

# **Genetic Issues and Huntington Disease**

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## **Huntington's Disease Society of America**

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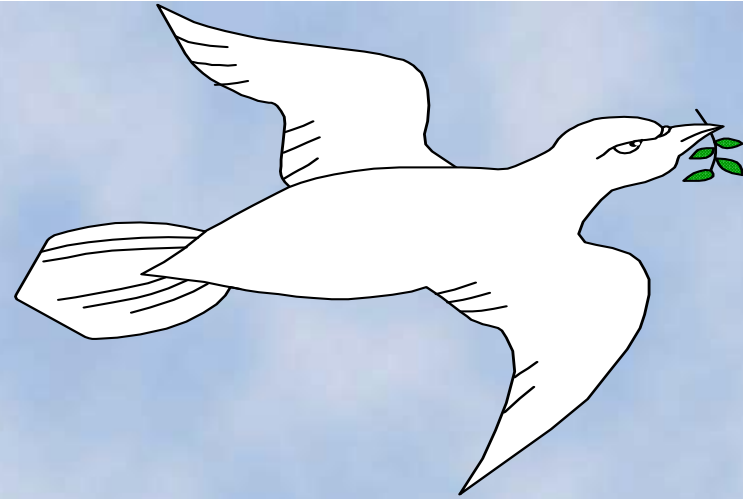
Katherine S. Hunt

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

No relationships to disclose  
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**Objective:**



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

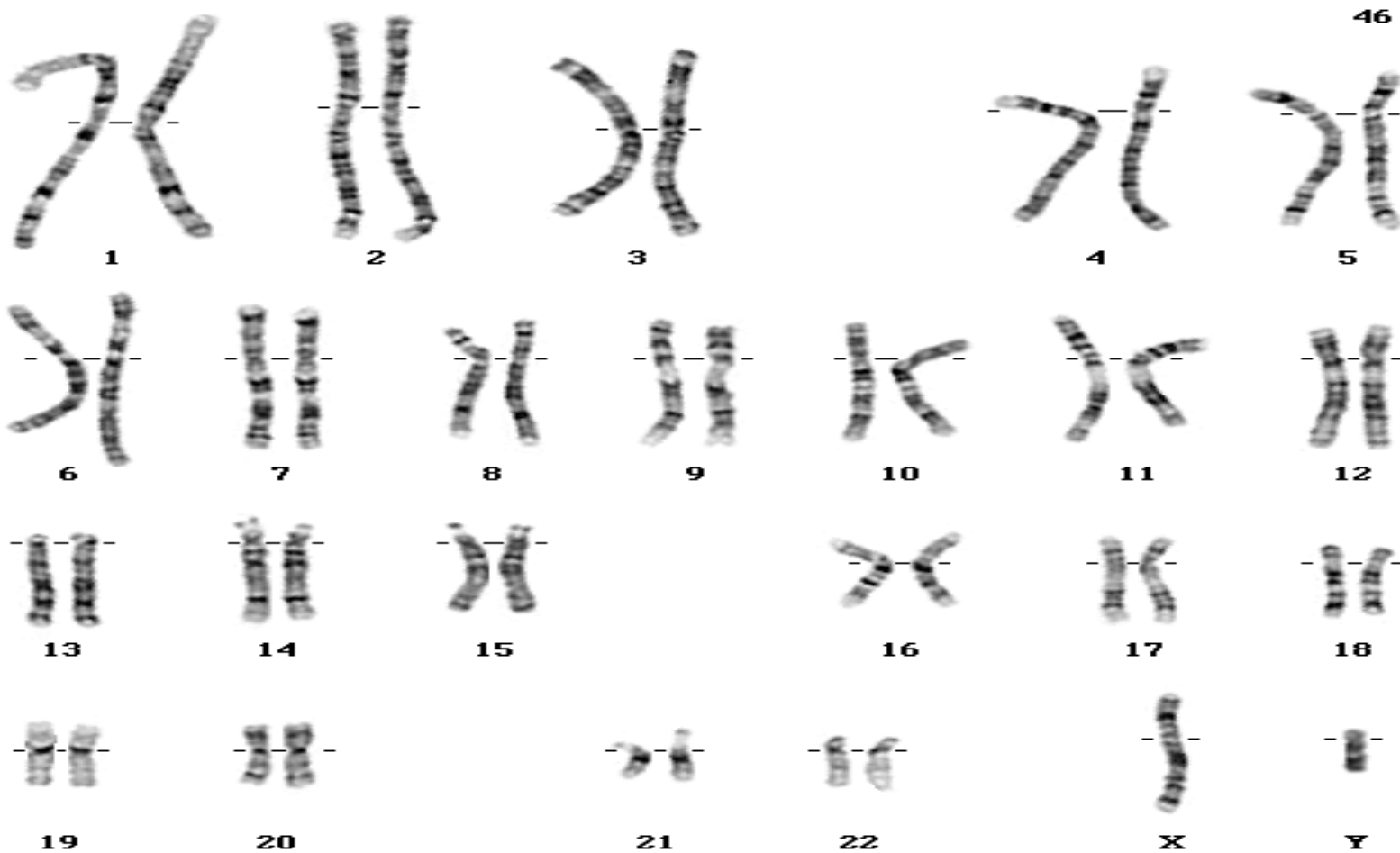
# Outline

- 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

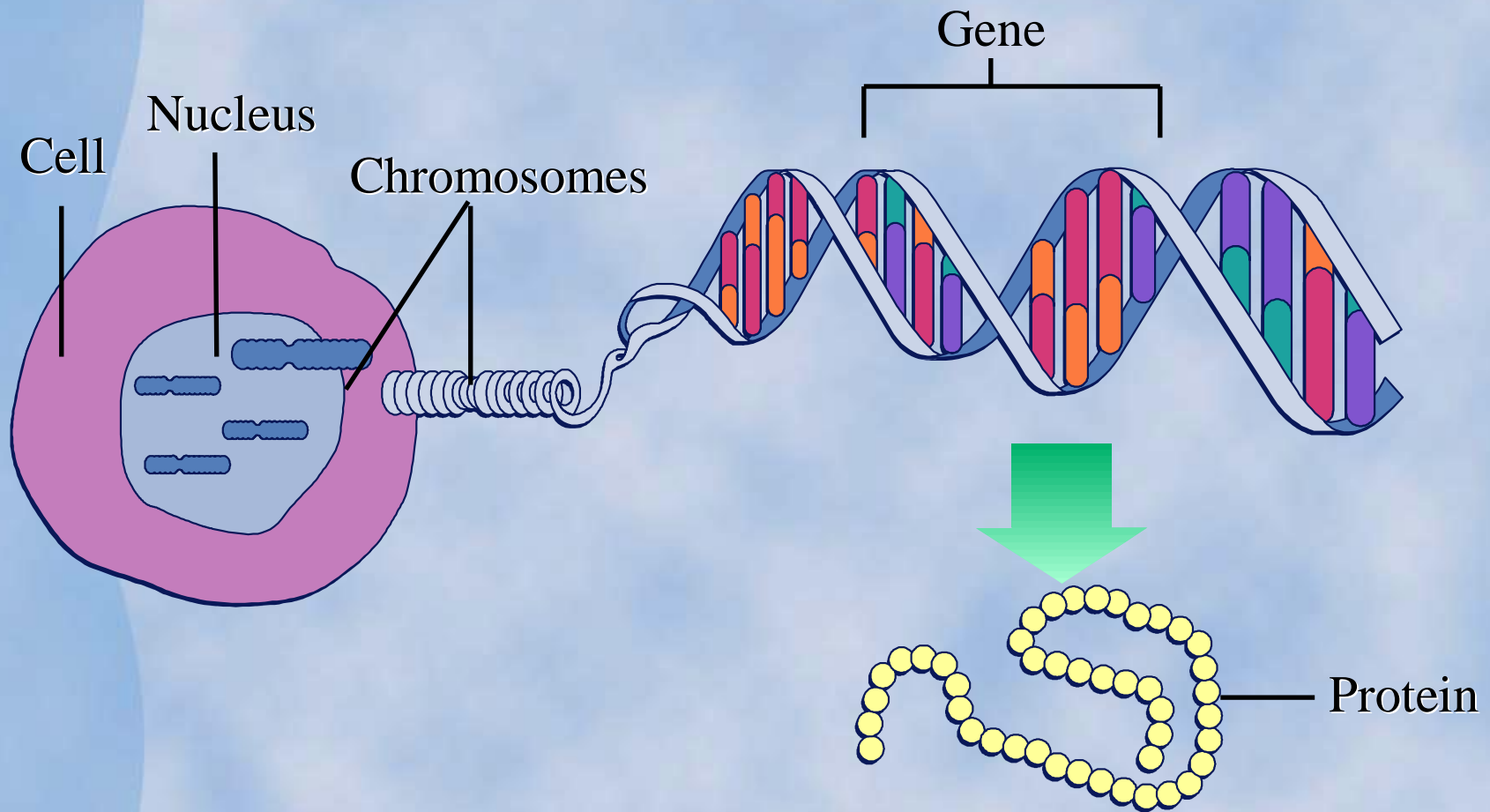
# Genetic terminology

- 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

# Human Chromosomes

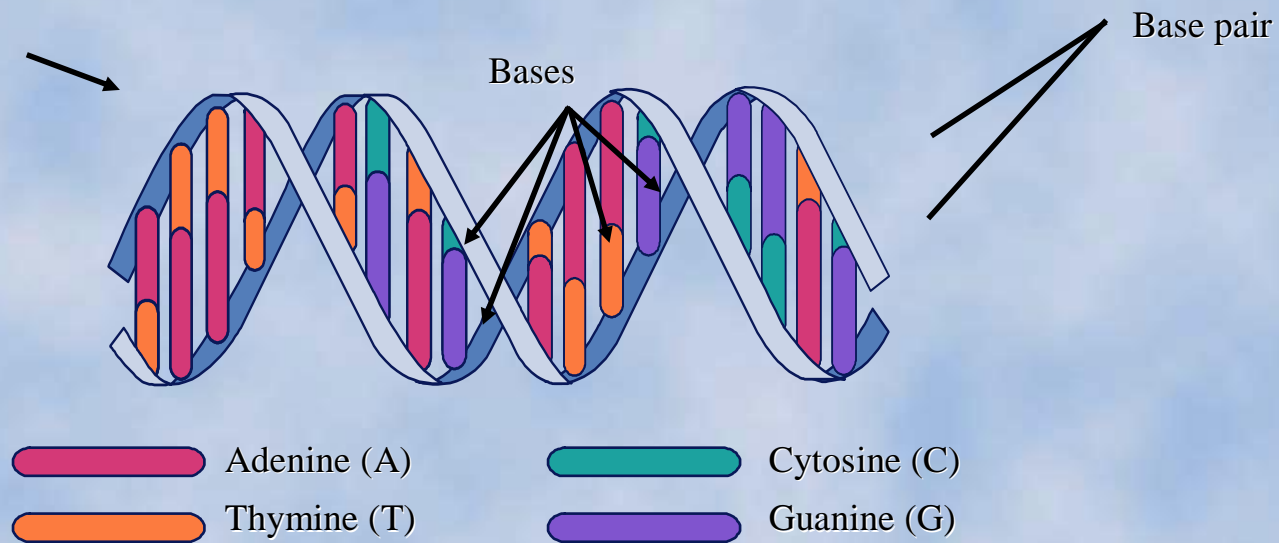


# Chromosomes, DNA and Genes





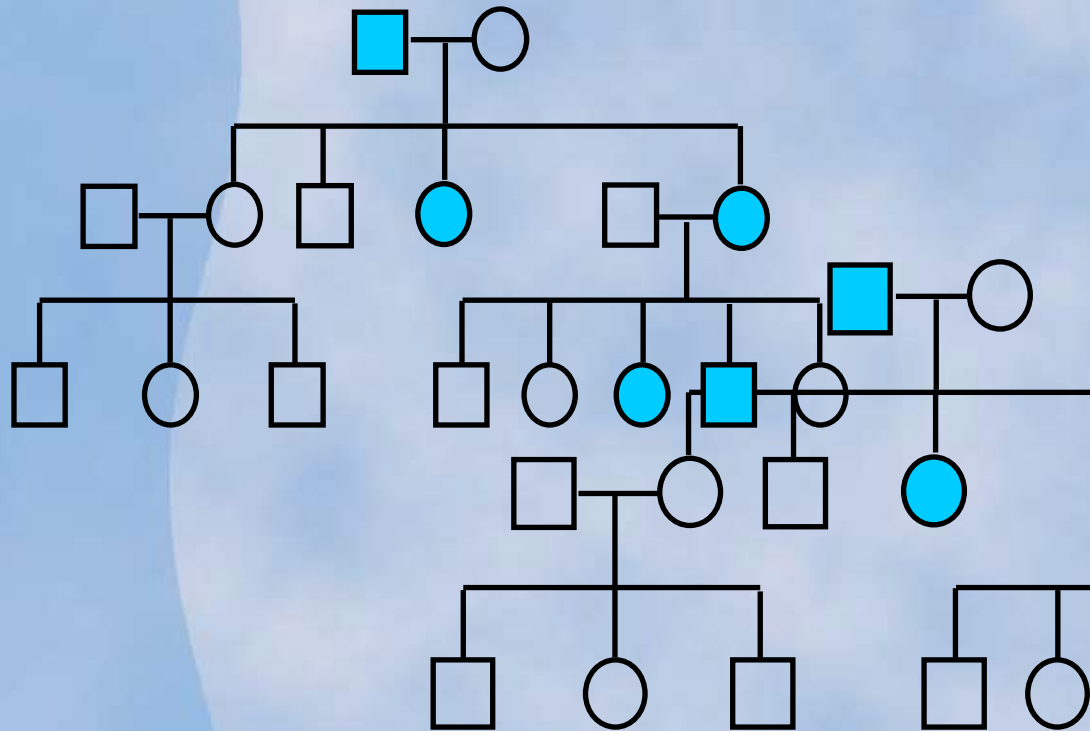
# DNA (deoxyribonucleic acid)



# Huntingtin Gene

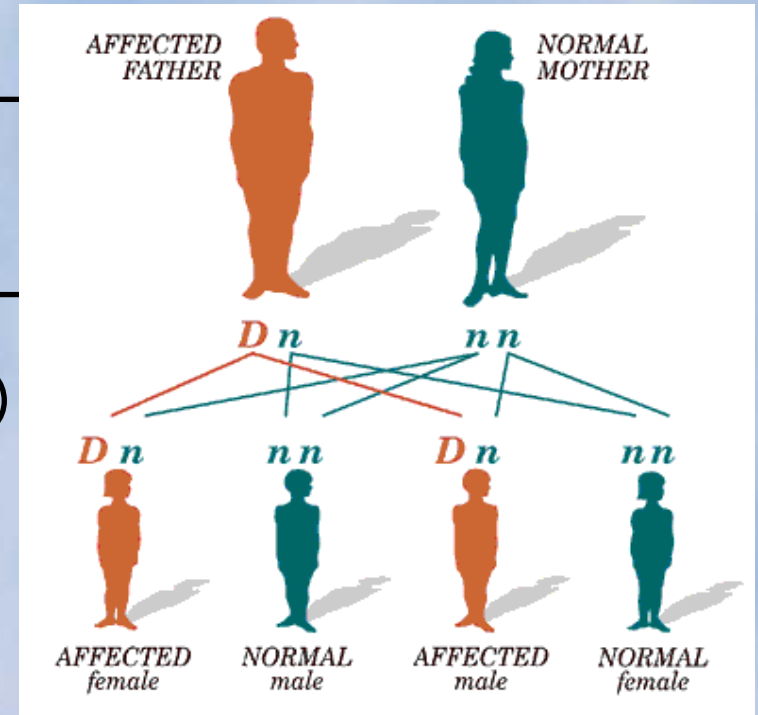
- Huntingtin gene is found on chromosome number 4-testing available since 1993
- HD is caused by an expanded number of CAG repeats on chromosome 4
- Number of CAG repeats can predict type of HD and other clinical characteristics
- HD is inherited in autosomal dominant manner-most families have an affected 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:
- [www.naela.org/memberdirectory](http://www.naela.org/memberdirectory)