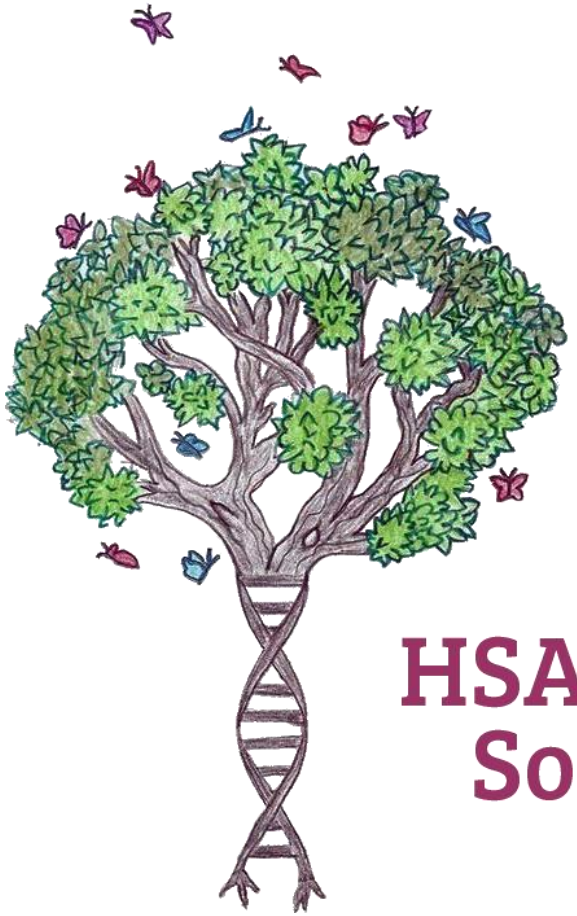


HSAN1E Society

Krista R. Hemming

Vice President

HSAN1ESociety.org

HSAN1E Society

- HSAN1E Society is a 501(c)(3) non-profit organization,
- Started in 2015, by members of an affected family.
- HSAN1E Society's purpose is:
  - create awareness of HSAN1E
  - provide emotional support to families affected by HSAN1E
  - help facilitate research through education and contributions

What is HSAN1E

- Hereditary Sensory and Autonomic Neuropathy, Type 1E
- Alternate Names:
  - Hereditary Sensory Autonomic Neuropathy with dementia and hearing loss
  - DNMT1 complex disorder with hearing loss and dementia

HSAN1E – The Basics

- Ultra Rare Genetic neurological disorder
  - Autosomal Dominant
  - Mutation in the DNMT1 gene
    - DNA Methylation
    - Embryonic Development
- Severely affects the Peripheral and Central Nervous System
- Three Main Symptoms:
  - Hearing Loss
  - Peripheral Neuropathy
  - Dementia
- 100% Fatal

Note
Related Symptoms or Disorders:

Narcolepsy, seizures, optic atrophy, myoclonus, cerebellar ataxia, FTD

HSAN1E – Onset and Progression

- Adult onset
  - Between 32 – 37 years
    - hearing loss, cognition or behavioral changes (dementia)
  - But can vary even among members of same family, De Novo mutations

- Progressive disease
  - 10-15 years between onset and death
    - Hearing loss – steady decline
    - Peripheral neuropathy - unsteady gait/balance, ulcers, immobility
    - Dementia – cognition decline, behavioral, psychiatric, psychosis
    - Other Issues: digestive tract, speech
    - Total Body/Mind failure

- Average life span of affected individual is 50 years

HSAN1E –Diagnosis

- HSAN1E Diagnostic Testing
  - Identification of 3 main symptoms
    - Hearing Tests
    - Cognitive Assessment
    - Nerve Conduction Studies
  - Patient/family history
  - DNA testing
    - Targeting mutations on the DNMT1 gene

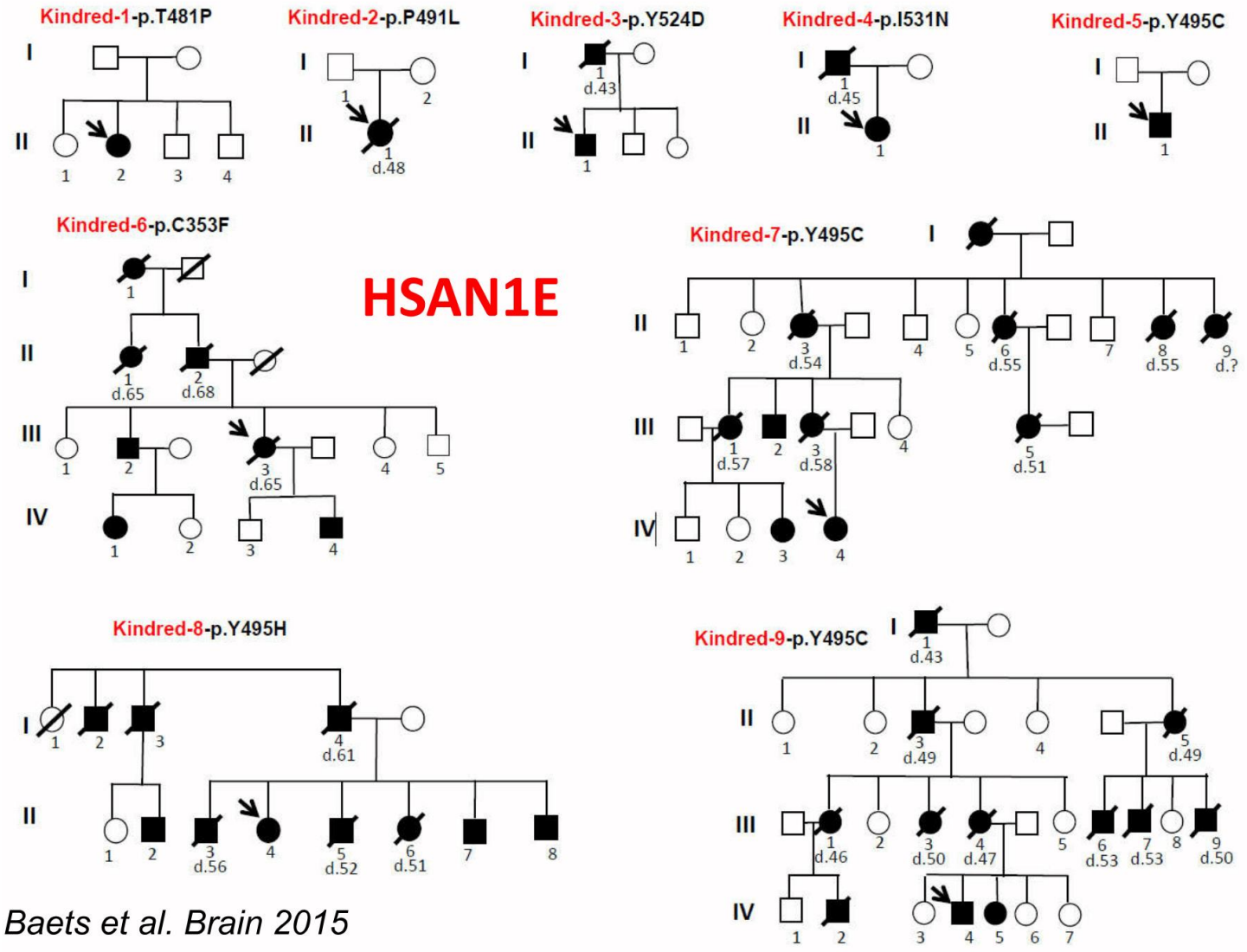
Due to lack of awareness, HSAN1E is largely misdiagnosed, or undiagnosed

HSAN1E - Treatments

- No Cure
- Can Manage symptoms
  - Hearing aids
  - Medications
  - Wound care
- Need team of specialists and caregivers
  - Neurologist, audiologist and Podiatrist
  - Caregivers, help managing life
- End of life care

HSAN1E Society

Frequency of disease and varied phenotypes



Baets et al. Brain 2015

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