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## Presenter Disclosures

Samuel Frank, MD

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

Consultant to Lundbeck  
Speaker for Allergan



## HD411: An Introduction to Huntington's Disease

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## Goals for Today

- Review what problems people with HD have
- Talk about some facts and myths
- Answer your questions

## George Huntington (1850-1916)

- Family physician
- Worked on Long Island
- Originally read his report to a group of family physicians in Ohio on February 15, 1872



## How Many People Have HD in the US?

- 3,000
- 30,000
- 300,000
- 3 million
- 3 bazillion (almost a google)

### What is a Gene?

- A comfortable pair of pants
- The guy who lives on your street
- Billy \_\_\_\_ from Michael Jackson's song
- The tiny part of our body that is the blueprint for making the building blocks for the rest of our body
- A treatment for Huntington's disease

### Who Gets HD?

- Boys and girls both affected
- All races and religions
- HD usually starts when people are in their 30's or 40's
- Some people show signs of the disease early
  - About 1 in 10-20
  - Called juvenile HD if it starts before age 20

### Genetic Disease

- Each child of an affected parent has 50/50 chance of inheriting disease (like the flip of a coin)
- Does not skip generations
- If you do not inherit the gene, you cannot pass it to the next generation

### Three Main Problems with HD

- Trouble with:
  - Moving
  - Thinking
  - Behaving

### Movement Disorder

- Uncontrolled movements
  - Dance-like movements (chorea)
  - Twisting or extra muscle contractions (dystonia)
  - Slow movements and stiffness (parkinsonism)
- Slurred or irregular speech
- Trouble swallowing
- Coordination problems
- Walking and balance problems

### Thinking Problems

- Slow thinking
- May make bad decisions
- Poor attention

## Behavior

- About half of patients with HD may have sadness or nervousness
- Thinking can get stuck on 1 thing
- Can believe things that are not true
- Personality changes
- Bad sleep
- Trouble controlling themselves

## How Do We Measure Disease Progression?

- Total Functional Capacity (TFC) scale
  - Work
  - Handling money
  - Doing things around the house
  - Caring for themselves
  - Where are they living?
- Not very good for kids, people with behavior problems or later stages of disease

## Myth or Fact?

- Huntington's disease is diagnosed with a blood test for the gene
  - Myth
  - Fact
  - I have no idea

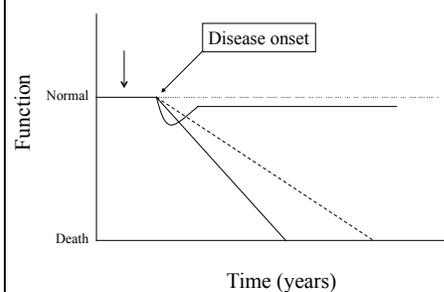
## Diagnosis

- Based on story that patients tell me about the problems they have been having, their family problems and what I see on their exam.
- Sometimes I order other tests like:
  - MRI, CAT scan, EEG or
  - Gene test

## When to Think About Getting the Gene Test

- "At risk" adult patient without symptoms
  - Family planning
  - Financial planning
  - Need to know
- When someone has the disease, but no one else in the family does
  - Adopted
- Juvenile case (when showing problems likely to be the disease)
- Unusual symptoms

## Changing Disease Progression

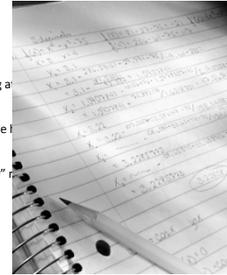


## Treatments

- Movements
- Depression or nervousness

## Hints to Help Patients with Thinking Problems

- Memory
  - Picking from a list is easier
  - Give hints, yes/no questions
- Learning
  - Give simple, clear instructions
  - Limit distractions - focus on 1 thing at a time
- Organization
  - Make lists
  - Keep a big, common calendar in the home
  - Routines
- Slow thinking
  - No response does not mean a "NO" response
  - Have patience



## Treatments – Not Just Medicines

- Social work
- Nutrition
- Social support
- Occupational therapy
- Physical therapy
- Speech therapy

## Questions

- I am confused about something and have a question.
  - (that means everybody else probably is too – just ask)
- I do not have any questions.
- I have a question but am too shy to ask.
- I am overwhelmed and need to think about this for a little while.