The Genetic Testing Process Unraveled

Katie Plunkett, MS, CGC
Baylor College of Medicine
Houston, TX

Huntington's Disease Society of America

The information provided by speakers in workshops, forums, sharing/networking sessions and any other educational presentation made as part of the 2012 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

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

Huntington's Disease
Society of America

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

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

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.

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

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

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.

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.

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

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.