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HDSA encourages all attendees to consult with their primary care provider, neurologist or other healthcare provider about any advice, exercise, medication, treatment, nutritional supplement or regimen that may have been mentioned as part of any presentation.
Presenter Disclosures

Kathleen Collins

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

No relationships to disclose or list
Getting to know each other

• Considering testing for HD?
• Had testing for HD?
• Received a positive HD result?
• Has symptoms of HD?
• Has a family member with a positive HD result?
• Provides care/support for someone with HD?
Objectives

• Briefly review HD testing recommendations – testing process and results
• Discuss unexpected/bad news and possible responses/reactions
• Discuss coping mechanisms and possible responses to HD results
• Hear from audience about their experiences (if applicable)
HD Genetic testing

- International Huntington Association (IHA) and the World Federation of Neurology (WFN) proposed guidelines for presymptomatic testing in 1994
- Guidelines evaluated and updated by European Huntington Disease Network (EHDN) “Genetic Testing and Counseling” Working Group (published in 2013)
Presymptomatic Testing

- Recommendations to reduce likelihood of an adverse event
- Very few adverse events occur
The Process of HD Testing

• Confirmation of a diagnosis in the family
  – Test an affected relative if possible
• Education and Counseling
  – Extensive discussion about implications of testing with a genetic counselor or geneticist
  – Neurological exam
  – Possibly evaluation by a psychologist or psychiatrist
• Support
  – The process of testing can be stressful
  – Even “good news” can take adjustment
  – It can be helpful to have a support person attend all appointments
Testing

• Very personal decision
  – Many studies show fewer than 10-20% of individuals at-risk choose to have testing
• Opinions may vary greatly within the same family
• Benefit of presymptomatic testing mainly psychological
  – Must be a personal decision with no coercion from others
The Decision to be Tested

- What do you think the test results will be?
- Who will you share the results with?
- How and when will you share the results?
- What will you do the day of the results disclosure?
- Have you thought about/arranged insurance such as life insurance?
# Possible Test Results

<table>
<thead>
<tr>
<th>Number of CAG Repeats</th>
<th>Expected Features</th>
</tr>
</thead>
<tbody>
<tr>
<td>26 or less</td>
<td>Negative – not at risk of HD or gene expansion</td>
</tr>
<tr>
<td>27 - 35</td>
<td>Negative – not at risk of HD; however gene expansion may occur in future generations</td>
</tr>
<tr>
<td>36 - 39</td>
<td>Uncertain – disease causing but reduced penetrance (may have later age of onset, may not show symptoms)</td>
</tr>
<tr>
<td>40 or more</td>
<td>Positive – full penetrance (symptoms of HD will occur at some time)</td>
</tr>
<tr>
<td>55 – 60 or more</td>
<td>Positive – juvenile HD (symptoms occurring before the age of 20 – accounts for 5-10% of all HD)</td>
</tr>
</tbody>
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Unexpected/Bad News

• Defining “Bad News”:
  – “Information likely to drastically alter a patient’s view of his or her future” – Buckman, 1992
  – “Something that can change a person’s understanding of themselves or a loved one as well as their future” – Uhlmann, pg. 148
Unexpected/Bad News

Think about a time YOU received bad news. Take 5 minutes and write down/think about answers to the following questions.

– What was the “bad news”?
– Who told you/how did you find out?
– What details do you remember?
– What were your initial feelings?
– How do you feel about the situation now? How long has it been?
Potential Reactions to Unexpected News

- Denial/disbelief/dismissal
- Shock
- Fear
- Loss
- Anxiety
- Sadness
- Despair
- Powerlessness/loss of control
- Anger – toward the situation, you, doctor, etc
- Guilt
- Shame
- Grief
What is Grief?

• Deep and poignant distress caused by loss or impression of loss
• Emotional, physical and psychological reactions to loss
• Process of finding “new normal”
  – Weil, 2000, pg. 17
  – Klass, 1997
Grief Triggers

• Reaction to the loss of
  – Person
  – Current capabilities
  – Future capabilities
  – Control
  – Expected outcomes or dreams
• Previous losses
Expressions of Grief

• Feelings
  – Sadness, anger, guilt, anxiety, loneliness, fatigue, helplessness, shock, relief, numbness

• Thoughts
  – Disbelief, confusion, preoccupation

• Physical sensations
  – Hollowness in the stomach, tightness in the chest, tightness in the throat, over-sensitivity to noise, feeling short of breath, weakness in muscles, lack of energy, dry mouth
Grief Behaviors

- Sleep disturbances
- Appetite disturbances
- Absent-mindedness
- Social withdrawal
- Sighing
- Restless over-activity
- Crying
Grief Process

- **Avoiding** the new reality
  - Shock, denial
- **Admitting** the new reality
  - Anger, guilt
  - Realistic view of loss
- **Adapting** to the new reality
  - Acceptance
  - Readjustment
  - Personal growth
Signs of Adaption to Grief

• Return to day-to-day activity
• Emotional stability
• Joy and enthusiasm for life
• Involvement in established relationships

• Not the end of grief
Grief Rules

• There is no right way to grieve
• There is no right time to be finished grieving
• Grieving occurs in complex patterns
• A person can reach a place where they have adapted to the loss and redefined their life
Things People Say...

- **Bad responses**
  - Everything will be fine
  - It’s all going to work out
  - It’s been long enough. Aren’t you ever going to get back to normal?
  - I know how much you are hurting
  - You’ll get over it
  - Other examples?

- **Good responses**
  - I’m really sorry this has happened to you
  - This must be really difficult
  - What can I do for you?
  - It is very normal to feel _________ when dealing with a loss
  - Healing after a loss is a very individual process. There is no set time limit.
  - Other examples?
Assess Coping Strategies

• How have you dealt with difficult situations in the past?
  – Short term?
  – Long term?
• How do you ask for help when you need it?
Coping Strategies

• May use several coping strategies
• Consider whether or not the response is adaptive/maladaptive
Coping Strategies

• Confronting
  – Trying to change the opinion of the person in charge
  – “You must be wrong. Are you sure those are MY test results?”
• Distancing
  – Going on as if nothing happened
  – “I wonder how Steve will feel about our plans this weekend”
• Self-controlling
  – Keeping feelings to oneself
  – “What other information do you have for me?”
More Strategies

• Seeking social support
  – Engaging in conversation in the hope of learning more
  – “Tell me more about the medical complications of HD”

• Accepting responsibility
  – Criticizing oneself
  – “I knew something was wrong the whole time. I just knew I shouldn’t have XYZ when I was younger.”
Even More Strategies...

- Escape-avoidance
  - Hoping for a miracle
  - “Maybe they will find a cure for HD next week. Research has come a long way.”
- Plan
  - Identifying and following an action plan
  - “It sounds like the best next steps are…”
- Positive reappraisal
  - Identifying existing or potential positive outcomes
  - “This isn’t what I was hoping for but it doesn’t sound as bad as when my father was first diagnosed with HD. I think I can handle this.”
Other Factors that may affect Coping

• Personality characteristics – ego strength
• Reasons for testing
• Expectations of test result
• Social support
• Timing
Positive test result: no symptoms

No previous molecular testing in family. 4 older siblings tested negative.
Positive result: no symptoms

• Isolation
• Concern about when they will develop symptoms
  – Difficult to be objective about occasional clumsiness, jerk or emotional outburst
Positive result: symptomatic

- d. 42 years HD
- d. 44 yrs HD Symptoms early 30s
- 30 years Balance problems Speech problems
Positive Test Result: Symptomatic

- May feel a sense of relief to have an explanation
- Reduce stress in the work environment – may provide for job reclassification or benefits
Advantages of Testing (if positive)

- Uncertainty of gene status removed
- Prepare for the future
- Arrange surveillance/treatment
- Inform children/family planning
Disadvantages of Testing (if positive)

• Removes hope
• Introduces uncertainty (if and when)
• Known risk to offspring
• Impact on self/partner/family/friends
• Potential problems with insurance/employment
Impact of Test Results

• Results usually evoke strong emotional feelings REGARDLESS OF OUTCOME
• “loss of identity” if result is different from what was expected
• Regretting past decisions
  – Family planning
  – Career paths
• Most people eventually adjust well to their test results
• Important to draw on support of professionals, family and friends
Advice from those who have tested positive

• It takes TIME to adjust to the result
• Normal to be angry, sad, depressed, etc
• Talking to someone who understands HD or has gone through the process can help
• Just because you test positive for the gene does not mean you automatically have symptoms
Coping with a Positive Test Result – Main Points

• Intense/unplanned feelings may occur
• Support is important – professional, friends, family
• REMEMBER – Most people eventually adjust to their result
Additional Advice/Thoughts?
Resources

• Huntington Disease Society of America (HDSA) – www.HDSA.org
• National Society of Genetic Counselors – www.nsgc.org
• Huntington’s Disease Lighthouse Families – www.hdlf.org
• Huntington’s Disease Youth Organization – en.hdyo.org
References

• Worden, J.W. Grief Counseling and grief therapy: A handbook for the mental health practitioner. 1982. Springer Publishing Company
References

Thanks!