Introduction to Huntington’s Disease

Huntington’s Disease Society of America
Center of Excellence at UC Davis
June 4, 2013
Welcome!

HDSA Center of Excellence
UC Davis Medical Center

Vicki Wheelock MD, Neurologist
Terry Tempkin RN ANP, Nurse Practitioner
Lisa Kjer LCSW, Social Worker
Rosy Chow LPT, Physical Therapist
Mara Sifry-Platt MS, Genetic Counselor
Lorin Scher MD, Psychiatrist
Care, Outreach, and Reach for a Cure

Our COE Mission:

- To provide excellent, comprehensive and compassionate care and outreach to HD families
- To provide expert education about HD to families, researchers and health care providers
- To advance HD research

Established 2001
21 HDSA Centers of Excellence

For a complete list of locations, please go to www.hdsa.org, click on "Living with HD, and then "Centers of Excellence"
Huntington’s Disease Definitions

- Slowly progressive, hereditary brain disease that causes changes in movement, thinking and behavior
- Diagnosis made at onset of movement disorder, typically with chorea and impaired voluntary movement.
- Autosomal dominant inheritance
  - High penetrance: everyone with the gene will get HD
  - Anticipation: earlier onset in children of fathers with HD
- 30,000 people with HD in US
  - 150,000 at-risk in US
  - About 2000 people are diagnosed each year
- World-wide occurrence in all populations
  - Lowest in Japan
Who Gets HD?

Each child with a parent with HD has a 50% chance of inheriting the disease.

Myers RH. J Am Soc Exper Ther 2004;255-262
The Search for the HD Gene

Introduction to HD

Dr. Nancy Wexler’s team visited Lake Maracaibo annually starting in 1979, identifying 18,149 individuals from HD families spanning 10 generations.

Dr. Ramon Avila-Giron, student of Dr. Americo Negrette, showed films of HD patients at Centennial Meeting to a skeptical audience, 1972.
1993: The HD Gene Is Discovered

**Huntington gene** → **huntingtin protein**

- Normal number: <31 CAG
- “Gray area”: 32-38 CAG
- Huntington disease: >38 CAG
The Huntington Disease Mutation

Gene

Protein

Normal number:          <31 CAG
“Gray area”:           32-38 CAG
Huntington disease:     >38 CAG
Brain changes in HD


MRI scan

Microscopic view
Symptoms in Huntington’s disease

- Impulsivity
- Balance Problems
- Creativity
- Slow Eye Movements
- Episodic Anger, Irritability
- Slowness of Movement
- Depression, Anxiety
- Chorea: Involuntary Movements
- Multi-tasking
- Restless, Fidgets
- Organizing Concentrating Prioritizing
- Fine Motor Tasks

Normal vs. Advanced HD
Psychiatric symptoms in HD

- Depression: 40 - 80%
- Anxiety: 30 - 40%
- Obsessions/compulsions: 10 - 20%
- Irritability: common
- Apathy: 20%
- Episodic anger: common
- Psychosis: 5%
Depression in HD

- Very common. May occur:
  - before diagnosis
  - at the time of diagnosis
  - later in the disease

- Thoughts of suicide may occur
  - Most commonly occurs around the time of diagnosis
  - Over 25% of patients with HD attempt suicide at some point in the illness.
  - Reported rates of completed suicide among individuals with Huntington’s disease range from 3-13%

- Treatment of depression with counseling, medications, and family and community support prevents suicide
Huntington Disease Stages

- Stage 0: Presymptomatic
- Stage 1: Slightly lower performance at work; independent at home
- Stage 2: Can still work (lower level), still mostly independent at home
- Stage 3: Difficult to work, starts to needs help with financial, home activities
- Stage 4: Unable to work. Needs major assistance with care
- Stage 5: Full-time nursing care required
What kinds of treatments do we have for HD?

- **Symptomatic**
  - Movement problems
  - Thinking/behavior
  - Mood problems
  - Swallow problems
  - Nutrition

- **Neuroprotection:**
  - Delay the onset or slow progression

- **Research is underway**

- **Cure:** Hope from research
Motor symptoms

- Chorea
- Slowness, rigidity
- Dystonia
- Poor balance
- Exercises, physical therapy, medications can help
Tetrabenazine: First drug approved for HD in the US

**Benefits:** Highly effective for chorea

**Side effects:**
- swallow dysfunction
- ↑depression/suicide
- restlessness

**Interactions:**
- Some anti-depressants
- Other anti-chorea drugs

**Dosing:**
- Genetic test
- Monitor EKG

**Cost:** Special program
Balance Problems

May occur early in HD, but more commonly occurs later in the disease.

*Physical therapy and exercise help tremendously!*

Please see the presentation

“Coping With Disability and Muscle Weakness in Huntington’s Disease”

for specific exercises.

Rosy Chow, Physical Therapist
Behavioral/Thinking Symptoms

- “Memory” problems
- Borrow meds used for Alzheimer’s disease
- Apathy
  - Structure, behavioral treatments
  - Stimulant medications
- Irritability
  - Seek triggers, set structure
  - ?Depressed or anxious – treat those symptoms
- Episodic anger
  - See above
  - Sometimes mood stabilizers (anti-epileptic drugs, neuroleptics)
Emotional Symptoms

- Medications used to treat psychiatric symptoms in the general population are usually effective
- Depression
  - SSRI meds (fluoxetine, paroxetine, sertraline, citalopram, buproprion, etc)
- Anxiety
  - SSRI meds, sometimes clonazepam
- Obsessions/compulsions
  - SSRI meds
- Paranoia, delusions (false beliefs), hallucinations
  - Antipsychotics (olanzapine, risperidone, haloperidol, fluphenazine, etc)
Medications for Treatment of Emotional Symptoms

Please see slide presentation “Medications to treat HD” for more specific details
Is there anything that can slow the progression of HD?

- There is no therapy that’s been proven yet to delay the onset of slow the progression.
- We recommend:
  - a good, healthy diet
  - regular exercise
  - avoiding smoking and excessive alcohol
- Some dietary supplements may be helpful
  - Fish Oil (ethyl-EPA)
  - CoQ10
  - Creatine
- Research trials are ongoing.
What should I do about my HD?

- Establish care – Center of Excellence
- Get information
- Take care of your health (not just HD)
  - Healthy diet, exercise, lifestyle
- Find out about medications and therapies
  - Talk to us about supplements
- Get connected:
  - HD Support Groups
  - Northern CA chapter of HDSA
- Consider research participation