



Coping with a Positive Test Result

Kathleen Collins, LGC

Genetic Counselor

Division of Human Genetics

Cincinnati Children's Hospital Medical Center



The information provided by speakers in workshops, forums, sharing/networking sessions and any other educational presentation made as part of the 2014 HDSA Convention program is for informational use only.

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

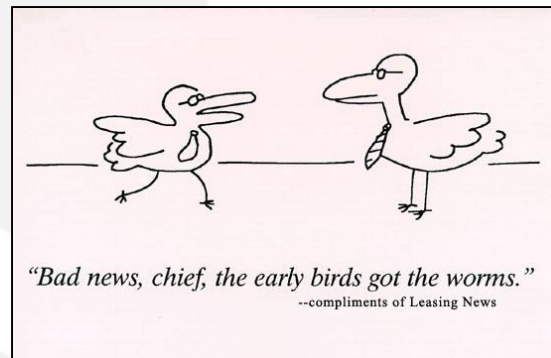
- What do you think the test results will be?
- Who will you share the results with?
- How and when will you share the results?
- What will you do the day of the results disclosure?
- Have you thought about/arranged insurance such as life insurance?

Possible Test Results

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

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.”

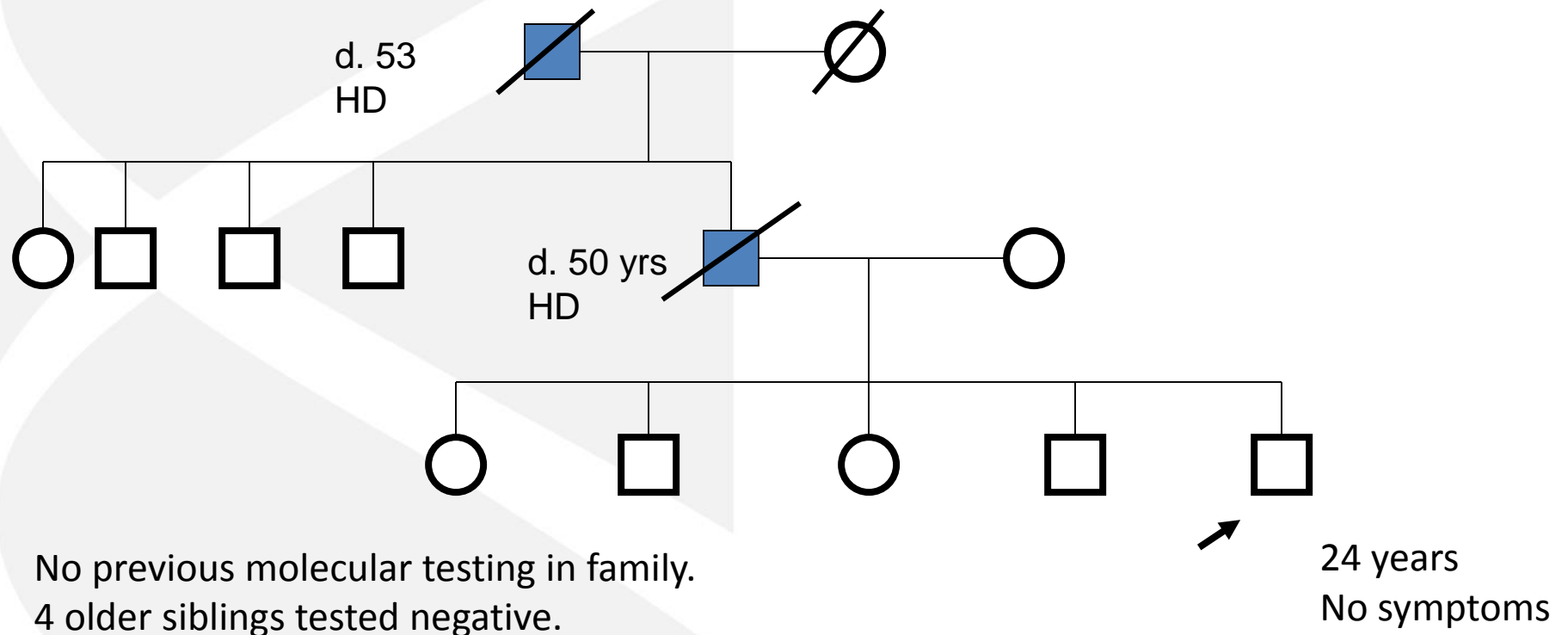
Even More Strategies...

- 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

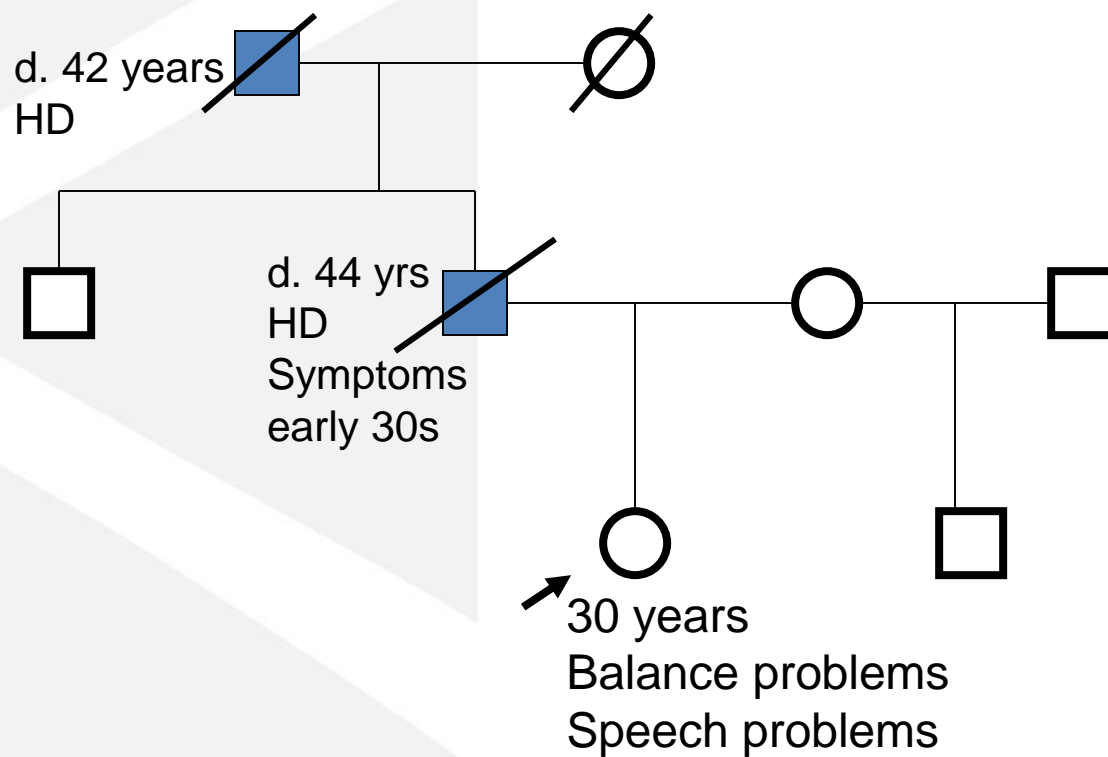
Positive test result: no symptoms



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



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

- Macleod, R. , et al. Recommendations for the predictive genetic test in Huntington's disease. *Clin Genet*. 2013; 83(3) 221-231
- Walker, O., et al. *Lancet* 2007; 369: 218-228
- American College of Medical Genetics/American Society of Human Genetics, Huntington Disease Genetic Testing Working Group, ACMG/ASHG statement: laboratory guidelines for Huntington disease genetic testing. *American Journal of Human Genetics* 1998; 62: 1243-1247
- Babul , R., et al. Attitudes toward direct predictive testing for the Huntington disease gene. *Journal of the American Medical Association* 1993; 270 (19): 2321-2325
- Weil, J. Psychosocial genetic counseling. 2000. Oxford University Press.
- Worden, J.W. Grief Counseling and grief therapy: A handbook for the mental health practitioner. 1982. Springer Publishing Company

References

- Uhlmann, W.R. Schuette, J.A. and Baker, D. (Eds). A Guide to Genetic Counseling, 2nd Edition. 2009. Wiley-Blackwell
- Buckman, Robert. Breaking bad news: the S-P-I-K-E-S strategy. *Commun Oncol* 2005; 2:138-142
- Decruyenaere, M., et al. Psychological distress in the 5-year period after predictive testing for Huntington's disease. *European Journal of Human Genetics* 2003; 11: 30-38

Thanks!