Juvenile Onset Huntington’s Disease

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Juvenile HD

- HD age of onset 10% < 20; 25% > 50
- Male transmission 70-80% JuvHD
- CAG > 60 considered most common; 46% < 60 (42, 45, ...) > 80 repeats rare (<5% JuvHD); largest CAG = 250
- Avg range before diagnosis 9y (0-21)
- 66% report psychiatric/behavioral 1st
- 80% have psychiatric disturbance
INTERESTING FAMILY

- Father: onset age 35 with 47 CAG repeats
- Daughter 1: onset age 4 with 99 CAG repeats
- Daughter 2: onset age 12 with 47 CAG repeats
- Daughter 3: onset age 15 with 47 CAG repeats
- Daughter 1 fits classical definition of paternal anticipation, but daughters 2 + 3 do not (other genes?, childhood onset of adult HD?)
Large Differences in JHD

- Children:
  - 1. selective vulnerability in children different from adults
  - 2. arrest of certain brain regions differs with age.
  - 3. degeneration occurs in arrested brain regions.
  - 4. unlike adults, compensation of dysfunction is limited by immature brain.
Brain Development

4 years old

SENSATION
Areas responsible for sensations like touch are almost as developed as they ever will be.

VISION
The part of the brain governing vision has already matured.

(top view)
Motor presentation in Juv HD

- Family history positive/confirmatory for HD
- Stiffness of arms and legs: rigidity and dystonia
- Clumsiness of arms and legs
- Seizures
- Bradykinesia rather than chorea
HD PRESENTATIONS IN CHILDREN FROM CLINICAL CASES: Reynolds

- RIGID-WESTPHAL JHD VARIANT
- MYOCLONIC JHD VARIANT W CORTICAL ATROPHY
- DYSTONIA JHD VARIANT W CORTICAL ATROPHY
- ATAXIA JHD VARIANT W SEIZURES
- ATAXIA JHD VARIANT W HYPERACTIVITY/ADD
- ATAXIA JHD VARIANT W CORICAL ATROPHY
- DELUSIONAL/OCD JHD VARIANT
- CHILDHOOD ONSET ADULT PATTERN (JHD?)
- PRECLINICAL JHD (CAG’S > 60)
- PRECLINICAL ADULT HD (CAG’S < 60)
JHD SCALE

☐ 1. Body Language (0-3)
☐ 2. Eye Movements (0-3)
☐ 3. Sialorrhea (0-3)
☐ 4. Ataxia (0-3)
☐ 5. Dyskinesias (0-3)
☐ 6. Manual Incoordination (0-3)
☐ 7. Hallucinations (0-3)
☐ 8. Compulsive/Obsessive Behavior (0-3)
☐ 9. Fluctuations In Abnormal Behavior (0-3)
Cognitive symptoms

- Trouble learning new things
- Inattention, less concentration
- Difficulty starting or completing a task
- Difficulty doing things in a sequence
- Regression or losses can be seen in many areas previously attained; request evaluations of standardized test performances
Cognitive interventions

- Individualized Education Plan for school aged, IFSP for young children (both mandated by IDEA legislation)
- Ongoing adjustments to home and school environments to allow adaptation to brain changes and their consequences
- School-home partnership (or childcare)
Psychiatric and Behavioral Symptoms

- Depression is most common symptom in children
- Sad affect, apathy, withdrawal, tearfulness, changes in sleep and appetite
- Suicide risk needs to be assessed
- May manifest as irritability in teens
- May be bipolar or cyclothymic
Medications

- Antidepressants
  - Tricyclics
  - SSRI
  - SNRI
- Mood stabilizers
- Beta-andrenergics, sedatives, neuroleptics for aggression
- Antipsychotics
- Anticonvulsants for seizures
- Treatments for chorea
- Treatments for spasticity/rigidity/dystonia
Non-medicine treatments

- Physical/Occupational therapy
- Speech therapy
- Assistive devices
- Safety evaluation
- Counseling
- Behavior management
- Independent educational plan
- Alternative therapies
- Professional support...hygiene coach
- Respite care
Efforts to enrich and normalize…

- racing
- skating
- dancing
- painting
Primary Principles: Behavior Management in Children

- Consider time perception and age
- Pick your battles
- Planning and Routine
- Intentional and Unintentional reinforcement
- Follow-through
Behavior Management Strategies

- ABC: Antecedent, Behavior, Consequences
- Parent Effectiveness Training
- Collaborative Problem-Solving
- Russell Barkley, Ross Greene
Prevention of behavior through anticipation

- Routines
- Clear structure and rituals
- Use cues via timer or voice prior to transitions
- Offer controlled choices
- Reward positive choices
- Identify common and recurring triggers
- Focus on major and important areas of conflict
Factors affecting behavior

- Frustration
  - Task complexity
  - Organizational need
  - Restraint needed for setting
  - Communication or other limitations

- Competition
  - Stimulation level in environment
  - Evident or presumed alternatives

- Fatigue
  - Time of day
  - Wellness
  - Tiredness
Anger

“Anyone can become angry, that is easy; but to be angry with the right person, to the right degree, at the right time, for the right purpose, and in the right way...this is not easy.”

Aristotle
Outbursts, temper...

- Identify the antecedent or “trigger”
  - Bath example

- Choose your basket
  - Basket A – safety
  - Basket B – priority
  - Basket C – what or how does it really matter?

- Acknowledge FEELINGS
- Assure safe environment(s)
- Check factors affecting behavior (FAB)
“Gates” in the Brain
What Happens When Gates Fall Out of Repair?

- Poor control of temper
- Poor control of recall
- Dysregulation of emotion and thinking
- Confusion
Inhibition: Being able to choose behavior

- Disease produces disinhibition of impulses
- Sexualized behavior can be poorly filtered
- Violence and aggression, either verbal or physical
- May lead to legal difficulties
- Can also lead to substance use in adolescents
Filtering, organizing, prioritizing

"Would you please pick up milk, bread, and eggs at the store?"

"Get milk, eggs..."

"I have a bad headache."

"Where's my wallet?"

"Today on Capitol Hill..."
Addressing attention and “on-task” behavior

- Establish routine
- Baby steps
- Modify requests to parts that can be attained
- Celebrate accomplishments
- Reinforce positively – go for it!
Obsessiveness

- May be very slow with beginning tasks or completing things due to rituals or compulsions
- Perseveration in thoughts, words or action
- May become very angry when disrupted or asked to deviate from the usual pattern
Managing Obsessions

- Allow child to talk about interests
- Be creative integrating interests into school and home activities
- Gently shift to new topic or activity
- Recognize as symptom, nor purposeful or willful
- Use prompts frequently to cue shift in activity or topic
- Agree to return to desired topic after “x”
Sexuality

- Changes of puberty may require acquisition of new personal care and social skills
- Inability to plan and direct attention
- May be exhibited in inappropriate settings, i.e. public masturbation
- May result in promiscuity or sexual abuse
- Can become victims or perpetrators of sexual assault
Team Approach

- Social services
- Financial planning
- School programs
- Physical supports
- Medical symptom treatments
- Nutrition/exercise schedules
- Behavior Management Skills
- Respite and Support for all Well-Being
Maximizing wellness and slowed progression

- Enriched environment
- Low stress
- Nutrition
- Exercise
- Wellness of caregiver
- Wellness of family
The rest of the family

☐ Take breaks
☐ Tag-team
☐ Allow change and flexibility
☐ Breathe
☐ Forgive yourself and others
☐ Each day is a new day