# RECESSIVE, DOMINANT, & SEX-LINKED DISORDERS

- some genetic traits depend on dominant and recessive alleles
- some gene expression depends on the chromosome it is on autosome or sex chromosome

#### DISORDERS BY RECESSIVE ALLELES:

- people with a disorder (affected) must have both recessive alleles (tt, rr, bb)
- parents that are homozygous dominant are considered *normal* (TT, RR, BB)
- parents that are heterozygous for a particular disorder are considered carriers and can pass the trait onto their offspring (Tt, Rr, Bb)
- Diseases/Traits: Albinism, Cystic Fibrosis, PKU (phenylketonuria), Tay-Sachs Disease



AA AA

NORMAL CARRIER

AA AA

CARRIER

AAA

CARRIER

AFFECTED

#### **ALBiNiSM**

- · 1 in 17,000 births
- · no pigmentation in hair, skin, eyes
- · eye sensitivity and vision problems
- UV radiation (sun) can easily damage skin





CLICK FOR CF

**AWARENESS** 

**ViDeO** 

## GENETICS PRACTICE #1

Craig and Nina are married and have four children. Both Craig and Nina have brown eyes and hair and light-medium skin. However, 2 of their 4 children have albinism. (Use the letters AA, Aa, or aa.)

- 1. What is Craig's genotype?
- 2. What is Nina's genotype?
- 3. What is the probability that their next child will also have albinism?

## CYSTIC FIBROSIS

- · 1 in approximately 2000 births
- defective CFTR gene...protein pump in cell membrane not made properly or at all
- · mucus clogs lung, liver, and pancreas
- · affects young children to young adults
- · high mortality rate
- life expectancy currently into the 30s (if given the best care/treatment)

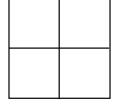
· NO cure yet



#### GENETICS PRACTICE #2

A couple who are both carriers of the gene for cystic fibrosis have two children who have cystic fibrosis. (Use the letters FF, Ff, or ff.)

- 1. What would be the genotypes of the parents?
- What would be the genotype for their children with CF?
- 3. What is the probability that their next child will have cystic fibrosis?



# PKU (PHENYLKETONURIA)

- · 1 in 10,000 births
- · cannot digest phenylalanine and it builds up causing severe complications including brain damage and even death
- · PKU diet does not allow consumption of meat, fish, poultry, milk, eggs, cheese, ice cream, legumes, nuts, or many products containing regular flour
- · NO cure yet



## Tay-sachs

- · most common in people of Jewish ancestry
- · metabolic disorder
- · fat builds up and destroys the brain
- · life expectancy is about 3 years



CLICK FOR AWARENESS VIDEO

# DISORDERS CAUSED BY DOMINANT ALLELES

- people with a dominant allele disorder need to have one allele to inherit the disease
- · homozygous dominant and heterozygous individuals would be affected
- · if a parent has a dominant allele, there is a 50/50 chance of passing it on to their offspring
- Diseases/Traits: achondroplasia and Huntington's disease



#### **ACHONDROPLASIA**

- · most common form of dwarfism
- Aa genotype = achondroplasia



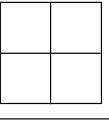




## GENETICS PRACTICE #3

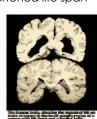
Matt and Amy Roloff have four children. Although Matt and Amy are both little people, they do not have the same type of dwarfism. Amy has achondroplasia, which is a dominant allele disorder. There children are: twins Zach and Jeremy, Molly, and Jake. Three of the four children are average height. Zach has achondroplasia like his mother.

Show the results of the Roloff family in a Punnett square. Matt (aa) is normal or does NOT have achondroplasia. Amy has achondroplasia (Aa).



# HUNTINGTON'S DISCOSC

- · 1 in 10,000
- · gradual deterioration of brain tissue during middle age
- · shortened life span

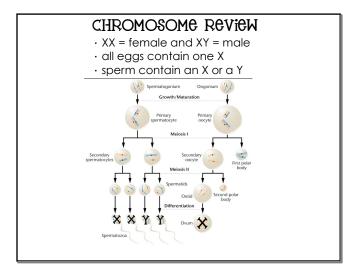


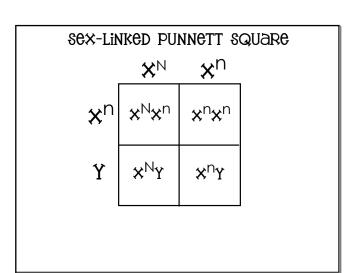


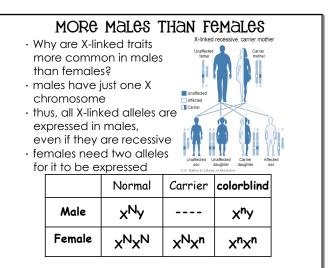


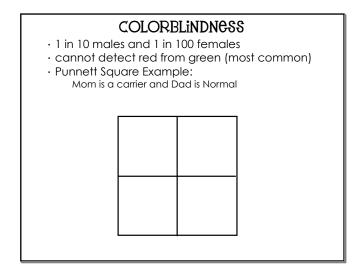
# Sex-Linked Traits

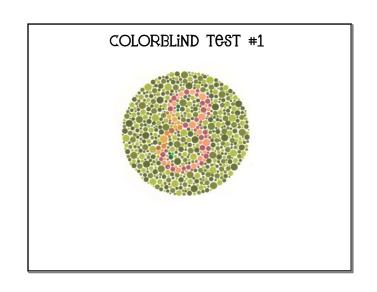
- some traits and disorders are located on the sex chromosomes (23rd pair)
- genes located on the sex chromosomes (X, Y) are said to be "sex-linked"
- · the probability of inheriting a particular trait depends on if your are a boy or girl
- · must use **XX** and **XY** in your Punnett squares
- Diseases/Traits: colorblindness, hemophilia, Duchene's Muscular Dystrophy, ALD (adrenoleukodystrophy)

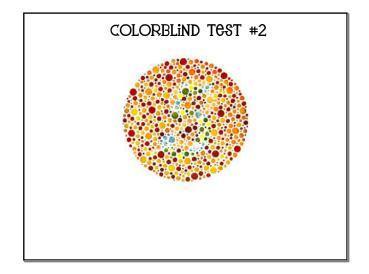


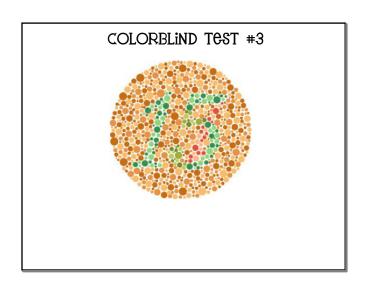


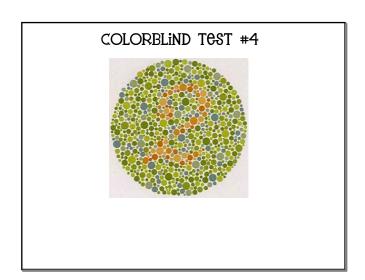


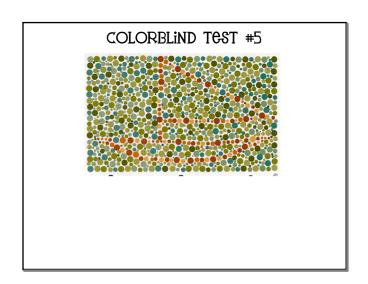


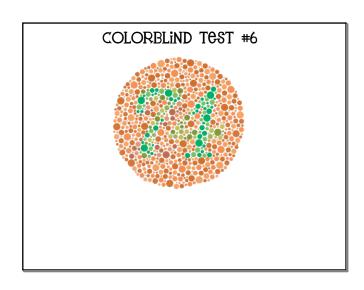












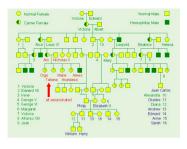
# GENETICS PRACTICE #4

Jeff and Emily are married and expecting their first child. Jeff has normal vision and so does Emily, but she is a carrier for colorblindness. (MUST USE XX and XY.)

- 1. What is the probability that their child will be colorblind?
- They find out that they will be having twins (one boy and one girl)
- 3. What is the probability that their son will be colorblind?
- 4. What is the probability their daughter will be colorblind? Carrier? Normal?

# Hemophilia

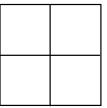
- · 1 in 10,000 males
- · defective gene for clotting factor XIII
  - · "Royal" disease



#### Genetics practice #5

Sofia, whose blood clots normally, has a hemophilic father. Her husband's (Tommy) blood clots normally. Their first child was a girl and their second child was a boy.

- 1. What is Sofia's genotype and phenotype?
- 2. What is Tommy's genotype and phenotype?
- 3. What are the possible genotypes for their daughter?
- 4. What are the possible genotypes for their son?
- 5. What are the chances their 3rd child will be a girl that is also a carrier for hemophilia?



# DUCHENE'S MUSCULAR DYSTROPHY

- · 1 in 3000 males
- · progressive weakening and loss of skeletal muscle
- · rarely live past early adulthood

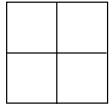




# GENETICS PRACTICE #6

Elizabeth and Sam have an 18 month old son named Scotty. He was late to walk, stumbles easily, and has difficulty getting up and moving around. After many tests, it was determined that Scotty inherited Duchene's Muscular Dystrophy (DMD), which is an X-linked trait. Both parents are unaffected.

- What is Elizabeth's genotype and phenotype?
- 2. What is Sam's genotype and phenotype?
  3. What is the genotype for their son, Scotty?
- 4. What is the probability that they will have
- What is the probability that they will have a girl with DMD?
- 5. Elizabeth is expecting another child. What is the probability she will have another child with DMD?
- She finds out that the child will be a girl.
   What is the probability that she will be
   normal and NOT carry the gene for DMD?



## ALD (adrenoleukodystrophy)

- · 1 in 20,000
- · fat builds up in the brain and disrupts normal activity
- · NO cure yet

