

# HD 101: THE BASICS

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What comes to mind when  
you hear the term HD?





# WHAT IS HD?

- HD is a **genetic, neurodegenerative brain disorder**
  - Genetic: means it is passed through families
  - Neuro: means it affects the brain
  - Degenerative: means it gradually gets worse over time

Simply put, Huntington's Disease is a condition that affects the brain and nervous system, and gradually gets worse as time wears on.



# WHAT ARE THE SYMPTOMS OF HD?

## Movement

- Involuntary movements
  - This is called chorea
- Facial Tics
- Trouble Balancing
- Hard time with grip
- Trouble swallowing
- Trouble sitting still

## Memory

Mental abilities are impacted by HD. Someone may have a hard time with:

- Multitasking
- Rational thinking
- Learning new information
- Decision making
- Communicating
- Memory

## Mood

Behavior and mood can be impacted by HD. It can appear in the form of :

- Depression
- Anxiety
- Denial
- Reduced ability to express emotions
- Anger
- Mood swings
- Repeating behaviors



# EVERYONE IS DIFFERENT

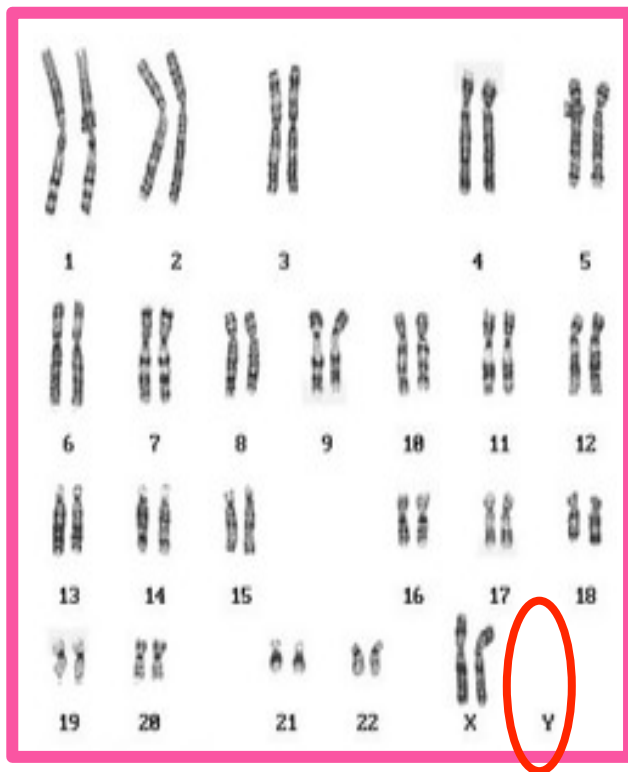
- HD impacts everyone differently
  - Some people may have a lot of movements while others may have more behavior or memory issues
- No two patients or even family members are alike.



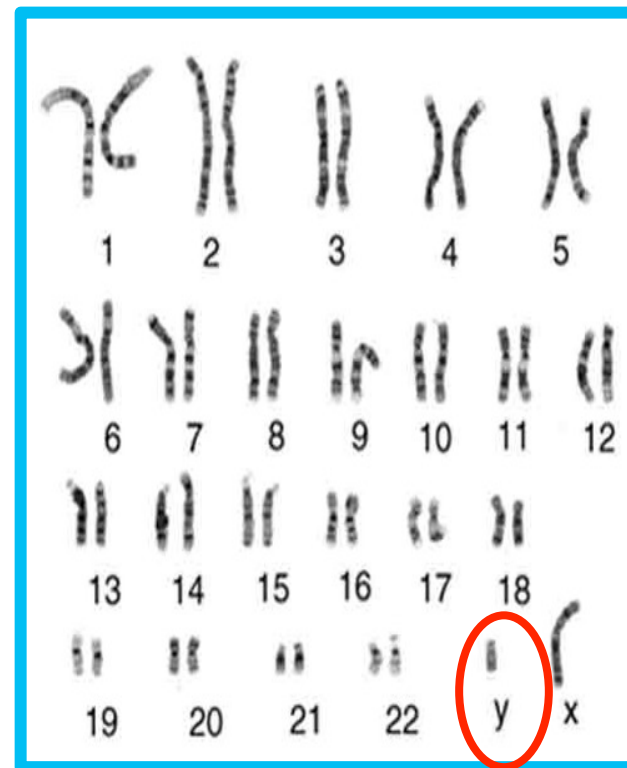


How do you get HD?

# LET'S TALK CHROMOSOMES



Mom's  
Chromosomes

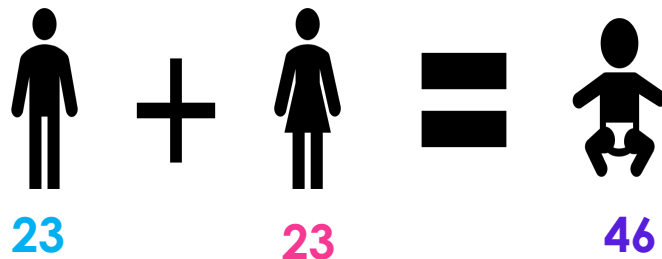


Dad's Chromosomes



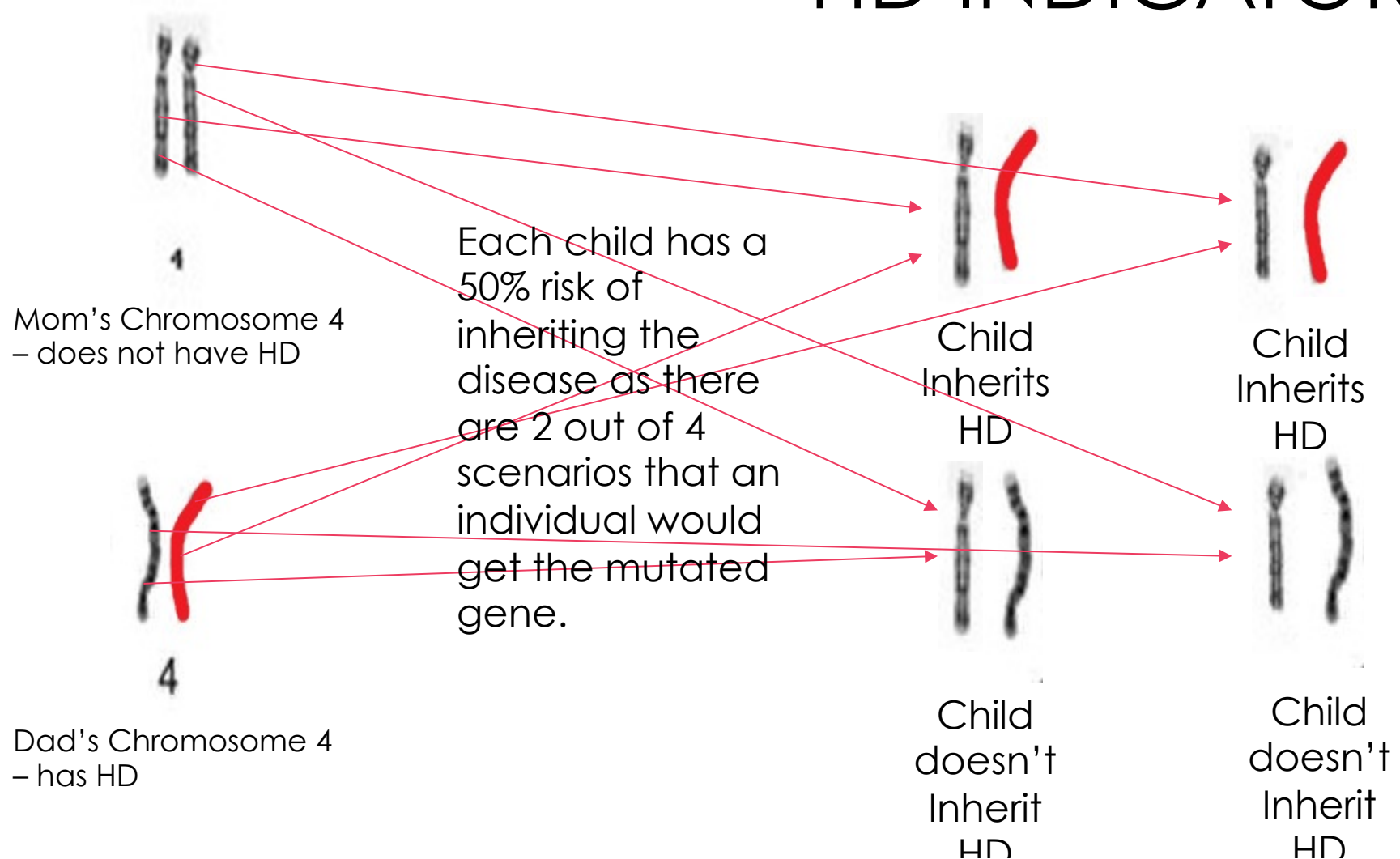
# BIOLOGY 101

- Every person has 46 chromosomes
  - 23 come from mom
  - 23 come from dad



- Chromosomes contain genetic information
  - They inform the cell whether you'll be a boy or girl, what color eyes you have, what color hair etc
- Chromosomes come in pairs
  - You have 2 chromosome 1's, 2 chromosome 2's etc

# CHROMOSOME 4 – THE HD INDICATOR



# SO HOW DOES SOMEONE GET HD?

- Remember we talked about HD being genetic
  - Every child who has a parent with HD has a 50% risk for inheriting HD
- There is a gene called the **Huntingtin Gene (HTT)**. It is located at the top of chromosome 4 (remember – you have two copies of chromosome 4. One from mom and one from dad)
  - The **HTT Gene** is made up of three of the four amino acids that make up our DNA
    - The three amino acids are represented by the letters “C”, “A” and “G”
      - You may hear people talk about their CAG Repeats



Chromosome 4

# IF WE ALL HAVE THE HD GENE, WHY DO ONLY SOME PEOPLE GET HD?



- As we said before, everyone gets two copies of chromosome 4. Someone with HD has an extra long gene and that is what causes HD.
- We can think about ranges when we talk about CAG Lengths:
  - **26 CAG Repeats or Less (Normal Range):** a “normal/typical” gene. This individual won't inherit HD
  - **27-35 CAG Repeats (Risk to Future Generations):** The individual with this length won't necessarily show symptoms, but they could pass the mutation on
  - **36-39 CAG Repeats (Reduced Penetrance Range):** The individual could have symptoms if they live long enough and still have a 50% chance of passing it on.
  - **40 CAG Repeats or Above (Fully Penetrant Range):** This individual will develop HD and has a 50% risk of passing the gene on



# LET'S RECAP ALL THAT INFO

- Every person inherits 23 chromosomes from Mom and 23 from Dad
- **Everyone** has 2 copies of chromosome 4.
  - Huntington's Disease is caused by a mutation at the top of **chromosome 4**
  - The mutation is an expansion of the CAG repeat on the Huntingtin Gene
    - 40 CAG Repeats or above means someone will get HD
    - 26 CAG repeats or less means someone has a "normal" gene
    - 27-39 CAG repeats can mean a few different things





QUESTIONS????