

# Juvenile Onset Huntington's Disease

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# Juvenile HD

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- ❑ 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
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# INTERESTING FAMILY

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- ❑ 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?)
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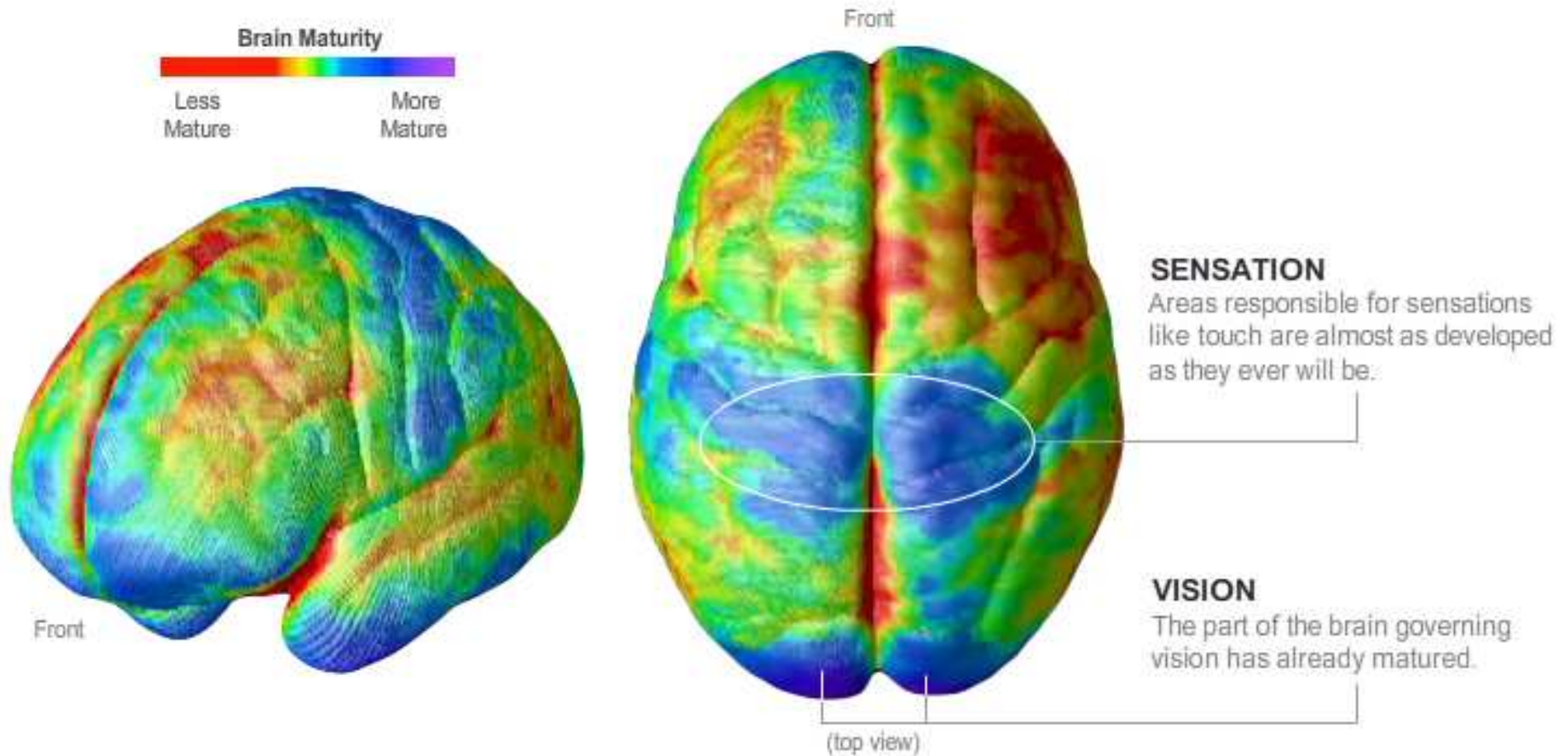
# Large Differences in JHD

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## □ 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.
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# Brain Development



4 years old

# **Motor presentation in Juv HD**

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

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- 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)
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# JHD SCALE

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- 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)
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# Cognitive symptoms

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- 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
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# Cognitive interventions

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- ❑ 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 child care)
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# Psychiatric and Behavioral Symptoms

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- 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
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# Medications

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



# Non-medicine treatments

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- 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...

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racing



skating



dancing



painting

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# Primary Principles: Behavior Management in Children

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

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

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- 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
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# Factors affecting behavior

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- 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

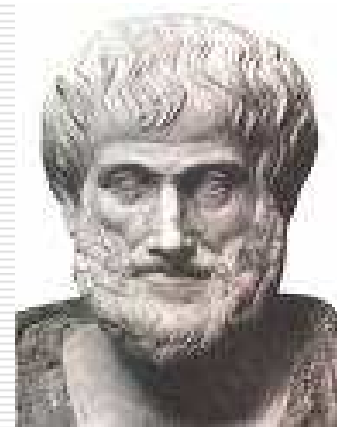


# Anger

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“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...

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- 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?



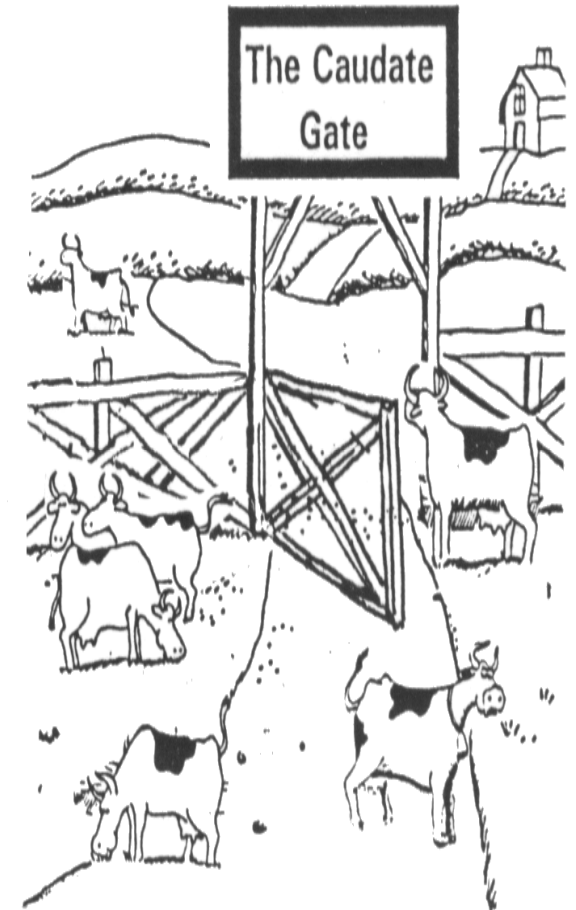
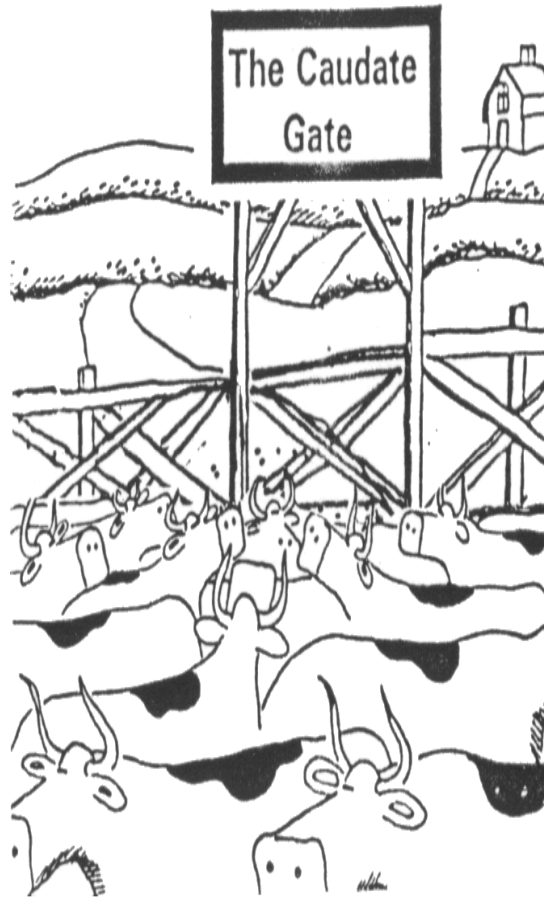
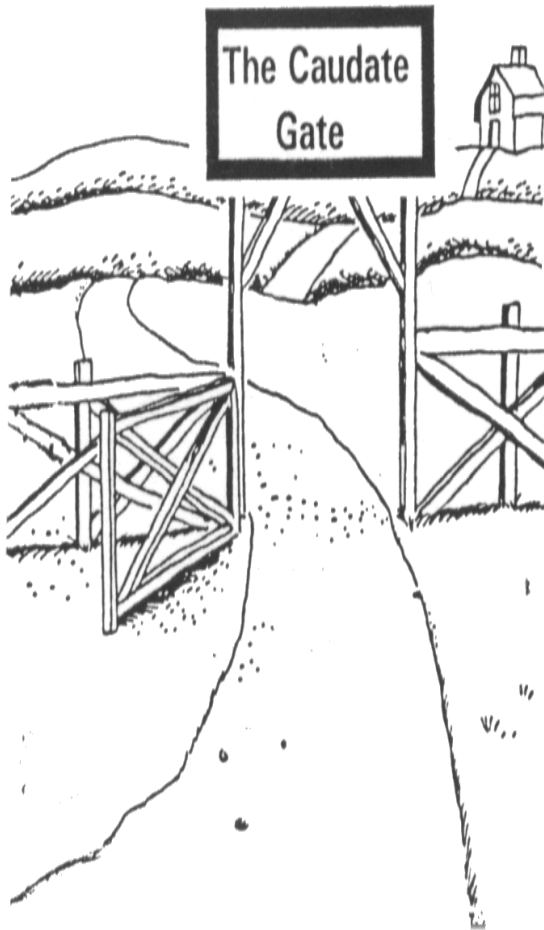
- Acknowledge FEELINGS

- Assure safe environment(s)

- Check factors affecting behavior (FAB)

# "Gates" in the Brain

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# What Happens When Gates Fall Out of Repair?

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- Poor control of temper
- Poor control of recall
- Dysregulation of emotion and thinking
- Confusion



# Inhibition: Being able to choose behavior

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- 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
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# Filtering, organizing, prioritizing

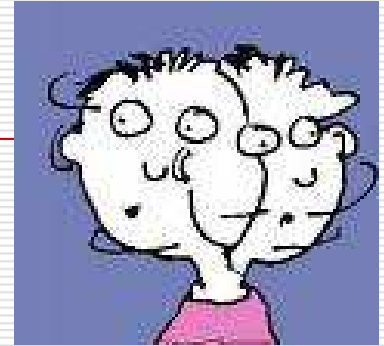
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# Addressing attention and “on-task” behavior

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

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- ❑ 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
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# Managing Obsessions

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- ❑ 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, not purposeful or willful
  - ❑ Use prompts frequently to cue shift in activity or topic
  - ❑ Agree to return to desired topic after “x”
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# Sexuality

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- ❑ 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

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

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

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