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

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

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

<table>
<thead>
<tr>
<th>Type of professional</th>
<th>Role in JHD management</th>
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<tbody>
<tr>
<td>Pediatrician</td>
<td>General, age-appropriate care</td>
</tr>
<tr>
<td>Dentist</td>
<td>Age-appropriate care</td>
</tr>
<tr>
<td>Teacher, school officials</td>
<td>Independent education program</td>
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<tr>
<td>Lay organization</td>
<td>Support for patient and family</td>
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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

