A HEALTHIER WORLD THROUGH BOLD INNOVATION






UCDAVIS HEALTH SYSTEM

Introduction to Huntington's Disease

Huntington's Disease Society of America
Center of Excellence at UC Davis
June 4, 2013

SCHOOL OF MEDICINE · UC DAVIS

Welcome!

HDSA Center of Excellence
UC Davis Medical Center

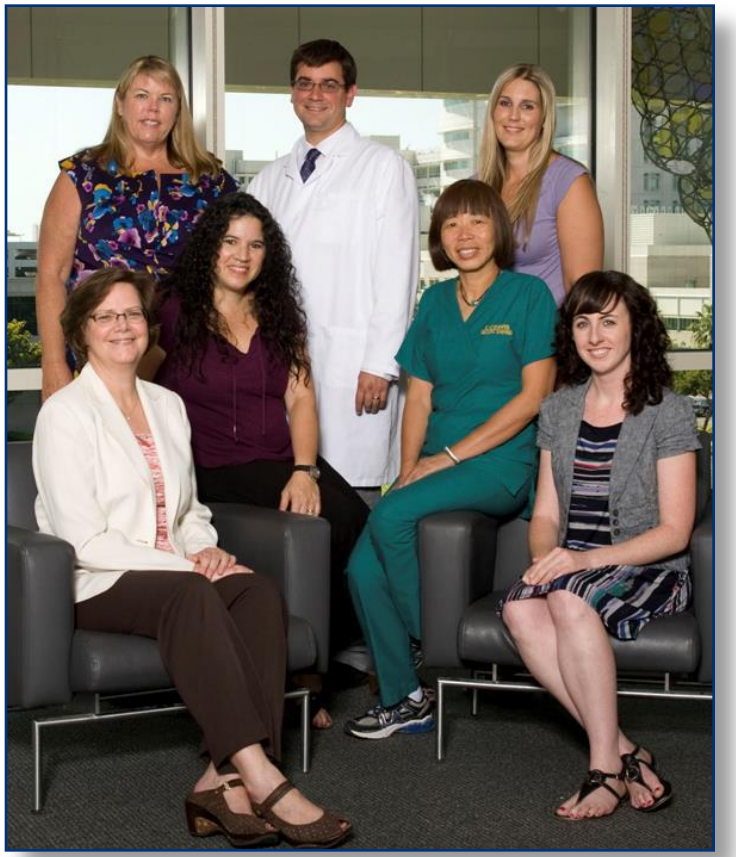
Vicki Wheelock MD, Neurologist
Terry Tempkin RN ANP, Nurse Practitioner
Lisa Kjer LCSW, Social Worker
Rosy Chow LPT, Physical Therapist
Mara Sifry-Platt MS, Genetic Counselor
Lorin Scher MD, Psychiatrist

Care, Outreach, and Reach for a Cure

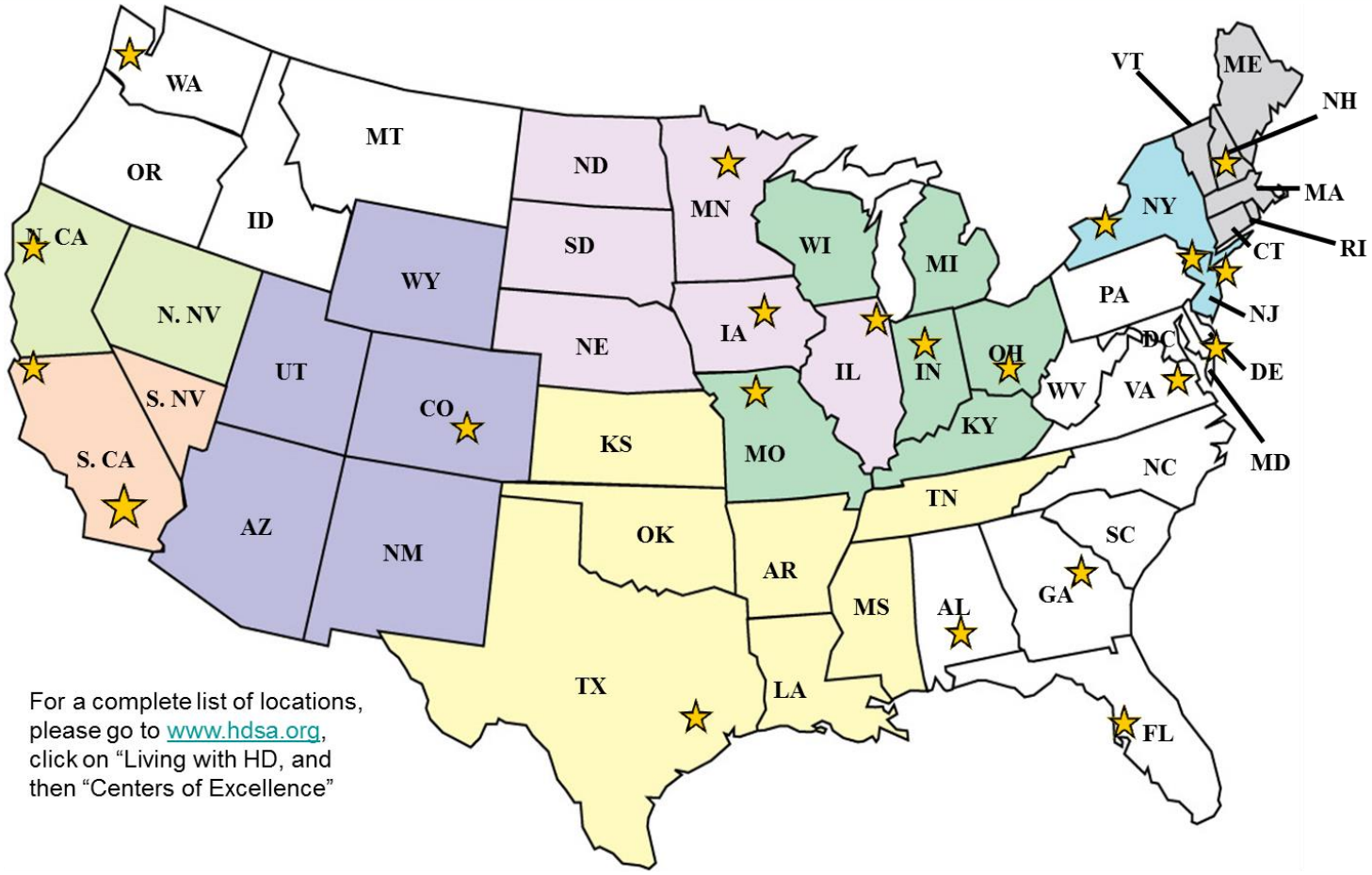
Our COE Mission:

- *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



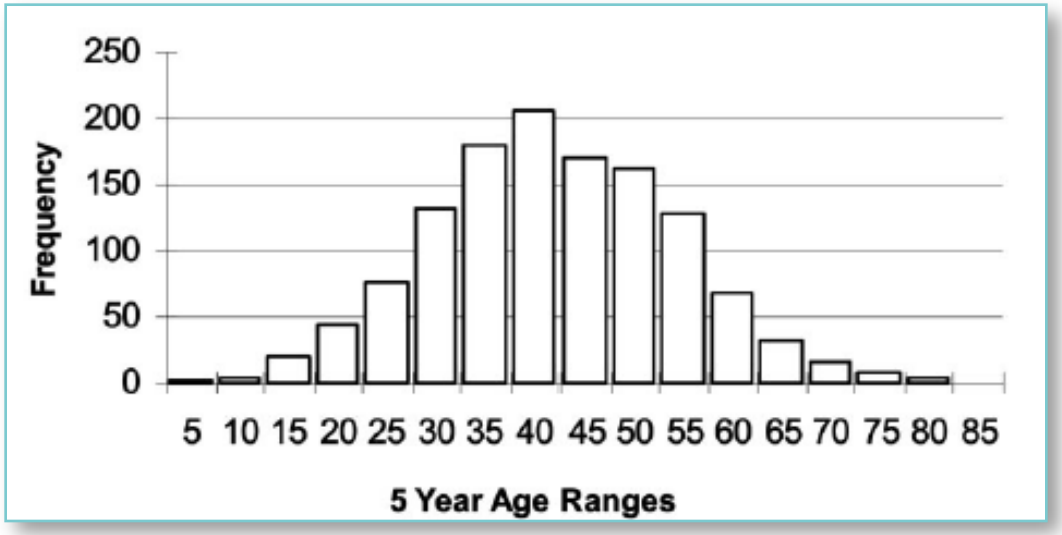
For a complete list of locations, please go to www.hdsa.org, click on "Living with HD, and then "Centers of Excellence"

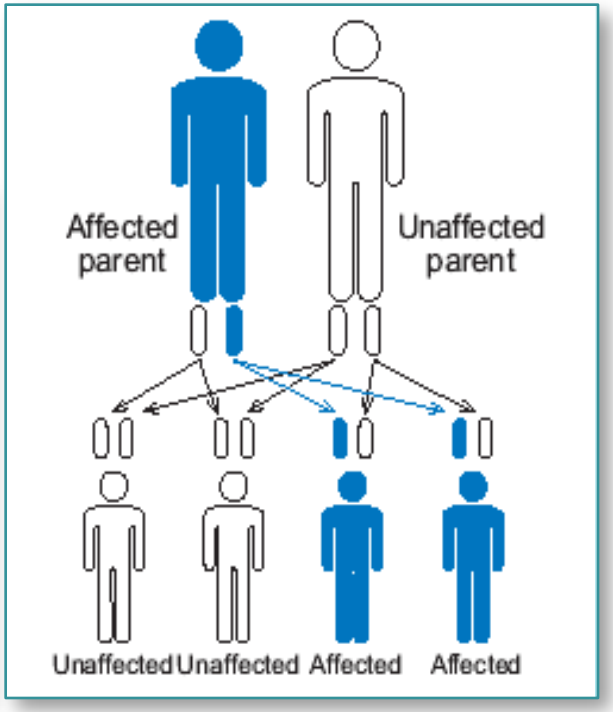
Huntington's Disease Definitions

- 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
  - Abut 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

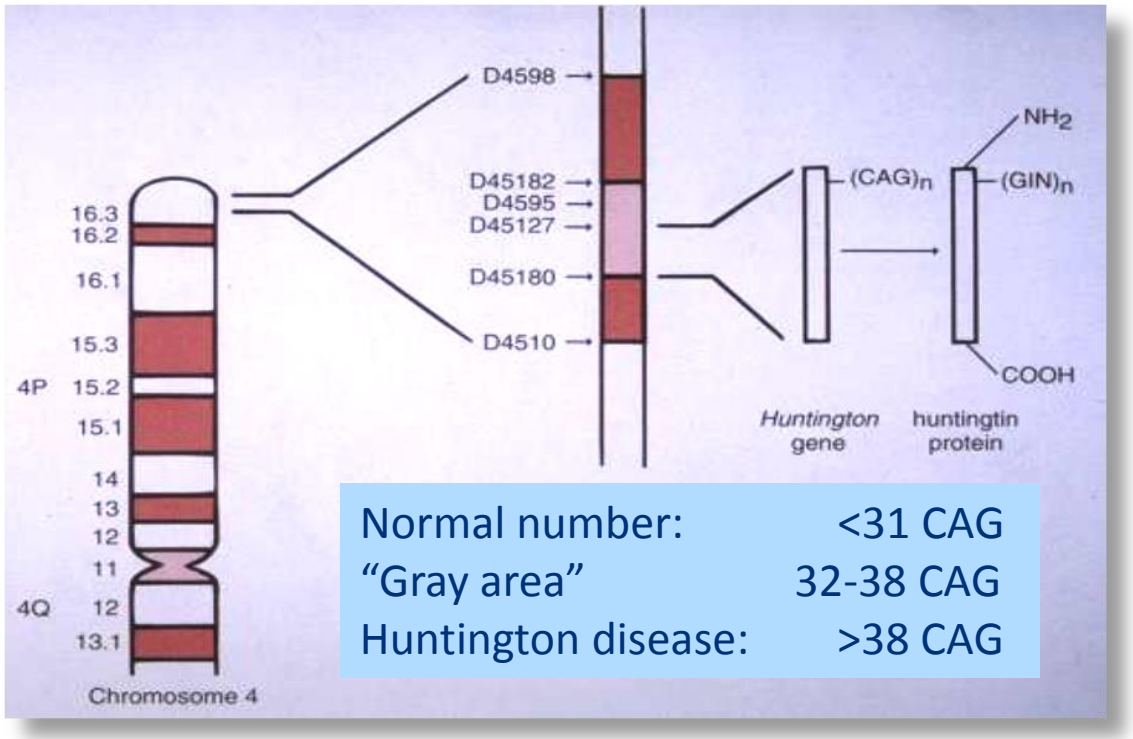
Dr. Ramon Avila-Giron, student of Dr. Americo Negrette, showed films of HD patients at Centennial Meeting to a skeptical audience, 1972.

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

A new study from Nancy Wexler, in Venezuela in the 1990s with a boy with Huntington's disease, suggests there may be ways to delay the onset of the disease.

1993: The HD Gene Is Discovered

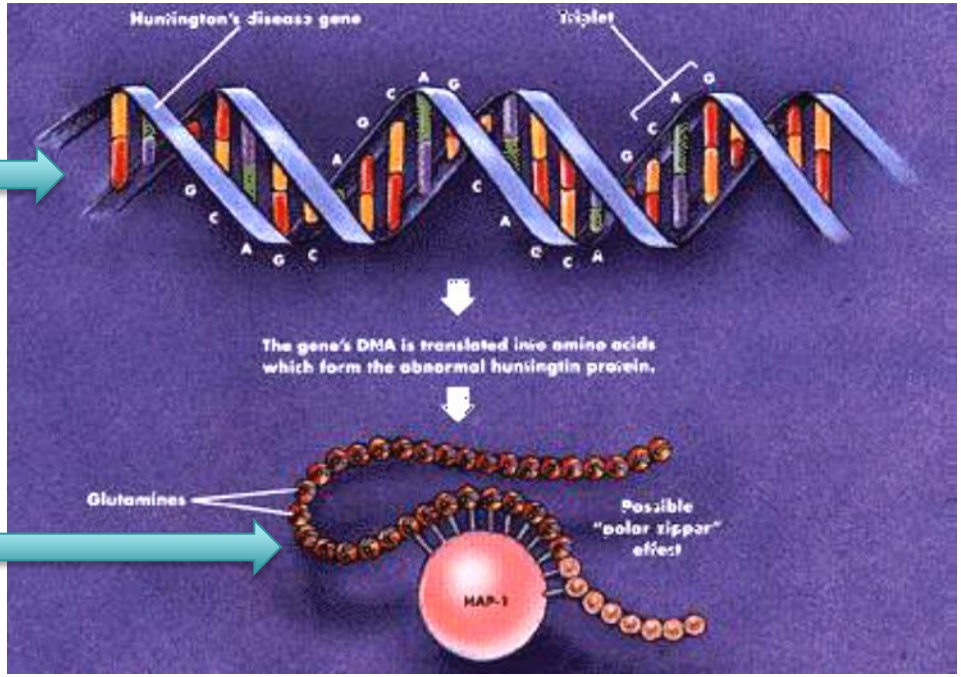
Huntington gene → huntingtin protein



The Huntington Disease Mutation

Gene

Protein

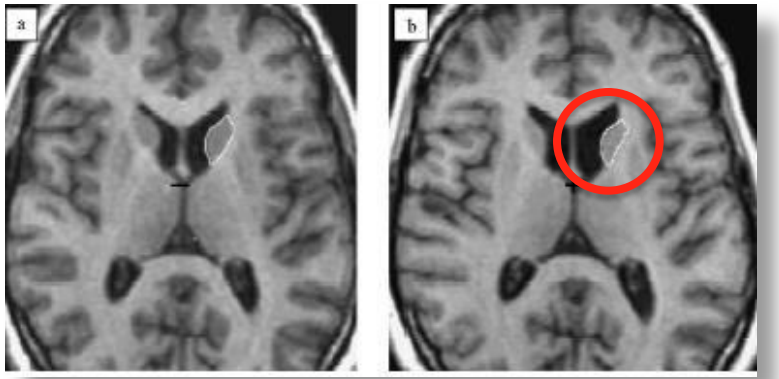


Normal number: <31 CAG
"Gray area": 32-38 CAG
Huntington disease: >38 CAG

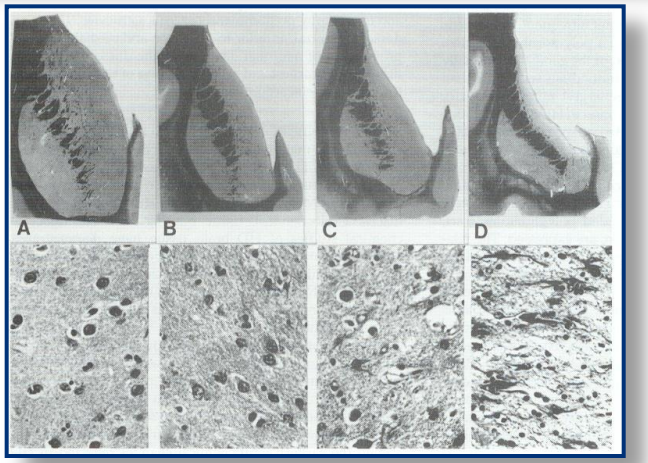
Brain changes in HD

Aylward EH. Brain Research Bulletin 2003;62:137-141

MRI scan



Microscopic view



Thinking

Movement

Behavior

Symptoms in Huntington's disease

Impulsivity

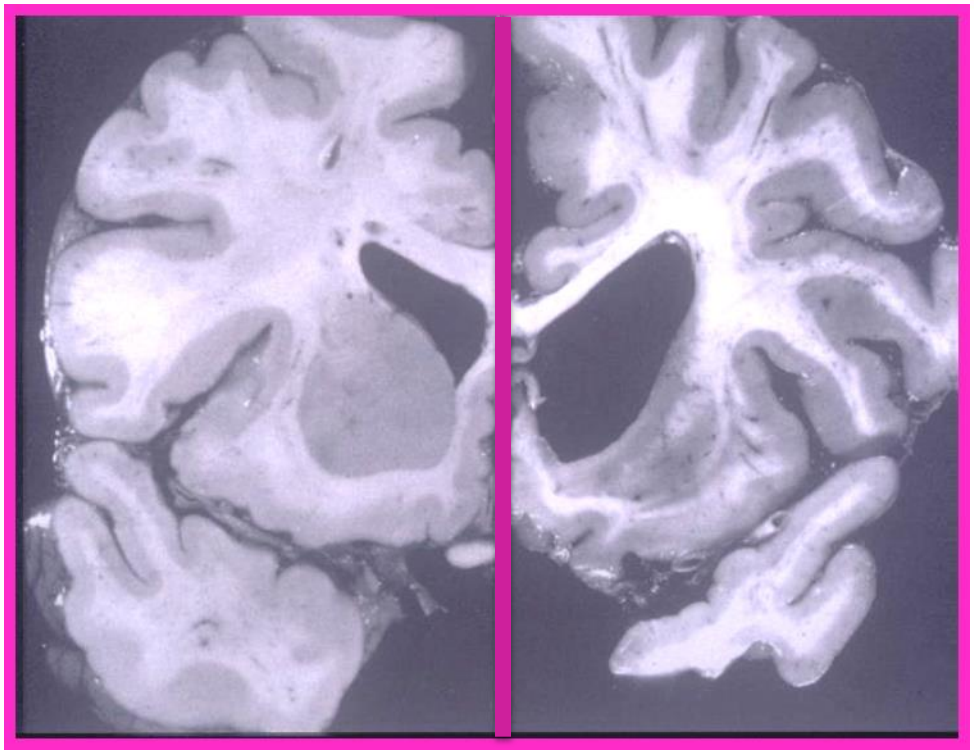
Balance Problems

↓Creativity

Slow Eye Movements

Episodic Anger, Irritability

Slowness of Movement



Normal

Advanced HD

Chorea: Involuntary Movements

↓Multi-tasking

Restless, Fidgets

↓Organizing Concentrating Prioritizing

↓Fine Motor Tasks

Depression, Anxiety

Trouble Swallowing

OCD Psychosis

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

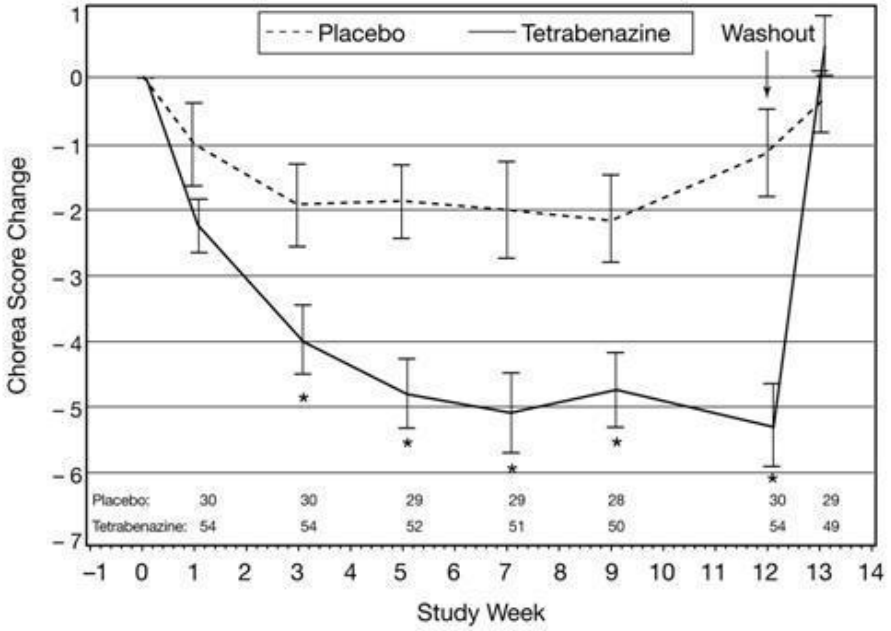
Interactions:

- Some anti-depressants
- Other anti-chorea drugs

Dosing:

- Genetic test
- Monitor EKG

Cost: Special program



Balance Problems

Rosy Chow,
Physical Therapist

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.

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