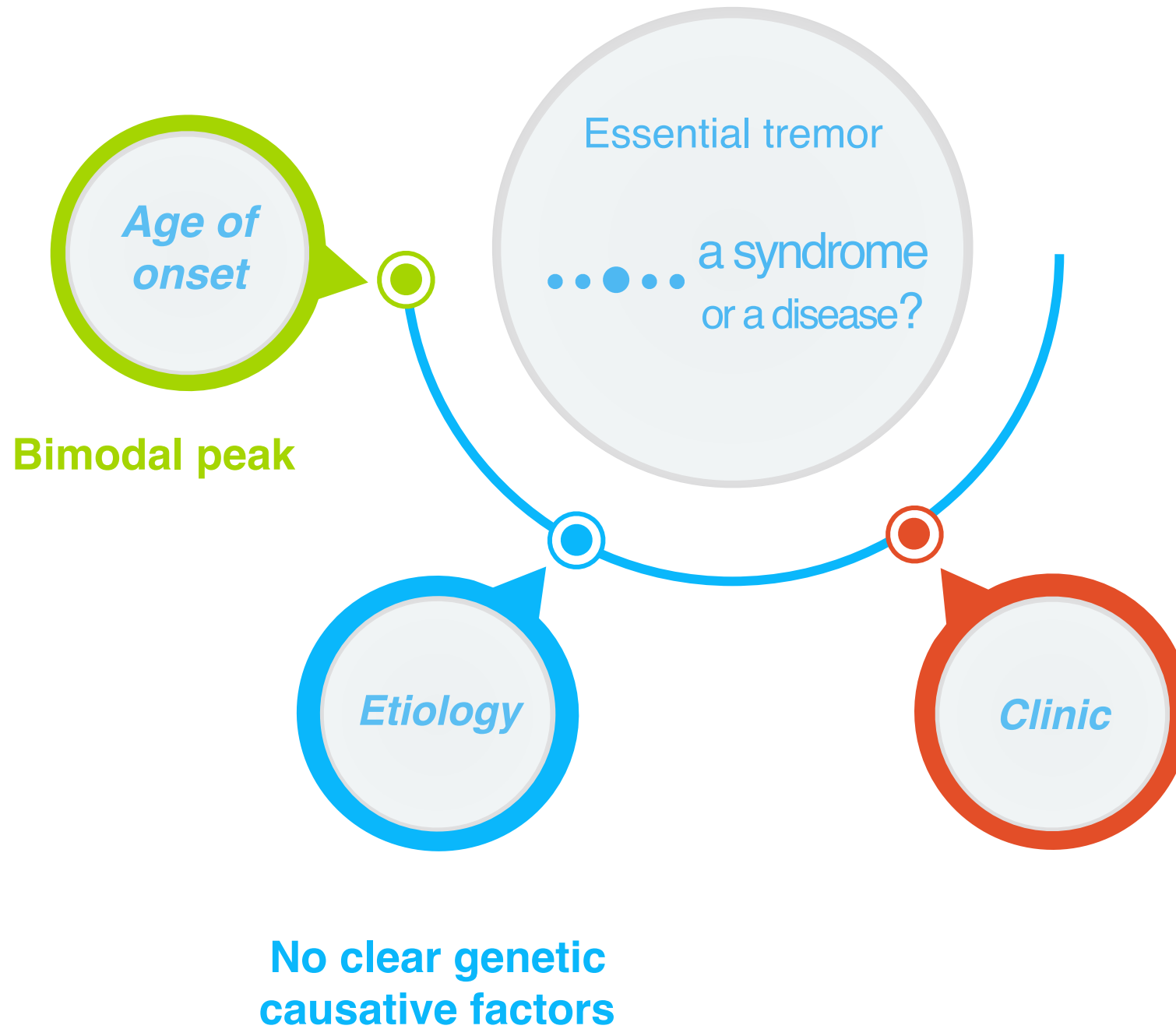


“

ET is a syndrome that may evolve into another tremor syndrome. Tremor of **less than 3 years' duration** that otherwise fulfills criteria for ET should be labeled during the observation period as **indeterminate tremor**.

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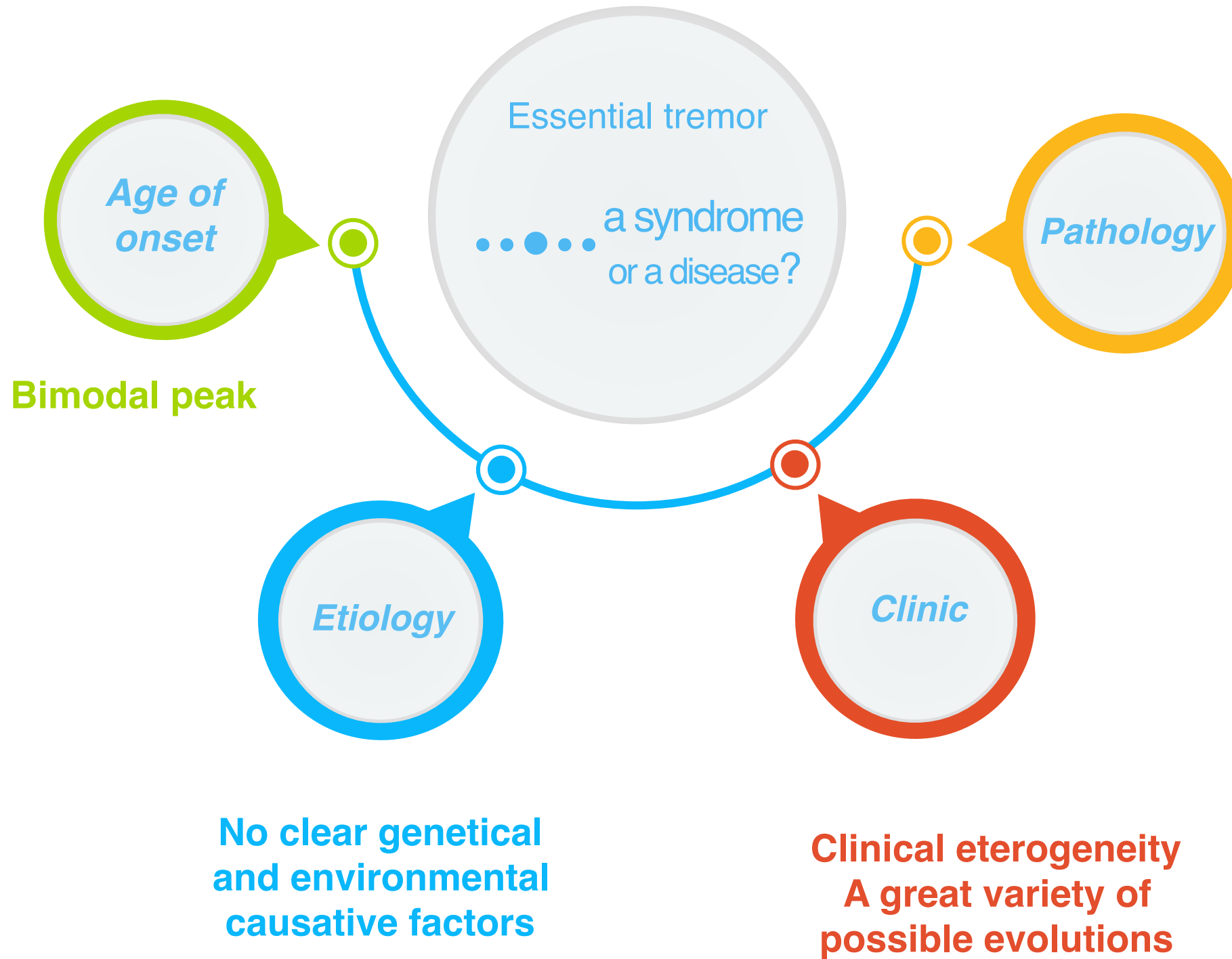


Clinical variability

- It can occur over a wide non-specific frequency range (4-12 Hz)
- It may exceed its postural amplitude at rest or on action as in cerebellar outflow tremor

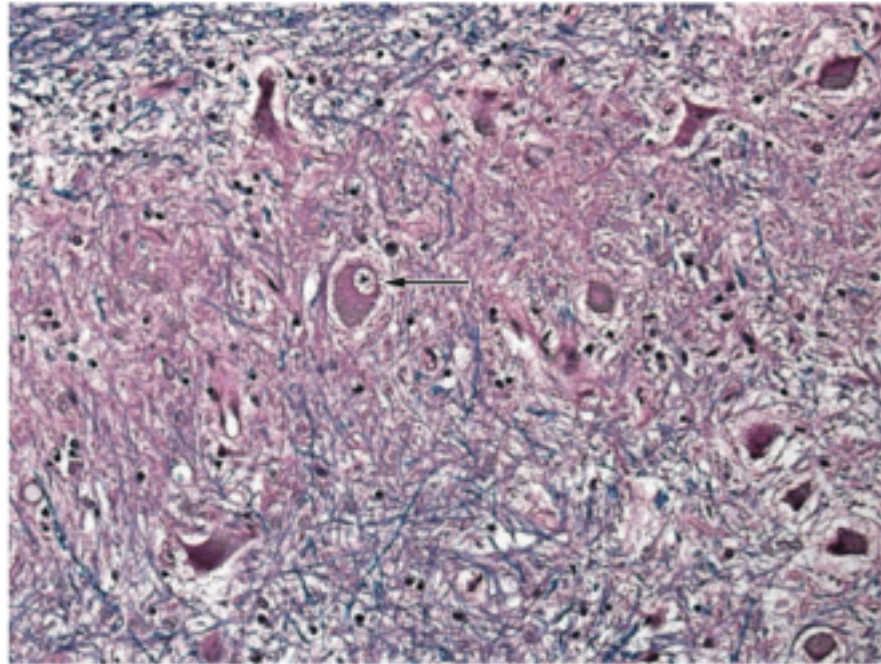
In the past, it has been associated with:

- mild cerebellar signs
- Cognitive impairment
- Psychiatric disturbances (e.g. anxiety, depression, social phobia)
- sensory abnormalities (olfactory deficits and hearing loss)
- Poor nocturnal sleep as well as RBD

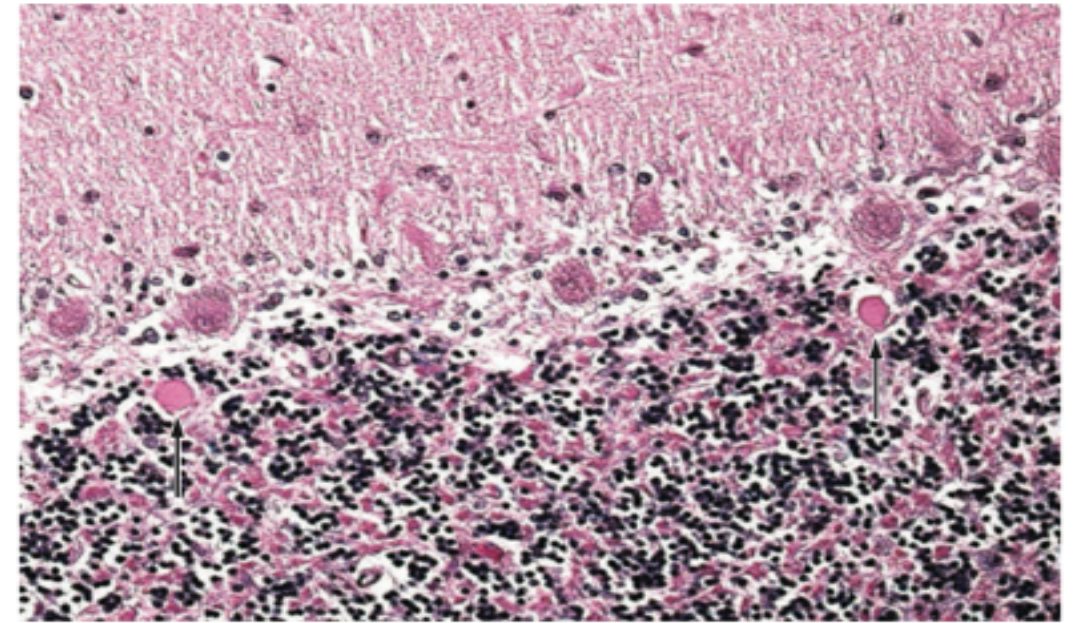




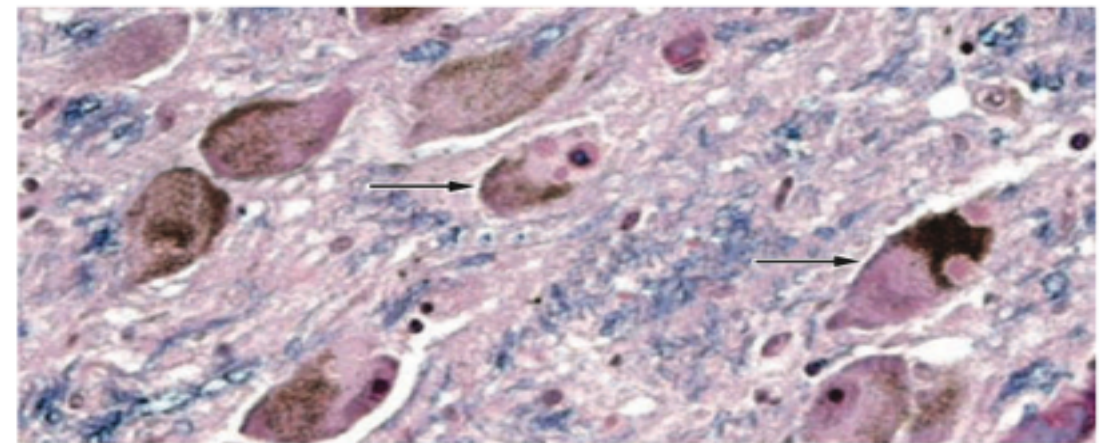
Different pathologies



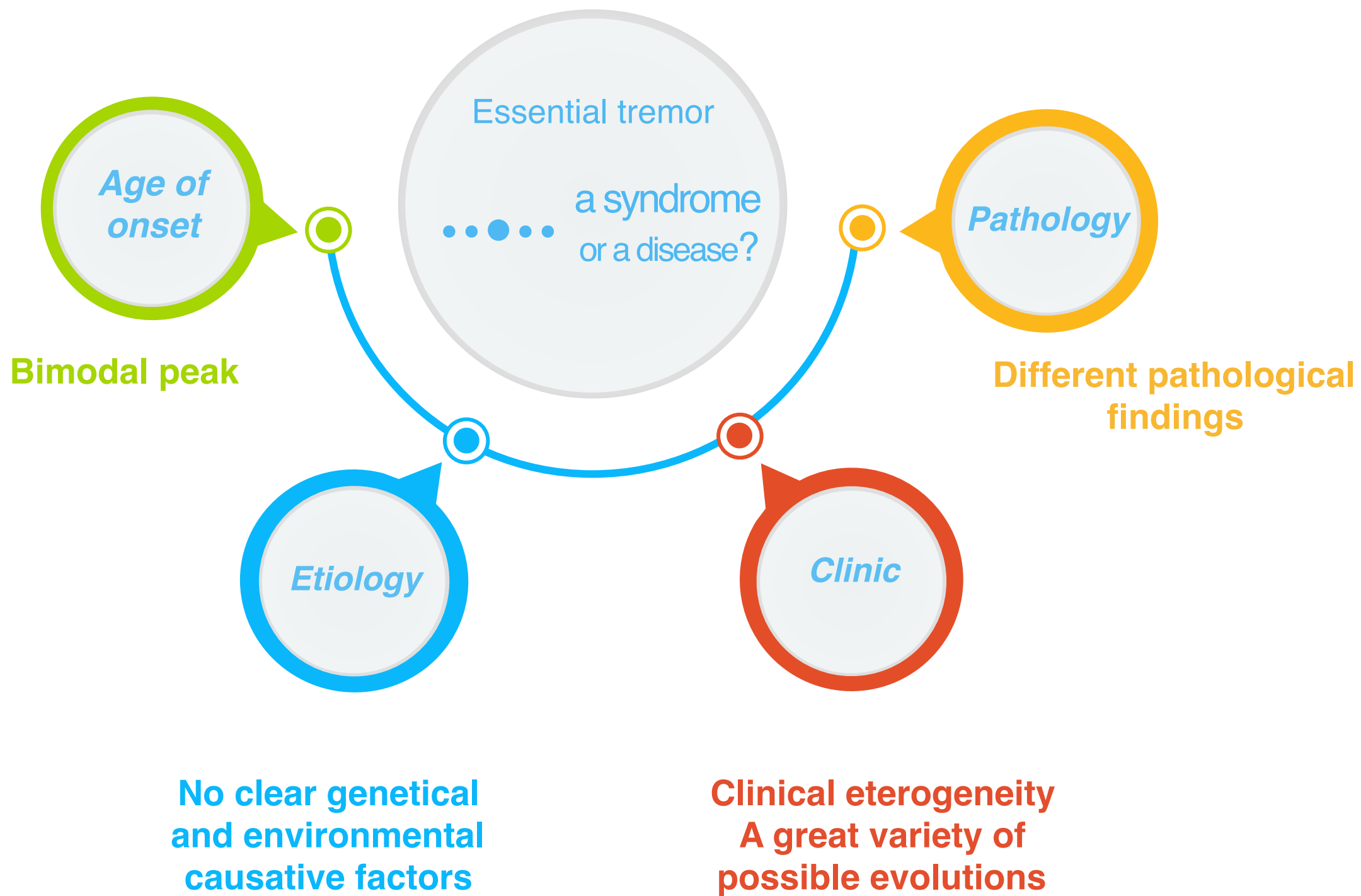
Section of the dentate nucleus from an individual with essential tremor, showing neuronal loss. The neurons that remain are small and angular. One normal, rounded dentate neuron remains in the center (arrow).

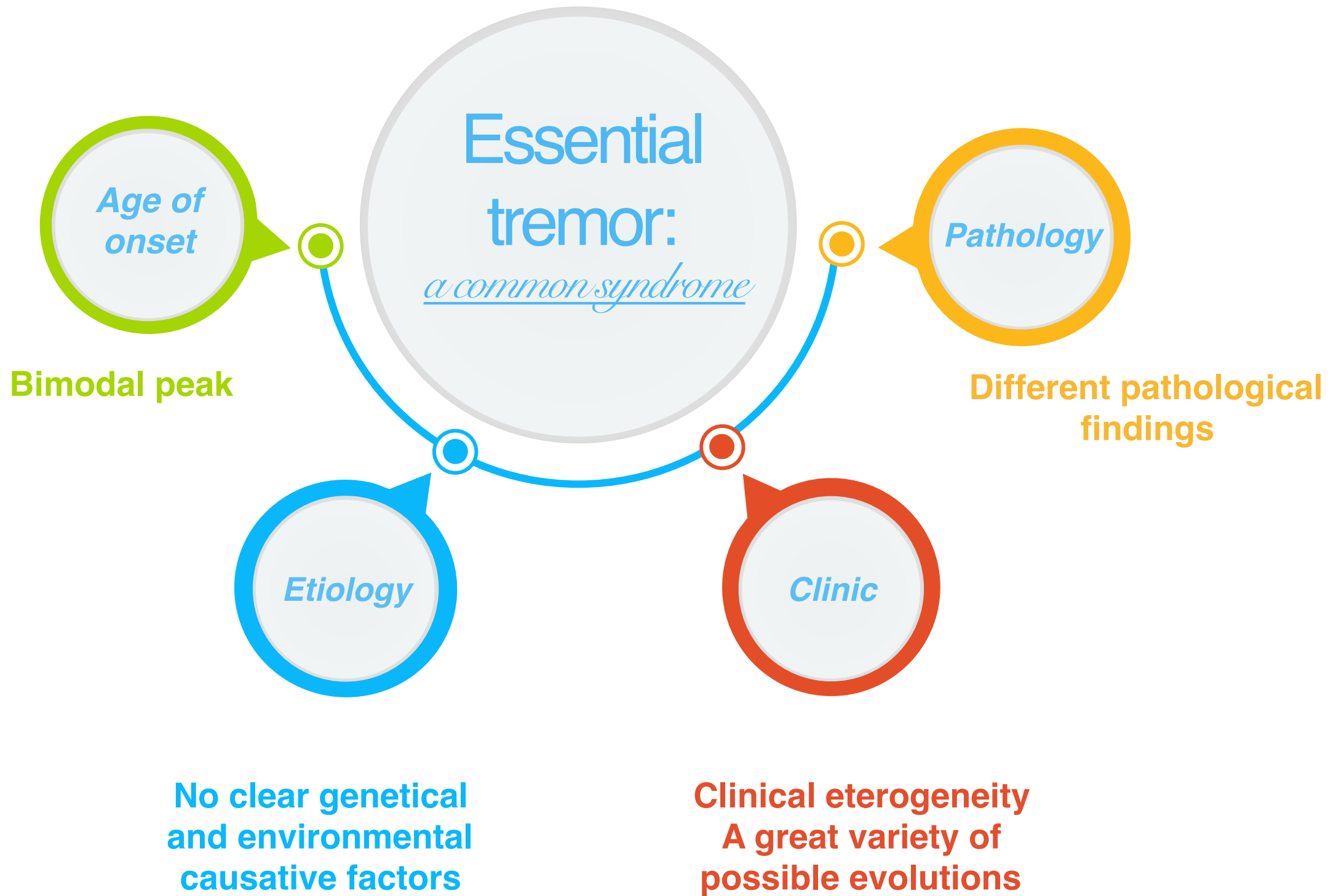


Section of cerebellar folium from a patient with essential tremor, showing two torpedoes (arrows).



Multiple Lewy bodies (arrows) in a section of the locus coeruleus from a patient with essential tremor.







Thank you for your attention