Huntington's Disease Society of America

Juvenile-onset Huntington's disease

Presenter Disclosures

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The following personal financial relationships with commercial interests relevant to this presentation existed during the past 12 months:

Lundbeck Advisory Board

Juvenile onset Huntington's disease (JHD)

- Definition and demographics
- Clinical differences between JHD and Adult-onset HD
- Diagnosis and management of JHD
- References

What is JHD?

- Onset of HD in childhood (<18, <20, <21)
- About 5-10% of HD cases
- About 90% have an affected father
- Partially explained by larger CAG repeat numbers in the huntingtin gene (often >60)
  - Bigger increases in CAG repeat number when the gene is passed on by a male (in general)

Clinical features of JHD

	Juvenile-onset HD	Adult-onset HD
Onset age	Under 20	20 and over
Movement disorder	Rigidity, dystonia, oral-motor dysfunction	Chorea, incoordination
Cognitive disorder	Failing school performance	Executive dysfunction
Psychiatric/ behavioral disorder	Wide range and variety; (severe) antisocial behavior sometimes seen	Wide range and variety
Other features	Seizures (25%)	Weight loss
Course	?10-15 years	15-20 years

Diagnosis of JHD

- Recognize a history of cognitive, motor decline
  - Declining academic performance
  - Declining athletic skills
  - Failing to achieve developmental milestones

Diagnosis of JHD

- Possible history of behavioral/ psychological/ psychiatric symptoms
  - Depression, suicidality, obsessive-compulsive
  - Antisocial (sometimes severe) behavior
  - Substance abuse
  - Inappropriate sexual behavior
  - (?attention deficit)

Diagnosis of JHD

- Family history of HD
- No family history of HD
  - At-risk parent (child diagnosed before parent)
  - nonpaternity
  - adoption

Diagnosis of JHD

- Motor examination
  - Abnormal eye movements
  - Dysarthria, oral-motor dysfunction (eg drooling)
  - Often increased muscle tone/rigidity
  - May have hyperreflexia
  - Abnormal gait and balance; (eg toe-walking, poor tandem, unable to stand on one foot)
- Cognitive examination
  - Age-inappropriate, or decline on formal testing

Diagnosis of JHD

- MRI may show caudate atrophy, diffuse atrophy, or cerebellar atrophy (in a very young child)
- Neuropsychological testing shows frontal/prefrontal dysfunction, declining performance over serial tests

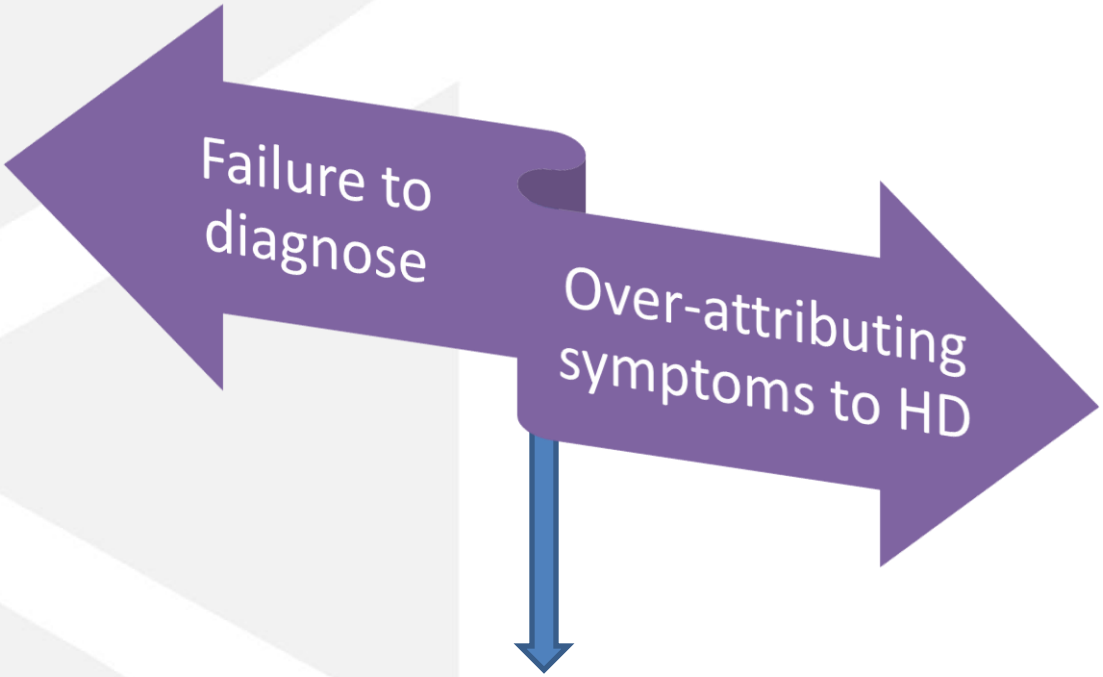
Diagnosis of JHD

- CAG repeat analysis of the huntingtin gene shows expanded CAG repeat sequence
  - >50 CAG repeats?

Challenges

- Children with developmental delay/ADD
- Missing/absent family history
- Affected mother
- Psychiatric-only presentation
- “soft signs”-only on examination
- CAG repeat numbers in the 30-45 range
- False-negative gene test result (PCR-based test may miss very large CAG repeats in very young children

Challenges


Failure to diagnose
Over-attributing symptoms to HD
MIDDLE GROUND

The middle ground

- Evaluate the child
- Brain imaging, any appropriate general labwork
- Formal cognitive assessment
- Formal psychiatric/psychological evaluation and treatment
- Optimize the family/school situation
- Re-evaluate in 6-12 months
- Consider gene test at that time (if declining)

Management of JHD

Health professional	Role in JHD management
Neurologist	Team leader, movement disorder, seizures, initiate referrals
Psychiatrist	Psychiatric/behavioral symptoms
Psychologist	Behavioral symptoms; family counseling
Neuropsychologist	Cognitive assessment and recommendations
Speech therapy	Assess, treat dysphagia, communication problems
Physical therapy	Gait disorder, liaise with school on physical education recommendations
Occupational therapy	Safety, functional assessment, equipment
Dietitian	Healthy eating; altered food textures; high calorie foods
Social worker	Identify community resources; assist with legal, financial issues

Management of JHD

Type of professional	Role in JHD management
Pediatrician	General, age-appropriate care
Dentist	Age-appropriate care
Teacher, school officials	Independent education program
Lay organization	Support for patient and family

Disability

- On April 11, 2012, the Social Security Administration announced a “compassionate allowance” for SSI/SSDI for children with JHD
- Goes into effect August 2012
- Thanks to Jane Kogan, Sam Frank MD and HDSA for negotiating with the SS Admin

References

- Nance MA, *The Juvenile HD Handbook*. New York, HDSA, 2007.
- Aubeeluck A, Brewer H. Br J Nurs 2008; 135: 260-263.
- Nance MA, Myers RH. Ment Retard Dev Disabil Res Rev 2001; 7: 153-157.