



Genetic Testing for Huntington Disease

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Genetic Counseling

- The process of helping people understand the medical, psychological, and family implications of a genetic disease
- This includes
 - Interpreting family and medical histories
 - Educating patients
 - Counseling to promote informed choices



Huntington Disease

- Affects:

- Movements – chorea, may appear fidgety at first
- Cognition – Problems with short term memory, ability to perform daily tasks
- Behavior – Emotion, personality, affect changes, mood swings, easily irritated

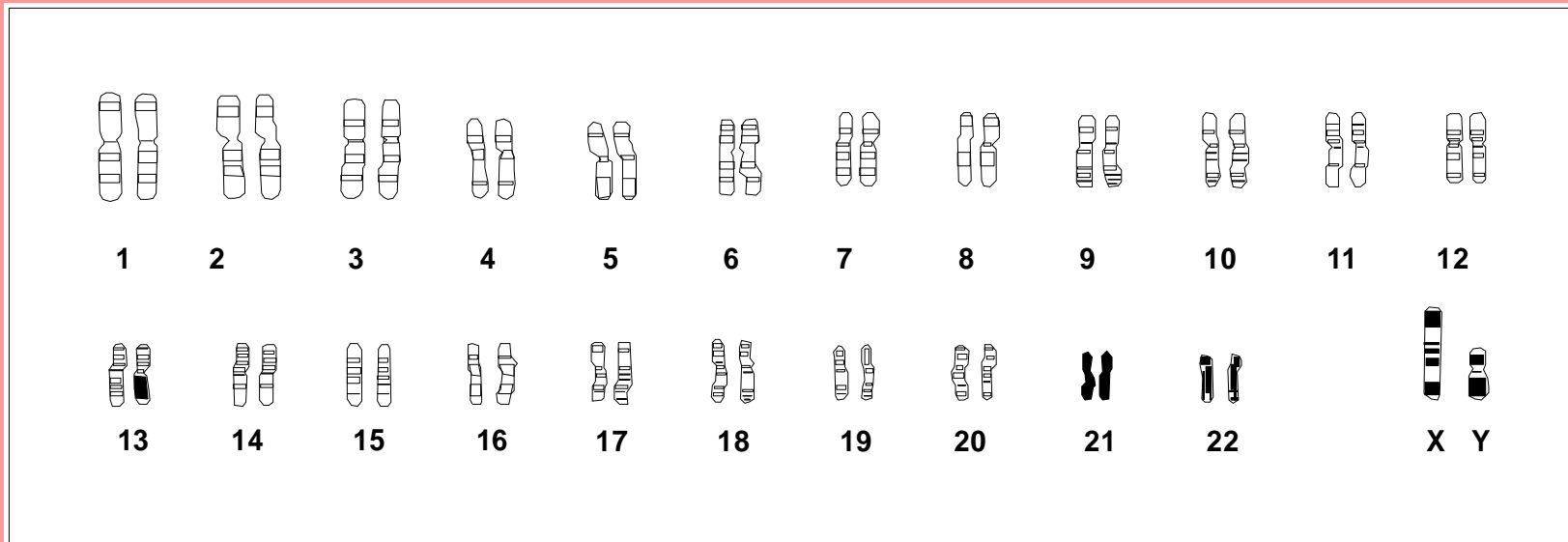
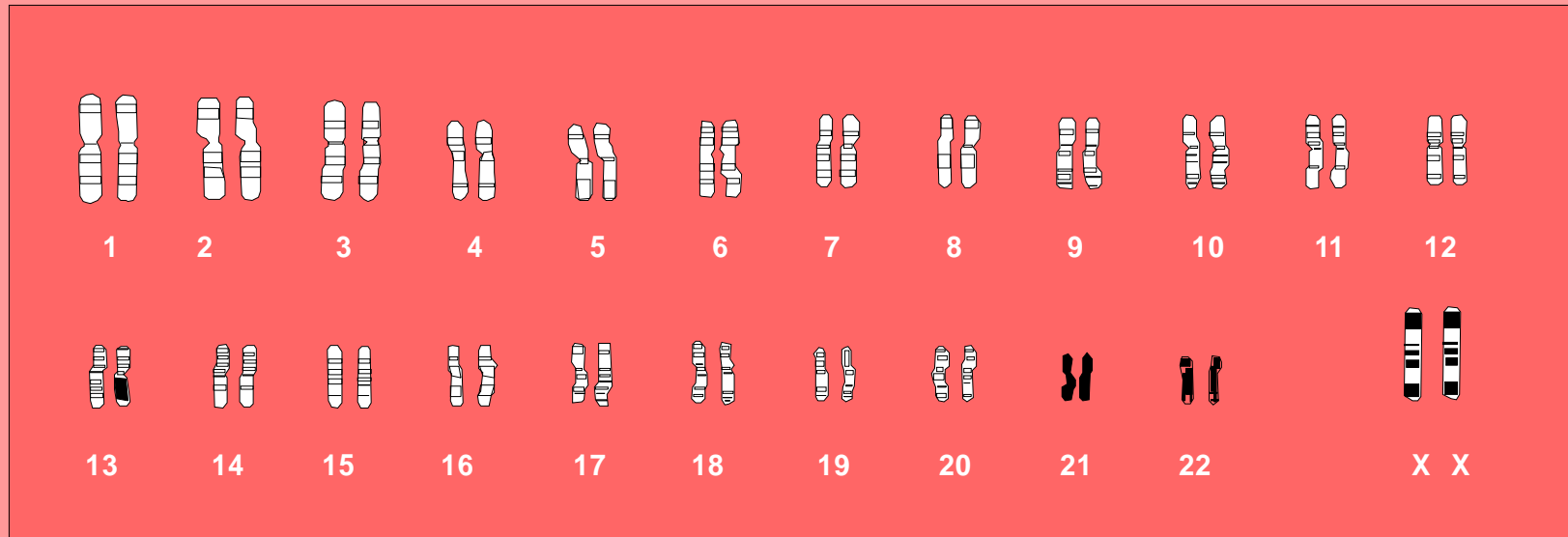
- Age of onset varies – ave age 35-40

- Course of disease 10-20 years

- Autosomal dominant

- Prevalence 3-7/100,000

CHROMOSOMES



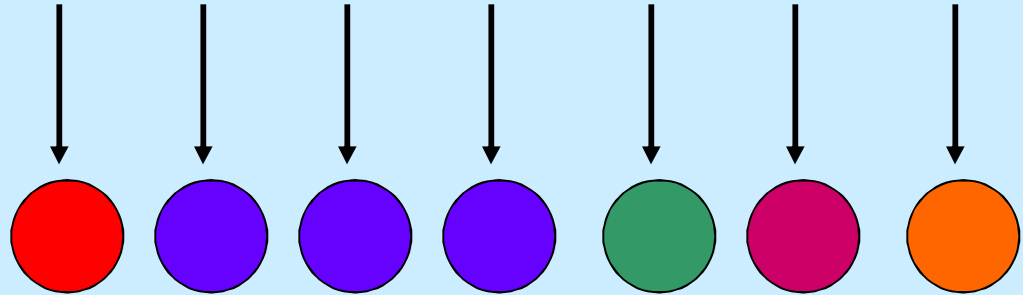
Chromosome



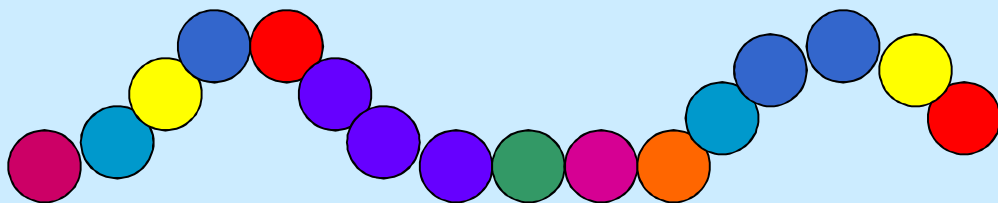
Gene

Gene

...**ATC****CAG****CAG****CAG** **GTG** **CTC** **AAG**...



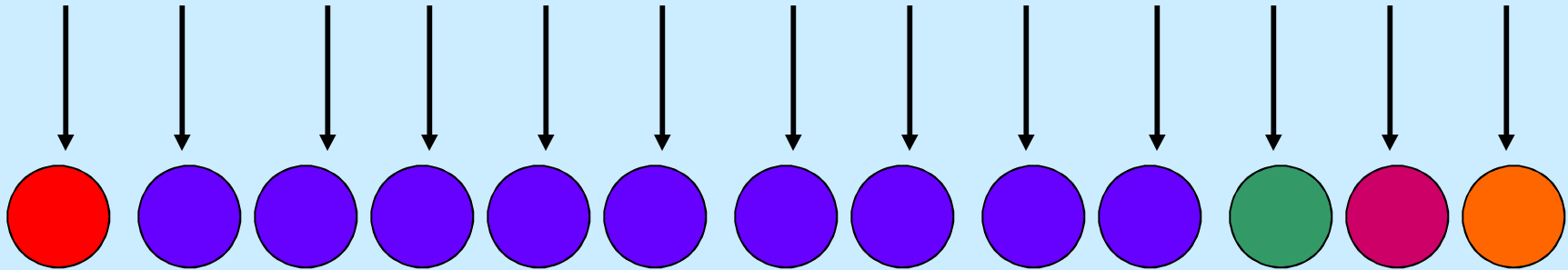
Amino Acids



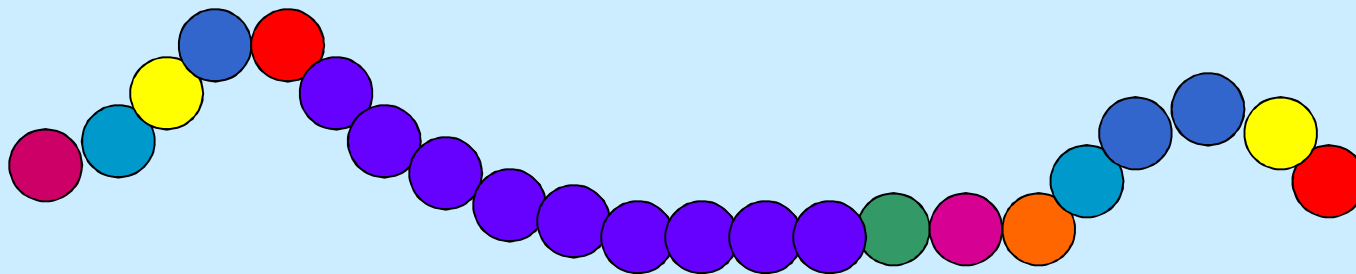
Protein

Huntington Disease

...**ATC** **CAG** **CAG** **CAG** **CAG** **CAG** **CAG** **CAG** **CAG** **CAG** **CAG** **GTG** **CTC** **AAG**...



Amino Acids



Abnormal Protein



Trinucleotide Repeats con't

- ≤ 26 repeats – No HD, cannot pass on
- **27-35** repeats – No HD, can expand when passed on
- **36-39** repeats – Gray area, some develop HD, some do not
- ≥ 40 repeats – Will develop HD if live long enough



Repeats Size Effects

- Rough correlation – the larger the repeat size the earlier the age of onset and more severe symptoms
- Repeats can expand or contract during transmission, more often expands
- **Anticipation** – repeat size expands during transmission
 - Occurs more often when passed from the father
- Juvenile cases usually paternally inherited



Presymptomatic Testing

- People who are at-risk, but do not have symptoms
- Guidelines put out (1994) by Huntington Disease Society of America, International Huntington Association and the World Federation of Neurology
 - Not meant to be restrictive
 - Designed to make sure that people can make informed choices about testing



Protocol Overview

- Initial phone contact
- Initial visit with genetic counselor, geneticist, neurologist
- One or more visits with psychologist or psychiatrist
- Blood draw visit
- Results disclosure
- Post-test counseling over next 1-2 years



Presymptomatic Protocol

- Need to confirm HD by genetic testing in affected family member
- Need to have support person accompany to all visits
- Minors will not be tested

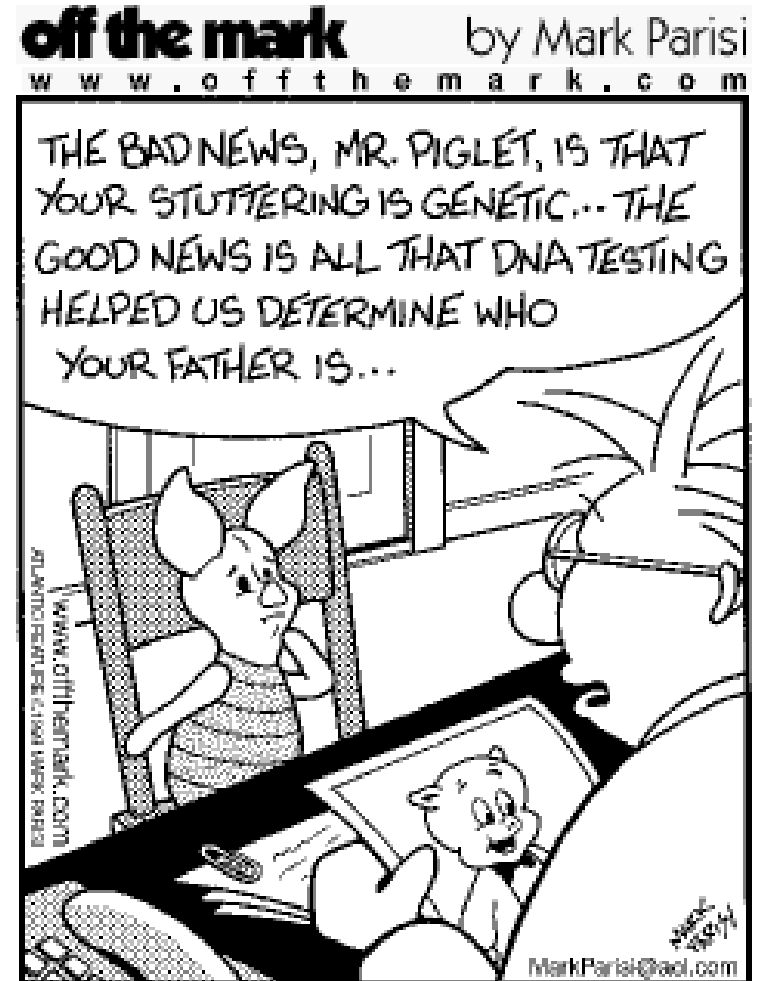


Emotional/Psychosocial Aspects

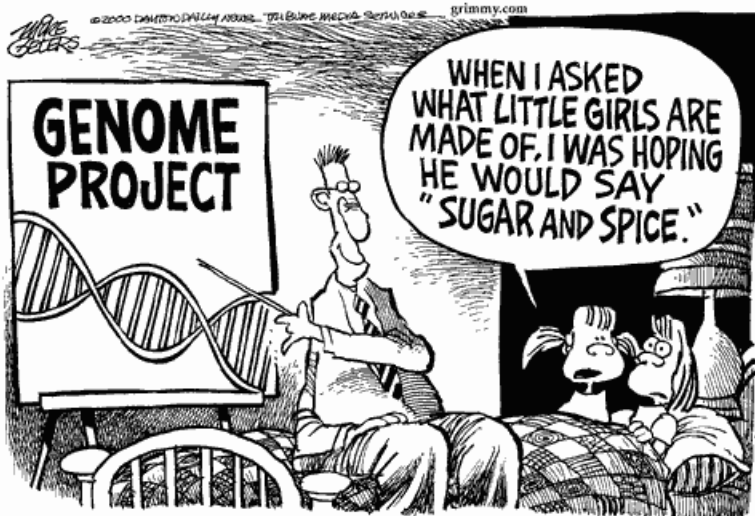
- Affects the whole family
- May have watched other family members with disease
- Caretakers may be caring for more than one family member
- Guilt/fear of having passed HD to kids
- Financial cost of having family member(s) with disability

Reasons for testing

- Reduction of uncertainty
- Preparations for future
- Financial arrangements along with health, life, and disability or long term care insurance
- Family planning



Reasons not to test



- Impact on self or family
- May have inconclusive results – what then?
- Emotional impact of positive *or* negative results
- Cost (approx. \$450, subject to change)
- Genetic Discrimination



Family Planning

- Prenatal Testing
- Pre-Implantation Genetic Diagnosis (PGD)
 - Non-disclosure PGD
 - Exclusion PGD
- Sperm/Egg Donor
- Adoption



Genetic Discrimination: MI State Regulations

■ **Employment Statute**

- Your employer cannot require, obtain, or perform genetic testing as a condition for your employment

■ **Genetic Privacy Law Statute**

- Consent is required to perform genetic testing

■ **Health Insurance Statute**

- Insurance company may not require or disclose genetic information without consent

■ **Life, Disability, and Long-Term Care Insurance**

- Not protected



Genetic Discrimination: National Regulations

■ **GINA**

- **Genetic Information Nondiscrimination Act**

■ **Employment Statute**

- Your employer cannot request, require, or purchase genetic information; they cannot use genetic information in employment decisions

■ **Health Insurance Statute**

- Insurance company may not request or require genetic information; genetic testing alone cannot make someone ineligible for group or individual health coverage

■ **Life, Disability, and Long-Term Care Insurance**

- Not protected

Thank You!

