What is Juvenile Huntington’s Disease?

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Summary:

We will review what is meant by “Juvenile” onset HD and how it can be similar and also how it is different from the adult form.

We will discuss symptom presentation and the difficulties associated with making a clinical diagnosis of juvenile onset HD.

We will briefly review the pros and cons of genetic testing of children.

Finally we will consider treatment strategies with a focus on the special issues related the dynamics of growing up with a diagnosis of HD.
Huntington’s disease is a neurodegenerative disorder that affects multiple aspects of how a person functions.

Symptoms can present at any age though symptoms most commonly present at age 35-55.

In a small percentage of cases symptoms start before age 20.
Juvenile Onset HD

Usually a family history of relatively young onset HD.

Often inherited from the father. (~80 %)

CAG repeat size usually (but not always) >60
HD is a “trinucleotide repeat” disorder

DNA code:

A (adenine)
C (cytosine)
G (guanine)
T (thymine)

Codon = groups of 3

HD is caused by an increased number of CAG repeats
Normally we have less than 10-26 CAG repeats


In HD there are more than 36 CAG repeats in a row
There is a **rough** correlation between repeat size and age of onset.

The higher the repeat size the early the disease onset.

Only 5-10% of individuals have repeat sizes greater than 50 or 60.

Symptoms in these individuals often start before age 20.
HD basics

• The disease starts insidiously and progresses continuously.

• Symptoms include changes in:
  – Behavior
  – Motor function
  – Thinking ability
HD basics

In adults motor symptoms include:

Chorea
Motor impersistence
Oculomotor changes
Impaired fine motor control
Balance and gait disorder
Dystonia
Speech/swallowing problems
Juvenile HD symptoms

- Stiffness of legs. Scissoring gait.
- Walking on toes.
- Clumsiness of arms and legs.
- Decline of mental ability and milestones.
- Changes in behavior.
- Seizures (~ 25% of children with HD).
- Swallowing and speech difficulties.
- Bradykinesia.
- Cerebellar signs.
- Chorea is less common in children but may be seen in teens.
Behavioral changes

• “A common early symptoms of juvenile HD is difficulty starting or finishing a task or doing all the steps of a task in the right order. As these can also be common symptoms of ordinary adolescence, parents teachers and physicians must look beyond single symptoms for a pattern of change that suggest HD.”

• – From Family guide series on Juvenile HD.
Behavioral issues

- Aggression
- Impulsiveness
- Adolescence and sexuality
- Depression
- Obsessive compulsive behavior
- Hallucinations (rare)
Juvenile HD

• Seizures are more common in children with HD than in adults

• Seizures seem to be more common in children who present with symptoms before age 10

• A neurologist can help prescribe anti-seizure medications
Genetic testing

• Usually for children who clearly have clinical symptoms and progression of symptoms.

• Genetic counseling before and after testing.

• Ensure that testing is in the best interest of the child.
Juvenile Onset HD
Why is it difficult to make a clinical diagnosis of JHD?

• The disease presents differently in each child.
• JDH is a rare form of a rare disorder.
• All children develop at different rates.
• Many children without HD or with other disorders may have symptoms that look like HD.
  – ADHD, depression, juvenile forms of bipolar disorder, mild cerebral palsy, seizure disorders, thyroid disease, etc.
Dangers of premature testing

- Incorrectly blaming symptoms on HD.
- Discrimination (insurance, future employment, others).
- Negative psychological and social effects.
- Testing of other children in the family is not encouraged.

- 44 subjects
- Of the 15 presenting under age 10, 12 had CAG repeats greater than 80.
- Symptoms included declining school performance, seizures, oral motor dysfunction, rigidity and gait disorder.
- Presenting symptoms in those over age 10 were more varied but usually included behavioral and motor symptoms.
A functional scale for assessing juvenile-onset HD

• A. School attendance.
• B. Academic/developmental performance.
• C. Chores.
• D. Activities of daily living.
• E. Residence.
A. School attendance

3- attends school, no special needs.
2- attends school, some regular classes, some modified classes.
1- attends school, few or no regular classes.
0- unable to attend school.

B. Academic/developmental performance

3- Reading/writing/math skills appropriate to age.
2- mild decrease in academic performance but still able to take a test or write.
1- unable to write legibly but able to communicate orally.
0- unable to read/write/communicate orally.
C. Chores.
- 2- Able to assist in age-appropriate manner with household chores.
- 1- Occasionally assists with chores.
- 0- Unable to participate in household chores.

D. Activities of daily living.
- 3- Performs self-cares in an age appropriate manner.
- 2- Requires some assistance for bathing dressing, grooming, or feeding.
- 1- assists others who bathe, dress, or feed him/her
- 0- unable to assist in self care.
E. Residence.

- 2. At home with only family assistance
- 1. At home/group home/foster care with assistance from non-family members.
- 0. Living in a skilled nursing care facility.

Add points to determine stage of HD

- Stage 1 .......... 11-13 points
- Stage 2 .......... 7-10 points
- Stage 3 .......... 3-6 points
- Stage 4 .......... 1-2 points
- Stage 5 .......... 0 points
Treatment: This is a progressive disorder. Treatment plan needs to be regularly reviewed.

• Symptomatic treatment:
  – Spasticity/rigidity. PT/OT, benzodiazepines, baclofen, dantrolene, tizanidine.
  – Speech swallowing and dental care issues.
  – Seizures: multiple medication options
  – Chorea. Benzodiazepines, amantadine, neuroleptics, tetrabenazine
The Juvenile HD treatment team

- Family and community
- pediatrician
- pediatric neurologist
- pediatric physiatrist
- physical therapist
- occupational therapist
- orthopedic surgeon
- orthotist
- speech/language pathologist
- home care nurse
- psychologist
- psychiatrist
- social worker
- educator/schools (individual education plans)
JHD educational resources
Juvenile Huntington's Disease

Martha Nance, MD, 2007. This book provides readers with information about Juvenile HD, including genetic testing, when it is appropriate to perform the test, how to understand your HD affected child, and what to do if you find yourself in compromising situations as well as where to turn for help.
Down load or order hard copy $7.00 each

Juvenile HD: A Guide for Families
Edited by Martha Nance, MD, Randi Jones PhD, Suzanne Imbriglio PT & Betsy Gettig MS/CGC. This pamphlet defines Juvenile HD, diagnosis, specific medical issues, prognosis of the disease, financial, legal & social issues and more.
Down load or order hard copy $2.00 each

The Juvenile Onset HD Resource Directory
Comprehensive resource guide for families of children affected by Juvenile Onset HD. Information about federal, state and local programs.
Down load or order hard copy $10.00 each
Pamphlet for families can be downloaded from Huntington's Disease Society of America Web site (www.hdsa.org)
HDSA PUBLICATIONS (con’t)

**JHD SPECIFIC**
The *Family Guide Series to Juvenile HD* by Martha Nance, MD (copyright 2009)
The *Juvenile HD Handbook* by Martha Nance, MD (copyright 2009)
*School and the Juvenile HD Experience* (copyright)
The *JHD Directory* (copyright 2009)

HDSA Webinars (archived on [www.hdsa.org](http://www.hdsa.org))

**GENERAL**
Palliative and End of Life Issues November 17, 2010
HD and Nutrition January 26, 2011
Team Management of HD February 22, 2011
Communication through Use of September 26, 2011
Assistive Technology

**JHD SPECIFIC**
Juvenile HD February 24, 2012
HDYO

- Huntington's Disease Youth Organization
- hdyo.org
- HDYO! A site created for young people to learn about HD in an age-appropriate, friendly and factually correct manner.
Resources Available through the Huntington Society of Canada
www.huntingtonsociety.ca

PUBLICATIONS (available in print and on line)
JHD: A resource for Family, Health Providers and Caregivers (copyright 2008)

FACT SHEETS (available on line)

GENERAL
Diet and HD June 2010
Palliative Care June 2010
End of Life June 2010
Strategies to Enhance Care August 2007
Other Websites with JHD Resources

**Stanford HOPES** (Huntington’s Outreach Project for Education at Stanford) – JHD section created 2010 [www.stanford.edu/group/hopes/cgi-bin/wordpress/category/hd-basics/juvenile-hd/](http://www.stanford.edu/group/hopes/cgi-bin/wordpress/category/hd-basics/juvenile-hd/)


**MDVU** (Movement Disorders Virtual University) [www.mdvu.org/](http://www.mdvu.org/) (primarily for health care providers)

**Patient Power**, JHD interview with Martha Nance and Sandra Kostyk, 2008 [www.patientpower.info/program/juvenile-huntingtons-disease](http://www.patientpower.info/program/juvenile-huntingtons-disease)

**HD Lighthouse** ([www.hdlf.org/jhd](http://www.hdlf.org/jhd))
International JHD Resources

PUBLICATIONS

*Juvenile Huntington's Disease* (and other trinucleotide repeat disorders), Quarrell, Brewer, Squitieri, Barker, Nance, Landwehrmeyer, 2009 (Amazon.com Price:$110.00) Mostly geared towards professionals.

WEBSITES

EHDN – [www.euro-hd.net](http://www.euro-hd.net)

JHD section [www.euro-hd.net/html/network/groups/jhd](http://www.euro-hd.net/html/network/groups/jhd)
International JHD Resources

HDA (UK): [http://hda.org.uk](http://hda.org.uk)

Information for Schools – also cites HDSA and HSC resources

Challenging Behavior – Huntington’s Disease in Children and Teenagers – A Guide for Professionals, Glendinning

Downloadable fact sheets:  [http://hda.org.uk/hda/factsheets](http://hda.org.uk/hda/factsheets)

Challenging Behavior in Juvenile HD

A Young Person with JHD at School

A Brief Guide to Juvenile HD for Children’s Hospices and Palliative Care Services
International JHD Resources

Scottish HD Association (www.hdscotland.org/youth)
JHD Section divided by audience
This site is mostly geared to helping those in the following groups deal with having a family member with HD
  Children Ages 8-12
  Teens Ages 13-17
  Young Adults Ages 18-25
  Professionals (very limited info for health care providers.)

Australia (www.ourhdspace.org)
Website dedicated to helping young people affected by HD
Other useful sites.

• National Information Center for Children and Youth with Disabilities
  
  http://nichcy.org/

Example of downloadable documents
  * Adaptions and accommodations for students with disabilities
  * Simple and inexpensive communication devices
  * Sexuality Education for children and youth with disabilities.
RESEARCH: Our hope for the future