HD 101—for newcomers

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Presenter Disclosures

Martha Nance

The following personal financial relationships with commercial interests relevant to this presentation existed during the past 12 months:

No relationships to disclose or list
Thank you to...

George Huntington

Nancy Wexler

Ira Shoulson
Clinical features of HD

- Clinical aspects
  - Movement disorder
    - Involuntary movements
    - Voluntary movements
  - Cognitive disorder
  - Emotional disorder
    - Wide range of symptoms
  - (weight loss)
Clinical features of HD

• Onset age
  – Average onset 35-40 years (range 2-90)
  – Average duration 15-20 years

• Prevalence 4-7/100,000
• ?30,000 affected in US
• 150,000+ at-risk
• Seen in all ethnic groups
  – ?higher incidence in Caucasians
Family histories in HD
Dominant inheritance

Each child of a person with HD has a 50% chance of developing HD.
What causes HD

• Caused by a mutation in the Huntingtin gene
• Type of mutation: “CAG repeat expansion”
  – 10-35 CAG repeats is normal
  – 36- above is not normal and can cause HD
• A blood test can “diagnose” HD
• Everyone with HD has a CAG repeat expansion in the HD gene
• CAG repeat expansions between 36-39 may not lead to symptoms within a normal lifespan
CAG repeats in a family

Normal repeat numbers don’t change much; abnormal repeat numbers tend to get bigger when passed from parent to child.
MRI Scans in HD

Normal Subject: Age 38

HD Subject: Age 31
Care-defining characteristics

• Brain disease
  – Neurologic and psychiatric symptoms predominate
• Progressive (neurodegenerative), fatal
  – Involves caregivers/family
  – Includes changes in relationships, work, ability to care for self, ability to live at home, and death
• Adult-onset (usually)
• Chronic
  – Wide-ranging effects on patient and family
  – Long-term relationship with health care team
• Genetic
  – Family members at risk
Stages of HD

• Shoulson-Fahn scale
  – Assesses work (0-3), money (0-3), chores (0-2), ADLs (0-3), Residence (0-2)
    • Stage 1 (11-13) — work, relationships, diagnosis
    • Stage 2 (7-10) — dx, driving, work, enjoyment
    • Stage 3 (3-6) — transition time: personal help needed
    • Stage 4 (1-2) — in-home help vs. nursing home
    • Stage 5 (0) — dignity in late stages; terminal/Hospice
Medications for HD

- Chorea
  - Neuroleptics (haloperidol, thiothixene, chlorpromazine)
  - Atypical neuroleptics (risperidone, olanzapine)
  - Tetrabenazine (FDA-approved for HD)
- Depression, anxiety, paranoia, explosive behavior, irritability, hallucinations
  - Many drugs
- Cognitive dysfunction
  - (Alzheimer’s drugs: donepezil, memantine, others)
- Weight loss
  - Eat!
Vitamins (and other unproven treatments)

- Probably won’t hurt, might help
  - Multivitamin, B, E (<400 IU), Coenzyme Q10 (>1000mg—expensive), omega fatty acids, creatine
- Probably won’t hurt, probably won’t help
  - Blueberries in human doses, most nutraceuticals, blue-green algae, acai berries,…
Vitamins and other unproven treatments

- I don’t know what it is or why it would help
  - Mangosteen, vinpocetine, homeopathy, etc etc
- Sure, why not?
  - Yoga, massage, acupuncture (if appropriate symptoms), tai chi, “Eastern medicine” (done appropriately)
- And there’s exercise, eat right, sleep right, avoid excess caffeine, nicotine, alcohol…
Principles of management: team-based care

Neurologist or psychiatrist
Psychology, neuropsychology
PT, OT, speech
Nurse-case manager
Social worker
Genetic counselor
Dietitian

Chaplain
Medical doctor
Dentist
Research team
Nursing home staff
Lay group volunteers
Centers of Excellence

- Designated by HDSA
- 21 Centers throughout the country with special expertise in HD
- COEs have multidisciplinary care, support services, research
- HCMC HD Clinic is an HDSA COE
You are not alone…

• Care
• Family support
• Genetic testing
• Advocacy
• Fundraising
• Research
• Community