

The Genetic Testing Process Unraveled

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Presenter Disclosures

Katie Plunkett

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

No relationships to disclose
or list



The Presymptomatic Testing Process

- ▶ Who should consider genetic testing for HD?

The Presymptomatic Testing Process

- ▶ Who should consider genetic testing for HD?
 - People who have a parent with HD and are at a 50% risk to have HD
 - What if your parent does not have HD but a Grandparent does? Should you be tested?

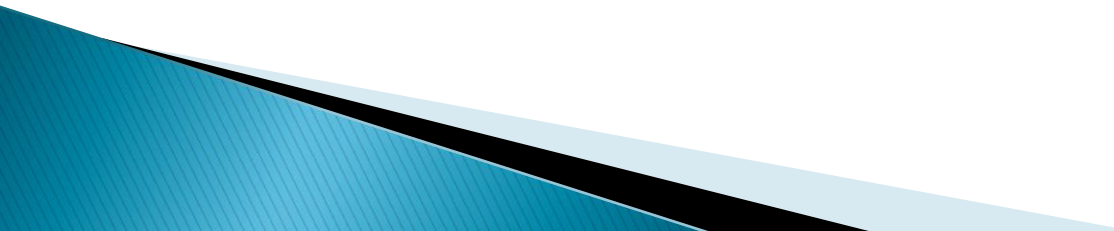
The Presymptomatic Testing Process

- ▶ At what age should someone have genetic testing for HD?

The Presymptomatic Testing Process

- ▶ At what age should someone have genetic testing for HD?
- ▶ Most programs will not test individuals under 18 years of age. Why do you think this is the case?

The Presymptomatic Testing Process

- ▶ At what age should someone have genetic testing for HD?
 - ▶ Most programs will not test individuals under 18 years of age
 - ▶ Want a person to be old enough to make their own decision about testing. Do not want a parent making that decision.
 - ▶ Legally an adult at age 18
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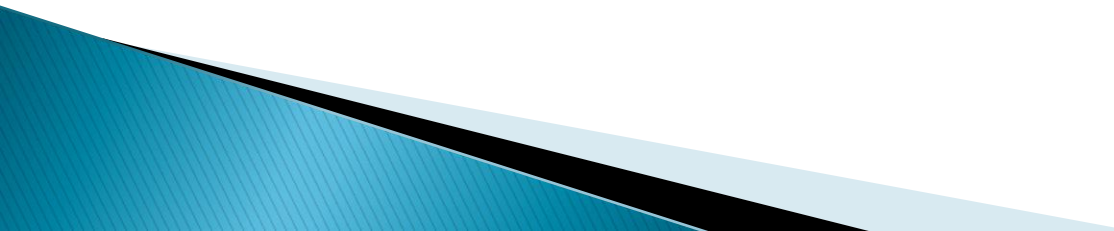
The Presymptomatic Testing Process

- ▶ Why might someone have testing for HD?

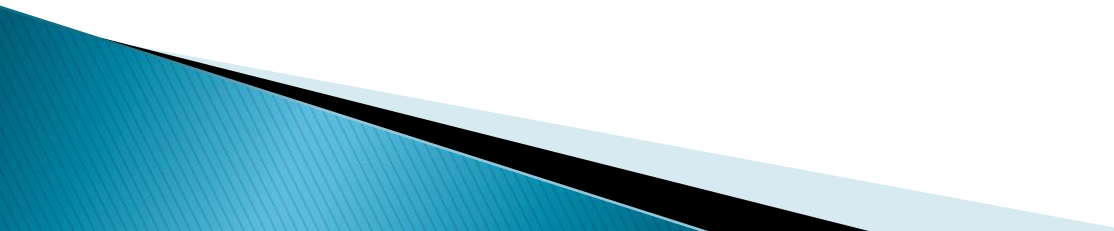
The Presymptomatic Testing Process

- ▶ Why might someone have testing for HD?
 - In a serious relationship and thinking of getting married
 - Thinking of having children
 - Has children and wants to know if they are at risk
 - Getting older and wants to know for planning
 - Thinks they are having symptoms

The Presymptomatic Testing Process

- ▶ Go to www.hdsa.org to find a genetic testing center
 - ▶ Each center has its own protocol but all involve pretest counseling
 - ▶ Different people are involved in the counseling including genetic counselors, doctors, psychologists, and nurses
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The Presymptomatic Testing Process

- ▶ Call a center and gather more information
 - How many visits?
 - Cost of testing – should you pay out-of-pocket or use insurance?
 - How long for results once the blood is drawn?
 - Do they have information they can send you?
 - Who can you bring with you?
 - Confidentiality – Who gets copies of the results?
 - Be ready to answer questions about why you want to be tested.
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The Presymptomatic Testing Process

- ▶ Where else can you get information about the testing process?

The Presymptomatic Testing Process

- ▶ Where else can you get information about the testing process?
 - Family
 - Friends
 - HDSA
 - Websites

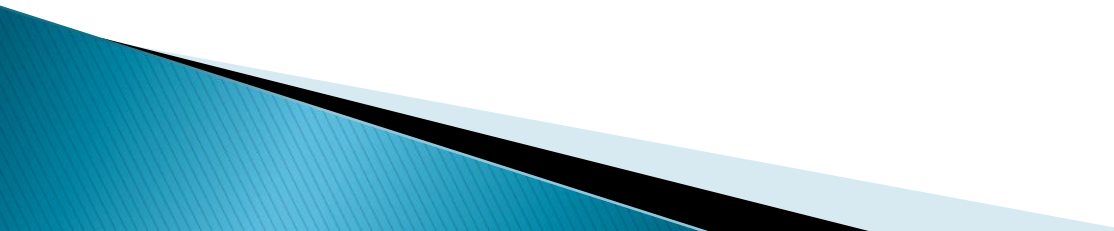
The Presymptomatic Testing Process

- ▶ The main question to ask yourself before being tested: Are you really ready to find out that you have the gene for HD?
 - No matter how much you prepare, it is still hard news to hear.
 - Do not have testing because “you know you don’t have HD.”
 - Is it better to know than not know?
 - Deciding not to be tested is a very real option.

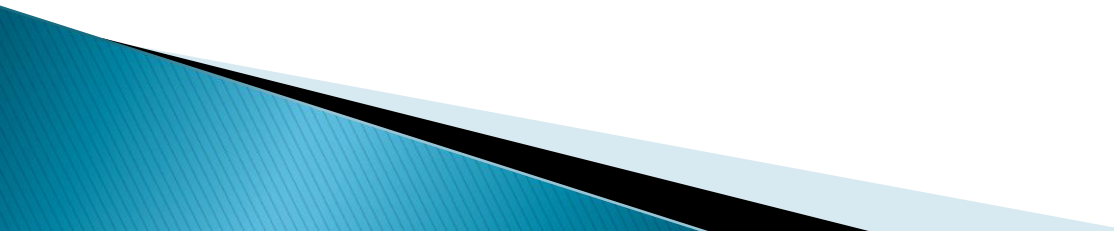
The Presymptomatic Testing Process

- ▶ The Baylor College of Medicine Protocol
 - Two initial appointments plus the blood draw
 - Meet with me (genetic counselor) to review the genetics, why doing testing – lasts about an hour
 - Meet with a Psychologist – talking plus some “tests”
 - Blood is drawn the same day
 - Results take about 3 weeks
 - Must come back for results in person
 - If person has the HD gene, refer to neurologist

The Results Visit

- ▶ Once I know results will be available, I call or email the person and schedule the results visit.
 - ▶ Let the person being tested decide when.
 - Typically take the entire day off
 - Often do not want it to be around a birthday or holiday
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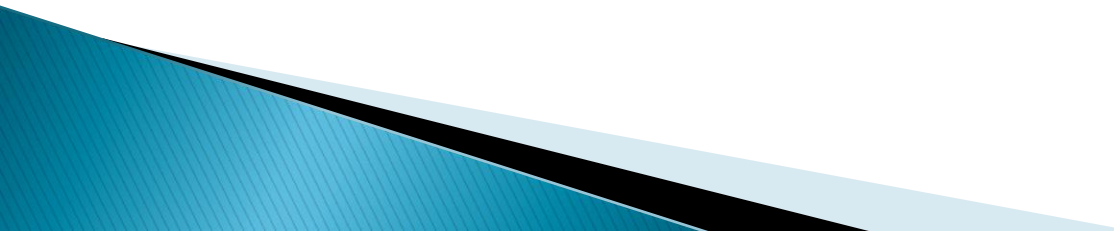
The Results Visit

- ▶ Immediately tell the person their results
 - ▶ Have a copy of the results for them to take
 - ▶ Take as much time as possible to answer any questions
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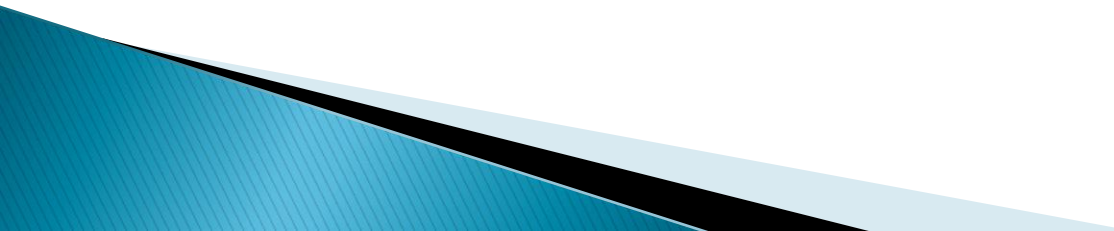
The Results Visit

- ▶ Person does not have HD
 - The absolute best part of my job!
 - Time for celebration
- ▶ What if the person is not happy? What might be some reasons?

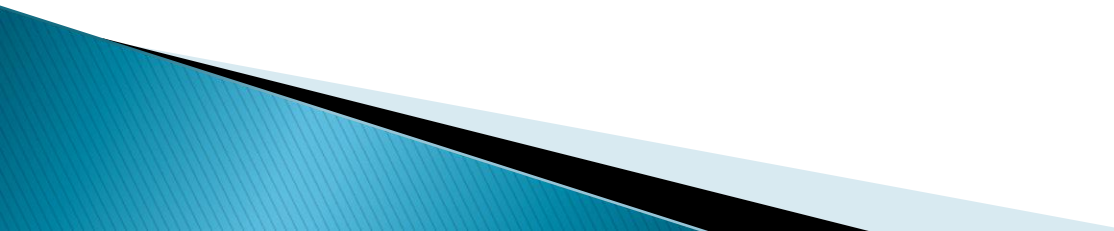
The Results Visit

- ▶ Person does not have HD
 - The absolute best part of my job!
 - Time for celebration
 - ▶ What if the person is not happy? What might be some reasons?
 - Thought they had HD. Weren't prepared to hear otherwise.
 - Concerned about other family members who have HD or are at risk for HD.
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What Next?

- ▶ You are not going to get HD. What do you do now?
 - ▶ Volunteer in your community to help beat HD!
 - ▶ Live your life.
 - ▶ Help other family members.
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The Results Visit

- ▶ Person has HD
 - ▶ Many different responses
 - Most people are sad.
 - Some people cry.
 - Some people just sit.
 - People who were having symptoms may be relieved to finally know for sure.
 - ▶ Essential to have someone with you at the results visit
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What Next?

- ▶ You will eventually get HD. What do you do now?
 - ▶ Volunteer in your community to help beat HD!
 - ▶ Live your life.
 - ▶ Consider enrolling in research studies.
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