Huntington's Disease Society of America

HD 101—for newcomers

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HDSA encourages all attendees to consult with their primary care provider, neurologist or other healthcare provider about any advice, exercise, medication, treatment, nutritional supplement or regimen that may have been mentioned as part of any presentation.

Presenter Disclosures

Martha Nance

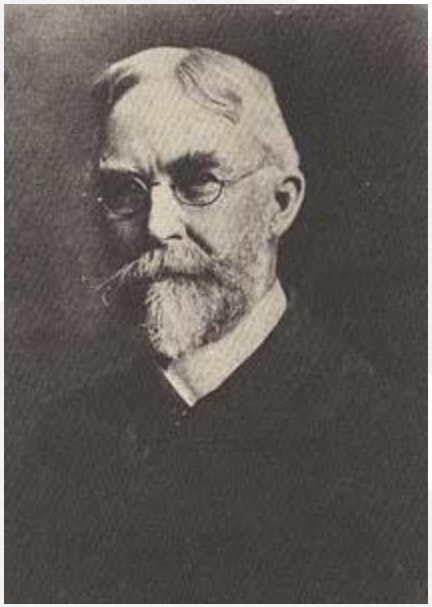
The following personal financial relationships with commercial interests relevant to this presentation existed during the past 12 months:

No relationships to disclose
or list

Huntington's Disease Society of America

Thank you to…

George Huntington



Nancy Wexler

Ira Shoulson

Clinical features of HD

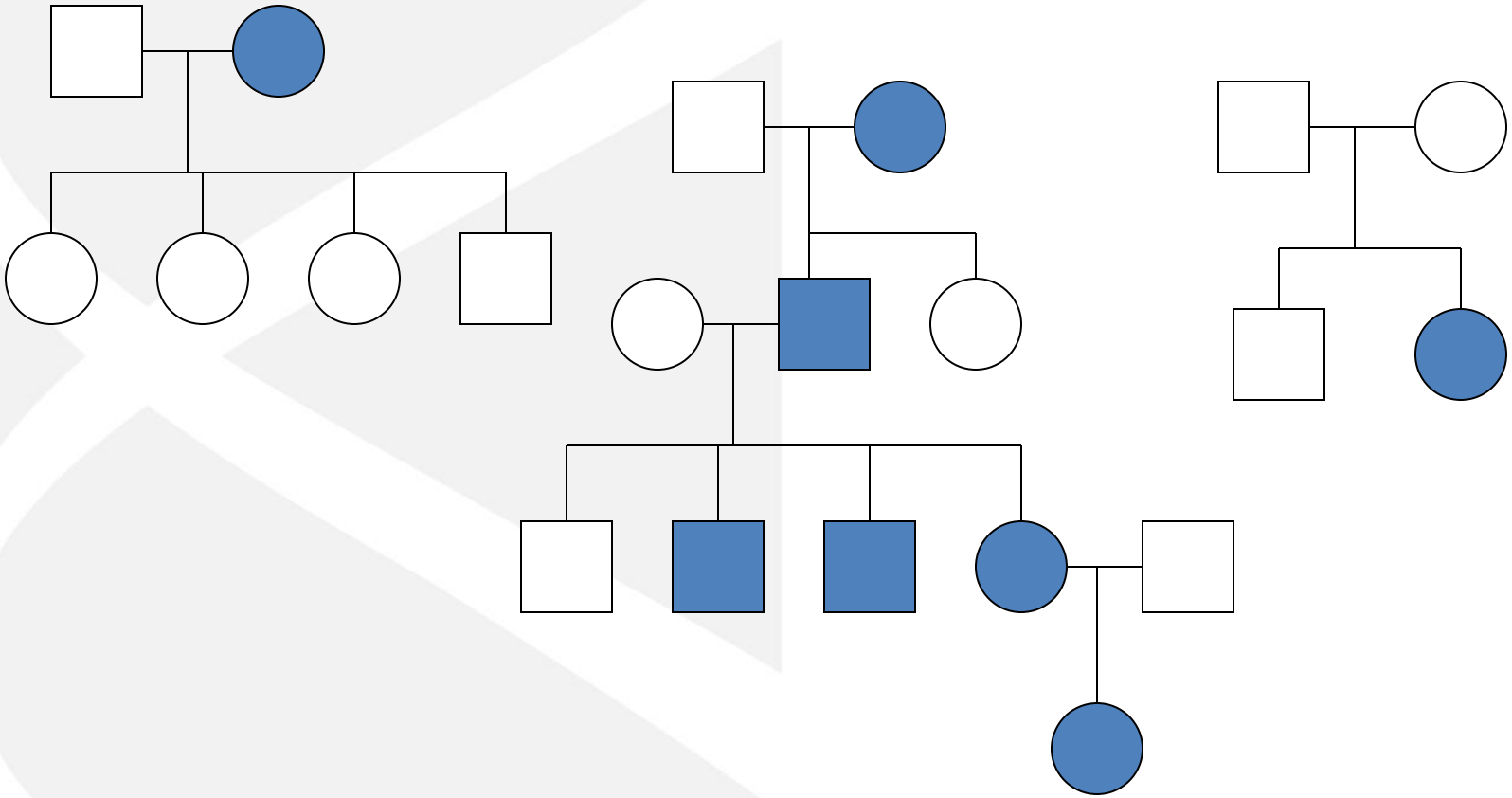
- Clinical aspects
  - Movement disorder
    - Involuntary movements
    - Voluntary movements
  - Cognitive disorder
  - Emotional disorder
    - Wide range of symptoms
  - (weight loss)

Clinical features of HD

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

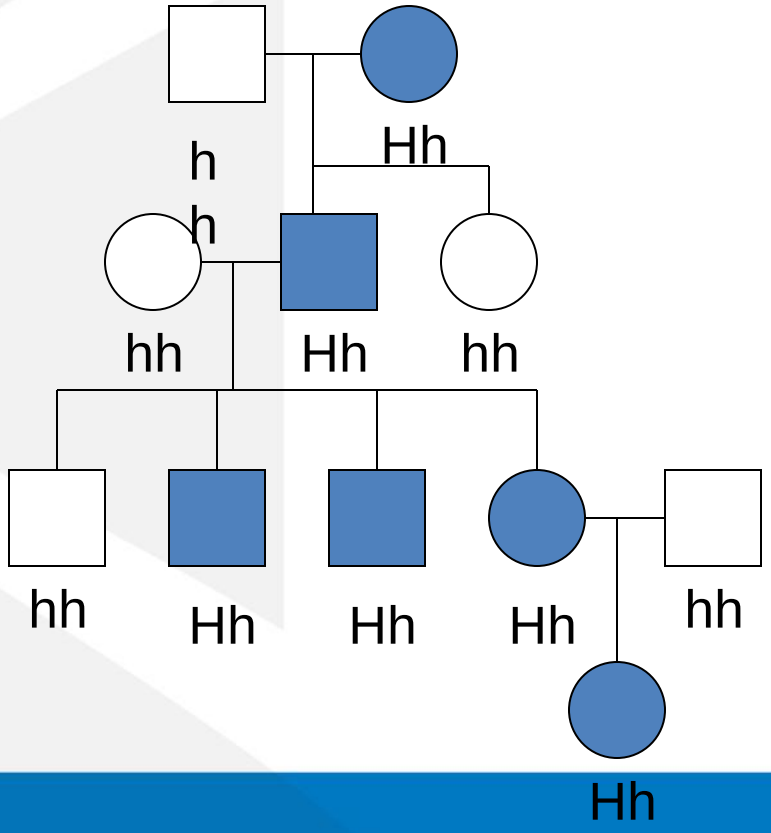
- Prevalence 4-7/100,000
- ?30,000 affected in US
- 150,000+ at-risk
- Seen in all ethnic groups
  - ?higher incidence in Caucasians

Family histories in HD



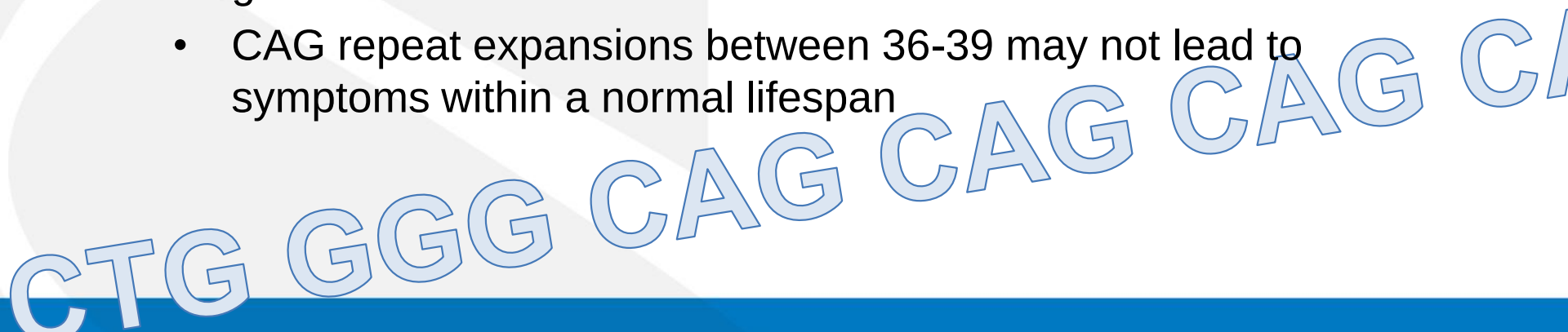
Dominant inheritance

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



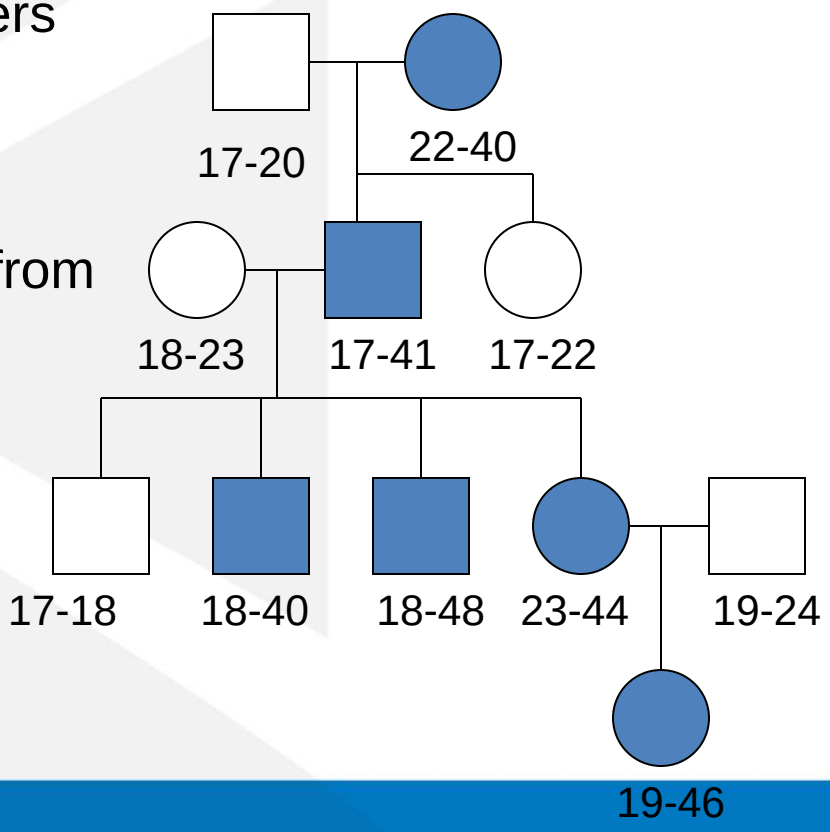
What causes HD

- 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

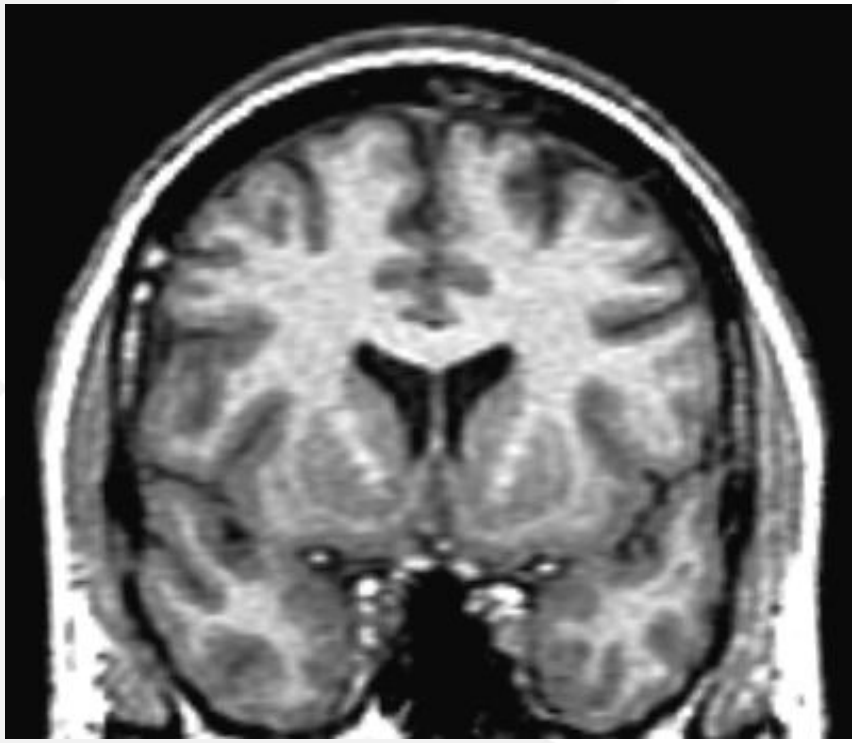


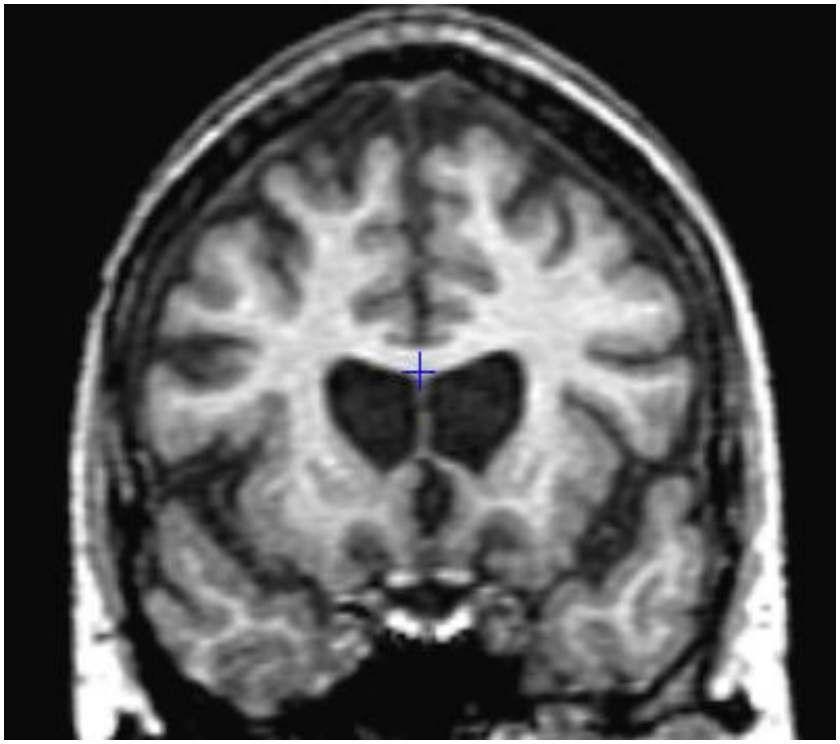
CAG repeats in a family

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