Genetic Issues and Huntington Disease

Katherine S. Hunt, MS, CGC
Assistant Professor in Medicine
Mayo Clinic Arizona
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Presenter Disclosures

Katherine S. Hunt

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

No relationships to disclose or list
Objective:

To review the genetics of HD and discuss the implications of undergoing gene testing.
Outline

- Explain underlying genetic cause of HD
- Outline protocol for pre-symptomatic gene testing
- Discuss psychological, financial and familial implications of gene testing
- Explore gene testing options available to couples at risk to have a child with HD
Genetic terminology

- **Gene**: Contains information needed for our bodies to function properly
  - Encoded in the DNA molecule which is composed of bases
  - DNA alphabet: A, G, T, C
- **Chromosome**: Contains all of our DNA
  - 46 XX-female karyotype
  - 46 XY-male karyotype
Chromosomes, DNA and Genes

Cell

Nucleus

Chromosomes

Gene

Protein
DNA
(deoxyribonucleic acid)

Bases

Adenine (A)
Thymine (T)
Cytosine (C)
Guanine (G)

Base pair
Huntingtin Gene

• Huntingtin gene is found on chromosome number 4-testing available since 1993

• HD is caused by an expanded number of CAG repeats on chromosome 4

• Number of CAG repeats can predict type of HD and other clinical characteristics

• HD is inherited in autosomal dominant manner-most families have an affected parent
Autosomal Dominant Inheritance

- □ = Affected
- ○ = Unaffected
CAG trinucleotide repeats.....

- **Unaffected**: HD gene = 26 or fewer CAG repeats
- **Indeterminate**: HD gene = 27-35 CAG repeats (at risk to have affected child)
- **Reduced penetrance**: HD gene = 36 -39 CAG repeats (may or may not be affected)
- **Affected**: HD gene = 40 or more CAG repeats
Genetic terminology

• Reduced penetrance: Individuals with the HD gene do not exhibit symptoms of the condition

• Anticipation: Increasing severity or decreasing age of onset seen in successive generations
  • Seen more frequently with paternal transmission of HD gene
Pre-symptomatic Gene Testing for HD

*Support person*

- Genetic Counseling
- Psychological assessment
- Neurological evaluation
- Blood draw for testing
- Cost of gene test ~$255
- Discussion of results
- Follow-up
Rationale for Genetic Counseling Prior to Testing

- Family disease-affects individuals beyond the patient
- Provide psychological support and assist patient in finding additional resources if necessary
- Educate patient and family on inheritance of condition
- Help patient understand implications of testing and how to prepare for results
Genetic Counseling Model

• Non-directive-patient decides what is best for them
• Provide accurate information about disorder and testing procedures
• Address concerns regarding health insurance, life insurance, disability, etc.
• Provide informed consent prior to test and disclose results following test
Questions to ask yourself prior to undergoing testing

• How will knowing the results change the way I feel about myself?
• How will knowing the results change my relationships?
• How will I communicate my results to my family, friends and co-workers?
• How have I coped with difficult information or situations in the past?
Questions to ask yourself prior to undergoing testing

- How will knowing the results change my life goals?
- What will I do differently with my life if I test positive?
- Am I afraid that I will lose my job if I test positive?
- What don’t I understand about this condition?
Questions to ask yourself prior to undergoing testing

• What will I do differently with my life if I test negative?

• Who can I talk with about my feelings?

• What will I do after I learn my results?

• What am I most afraid of?
Necessary prior to testing…

- Discuss financial future:
  - If you are primary caregiver, who will provide for family?
  - Do you have life insurance?
  - Do you have long term care insurance?
  - Do you have health insurance?
  - Do you understand Medicare/Medicaid laws?
Understand best timing for testing

- Lifecycle-
  - Adolescent/young adult-tested only when 18yo or older
    - Unmarried-do you want to get married?
    - Married-do you want to have children or how will you inform your children?
  - School and career choices
- Adult
  - Unmarried
  - Married
Benefits of Knowing

- Reproductive planning—for current children and for future offspring
- Need to know—reduce anxiety of not knowing
- To plan future life goals
Reason to Decline Testing

- Emotionally not prepared to handle results
- Unresolved grief over loss of an affected relative
- Conflict with family or friends over decision to be tested
- No insurance or long-term disability or life insurance
Options for Couples Who Want to Have Children

• Donor egg/donor sperm
  • Use the egg or sperm of an unaffected individual

• Pre-implantation genetic diagnosis
  • Test a developing fetus prior to implantation into mother’s uterus

• Prenatal testing
  • Test the fetus during pregnancy for the HD gene
Resources

• To find a genetic counselor to discuss genetic testing or prenatal testing:
• [www.nsgc.org](http://www.nsgc.org) National Society of Genetic Counselors
• Caring for People with Huntington’s Disease: [www.kumc.edu/hospital/huntingtons/index.html](http://www.kumc.edu/hospital/huntingtons/index.html)
Resources

• To find an attorney who specializes in Medicaid law:

• [website](http://www.naela.org/memberdirectory)