General Biology 1
BIO1101
Syllabus & Textbook: http://goo.gl/rvgdrH

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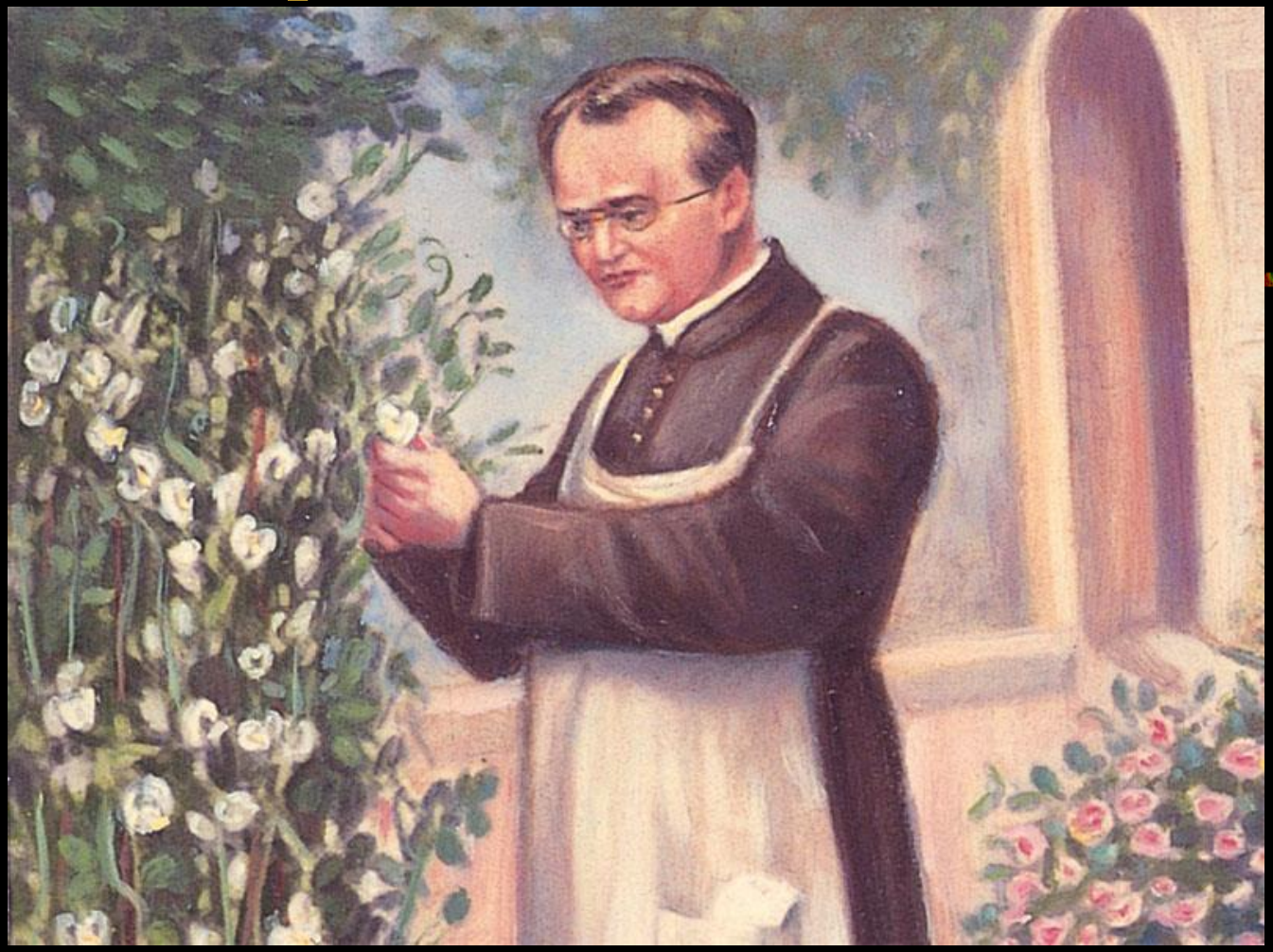
Letter Grade	Numerical Ranges
A	93-100
A-	90-92.9
B+	87-89.9
B	83-86.9
B-	80-82.9
C+	77-79.9
C	70-76.9
D	60-69.9
F	59.9 and below

OER
Lecture: https://openlab.citytech.cuny.edu/bio-oer/page/2/
Lab: https://openlab.citytech.cuny.edu/bio-oer/

Grade Breakdown:
Exams (4): 20% Each
Quizzes: 20% Average

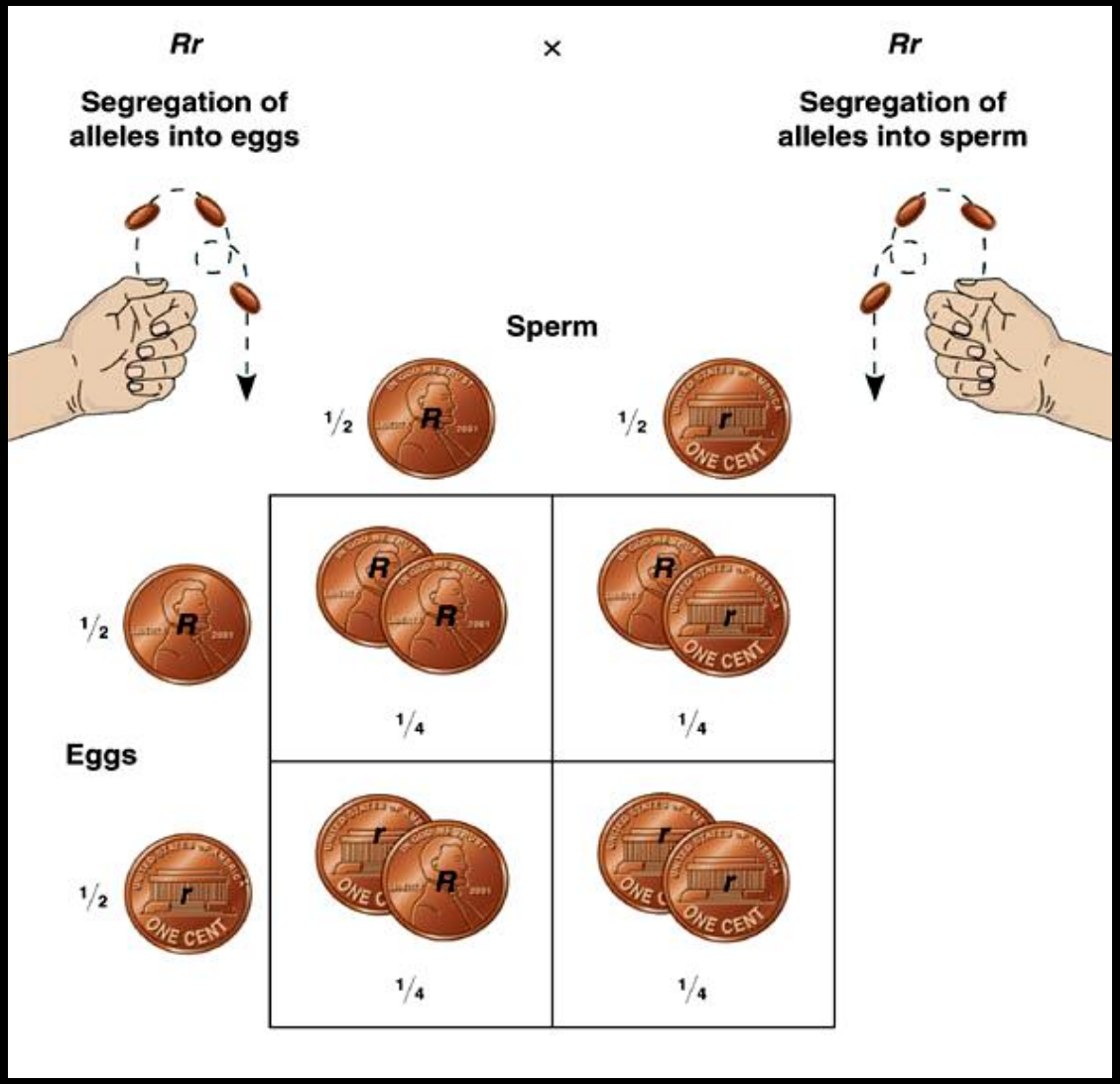
Recap of GENETICS



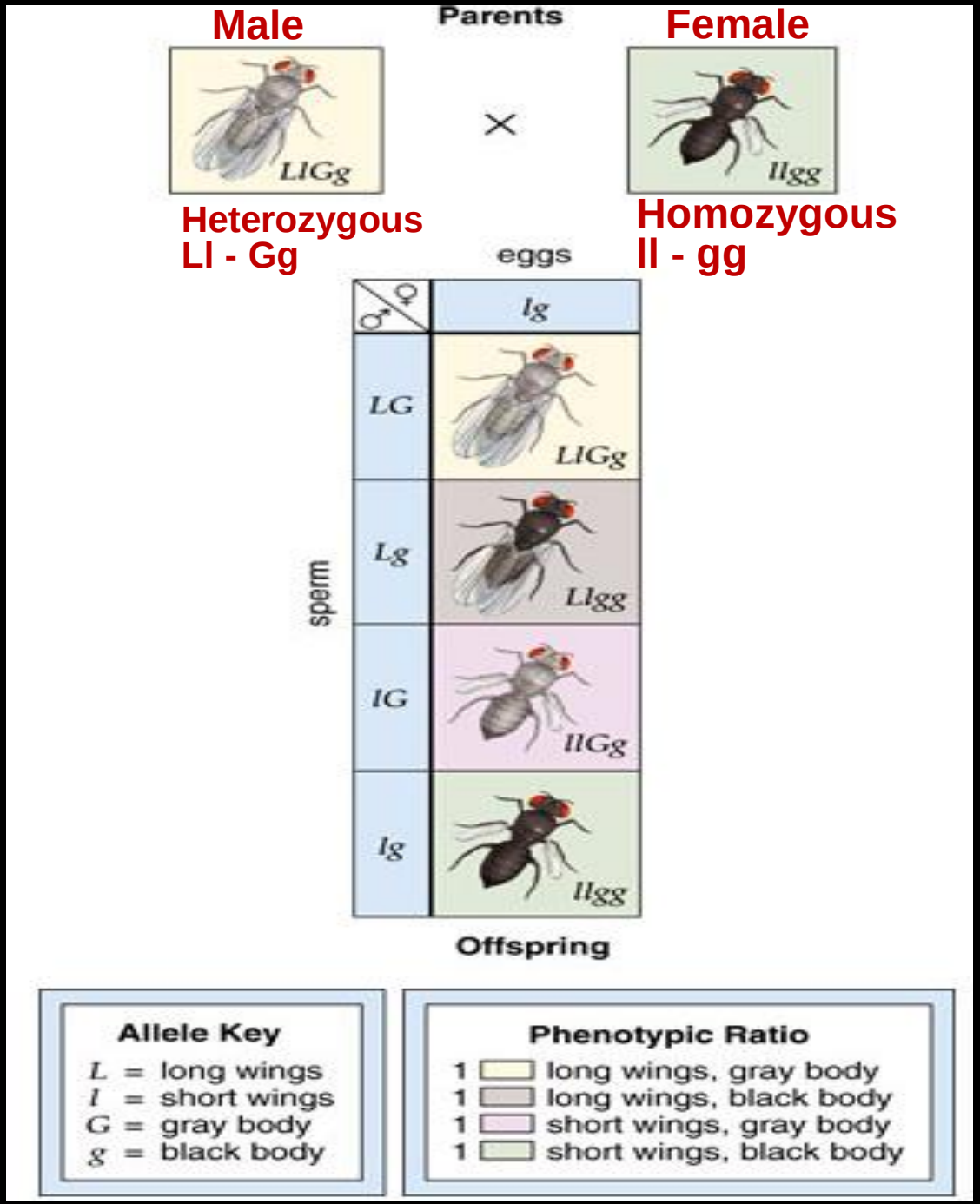
Probability

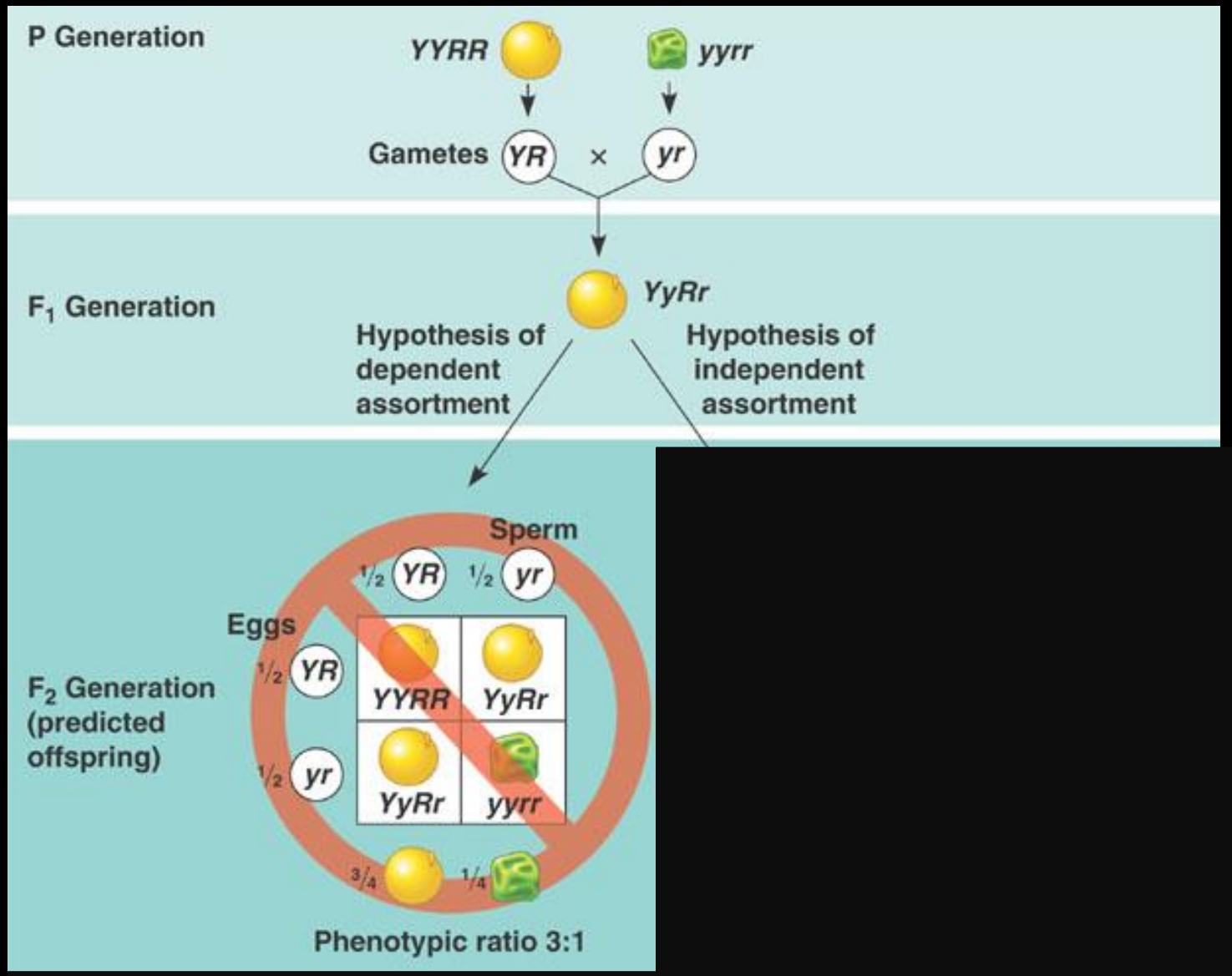
- Assortment of alleles into gametes is random
- Fertilization (which gametes) is random
- Probability = number of ways a certain event could occur divided by the total number of possible outcomes
  - If probability = 1, the event is CERTAIN to happen
  - If probability = 0, the event CANNOT happen
  - Everything in between is a fraction (or percent)
  - Example = flipping a coin: prob. of heads = ½, prob. of tails = ½
  - The probability of all outcomes adds up to 1

Possible outcomes of flipping a coin twice



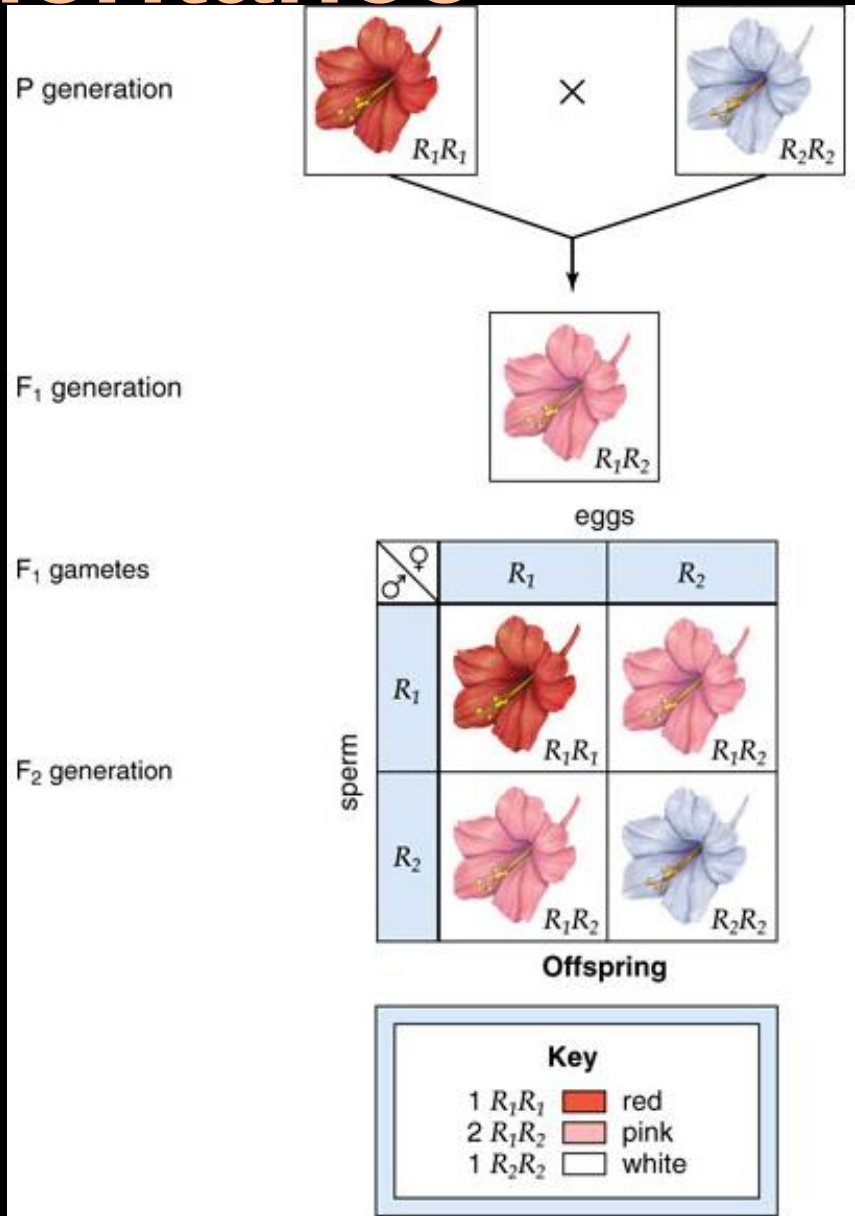
Two-Trait Test Cross -- Independent Assortment




P Generation
YYRR
yyrr
Gametes
YR
×
yr
F1 Generation
YyRr
Hypothesis of dependent assortment
Hypothesis of independent assortment
F2 Generation (predicted offspring)
Sperm
1/2 YR
1/2 yr
Eggs
1/2 YR
1/2 yr
YYRR
YyRr
YyRr
yyrr
3/4
1/4
Phenotypic ratio 3:1

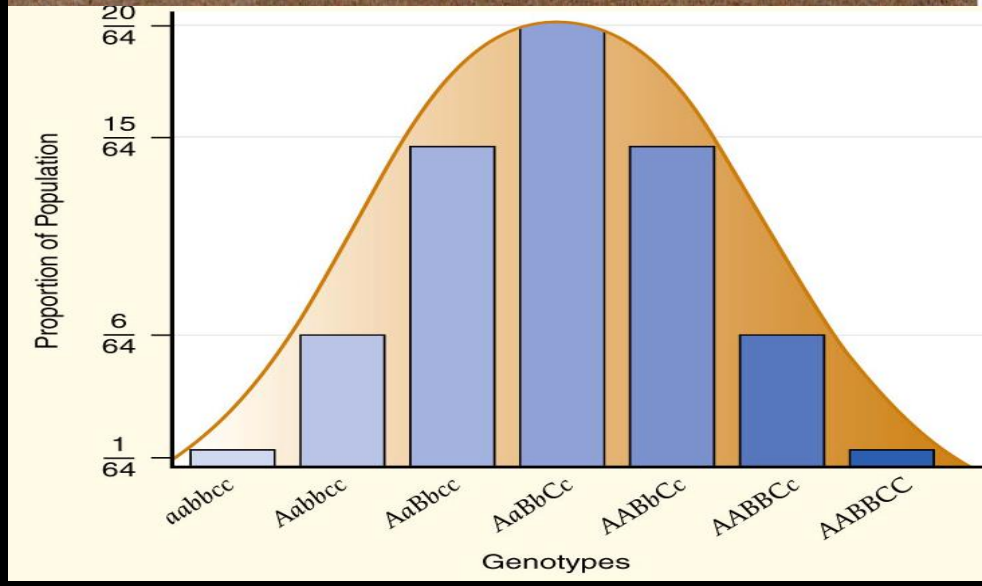
Incomplete Dominance -- Blending Inheritance

- Heterozygote has phenotype intermediate between that of either homozygote
  - Homozygous red has red phenotype
  - Homozygous white has white phenotype
  - Heterozygote has pink (intermediate) phenotype
- Phenotype reveals genotype without test cross



Polygenic Inheritance

- Occurs when a trait is governed by two or more genes having different alleles
- Each dominant allele has a quantitative effect on the phenotype
- These effects are additive
- Result in continuous variation of phenotypes



Nature vs. Nurture

- Environment can influence the expression of genes, so phenotype is not always strictly dependent on genotype
  - E.g., identical twins have the same genes, but are NOT perfectly identical Any Ideas How?
- Genes often establish a **norm of reaction**, which is a range of possible phenotypes for a given genotype.
- So “nature” and “nurture” cooperate to establish phenotypes

Human Genetic Disorders

- **Autosom**e - Any chromosome other than a sex chromosome

- Genetic disorders caused by genes on autosomes are called **autosomal disorders**

  - Some genetic disorders are autosomal dominant
    - An individual with AA has the disorder
    - An individual with Aa has the disorder
    - An individual with aa does NOT have disorder

  - Other genetic disorders are autosomal recessive
    - An individual with AA does NOT have disorder
    - An individual with Aa does NOT have disorder, but is a carrier
    - An individual with aa DOES have the disorder

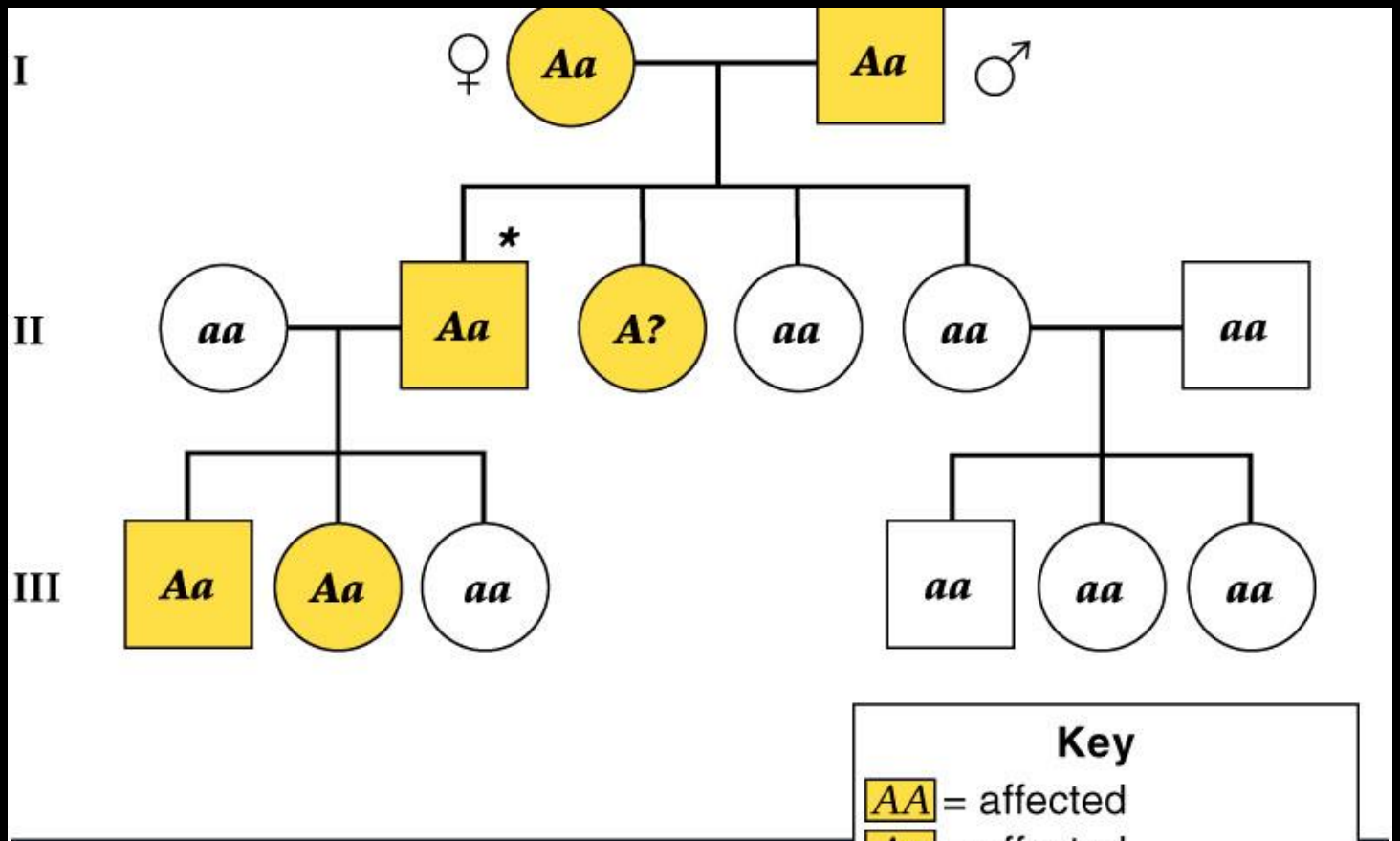
Autosomal Dominant Disorders

- Neurofibromatosis
  - Tan or dark spots develop on skin and darken
  - Small, benign tumors may arise from fibrous nerve coverings
- Huntington Disease
  - Neurological disorder
  - Progressive degeneration of brain cells
    - Severe muscle spasms
    - Personality disorders

Autosomal Recessive Disorders

- Tay-Sachs Disease
  - Progressive deterioration of psychomotor functions
- Cystic Fibrosis
  - Mucus in bronchial tubes and pancreatic ducts is particularly thick and viscous
- Phenylketonuria (PKU)
  - Lack enzyme for normal metabolism of phenylalanine

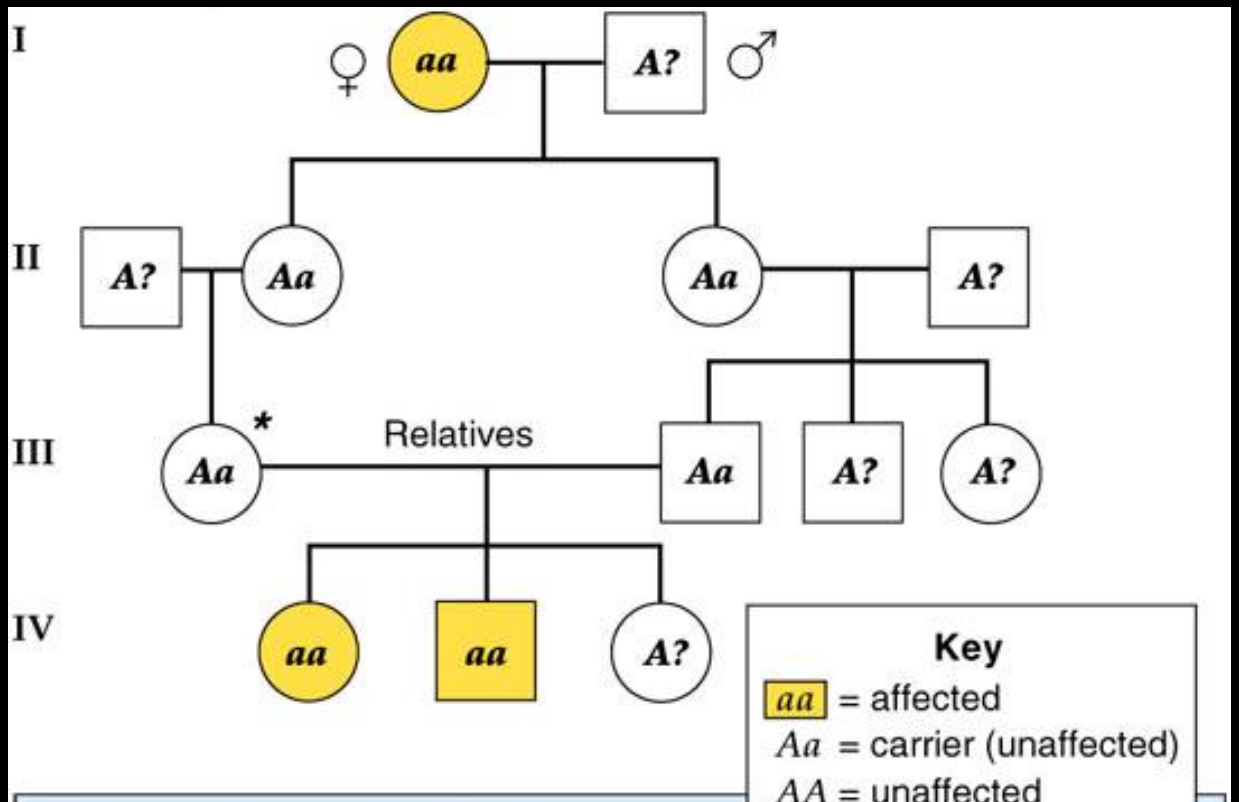
Autosomal Dominant Pedigree Chart



Autosomal dominant disorders

- Affected children will usually have an affected parent.
- Heterozygotes (*Aa*) are affected.
- Two affected parents can produce an unaffected unaffected child.
- Two unaffected parents will not have affected children.
- Both males and females are affected with equal frequency.

Autosomal Recessive Pedigree Chart



Autosomal recessive disorders

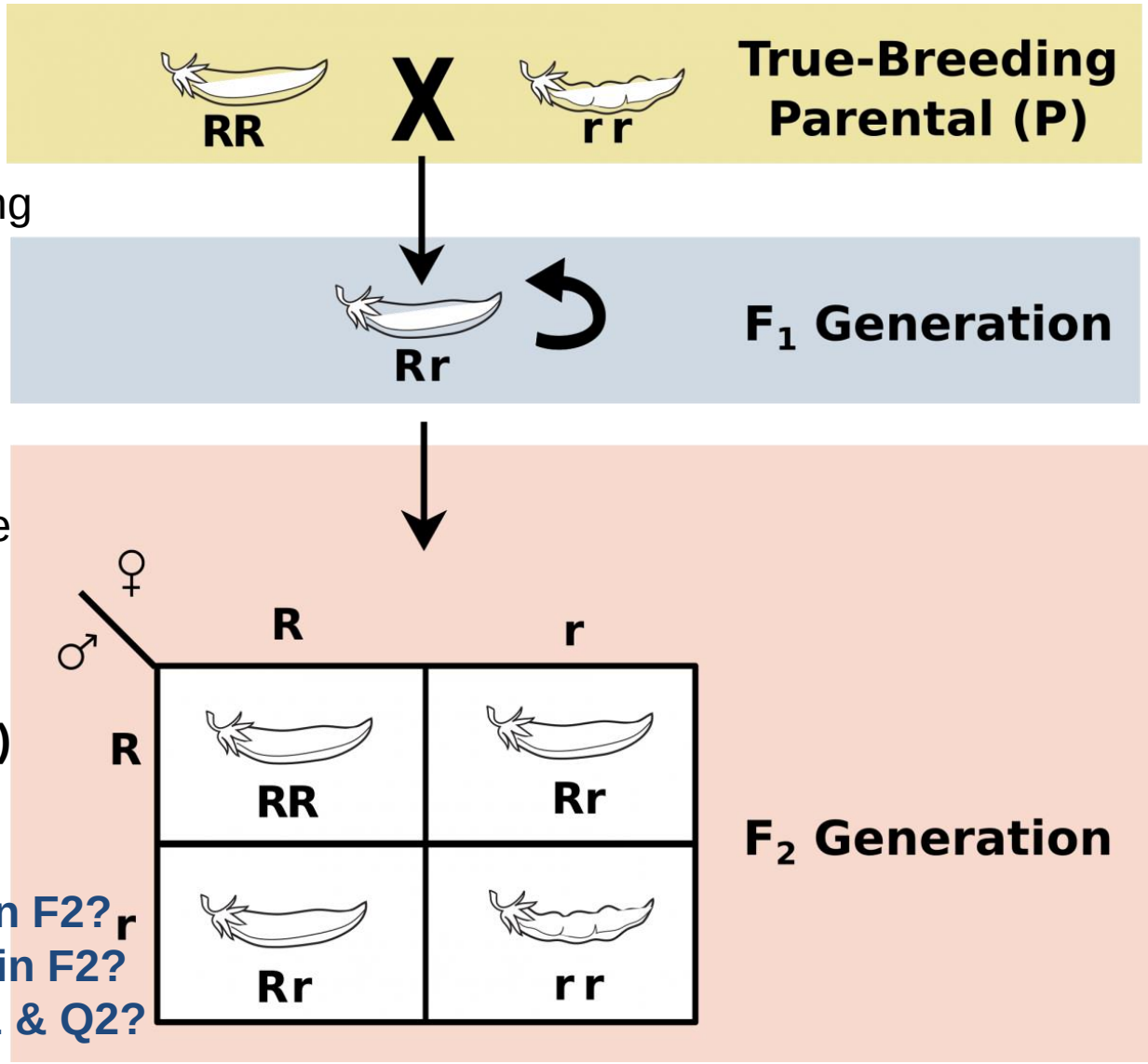
- Most affected children have unaffected parents.
- Heterozygotes (*Aa*) have an unaffected phenotype.
- Two affected parents will always have affected children.
- Affected individuals with homozygous unaffected mates will have unaffected children.
- Close relatives who reproduce are more likely to have affected children.
- Both males and females are affected with equal frequency.

Review Questions

2. Mendelian Genetics: Breeding

https://openlab.citytech.cuny.edu/bio1-oer/genetics/6/

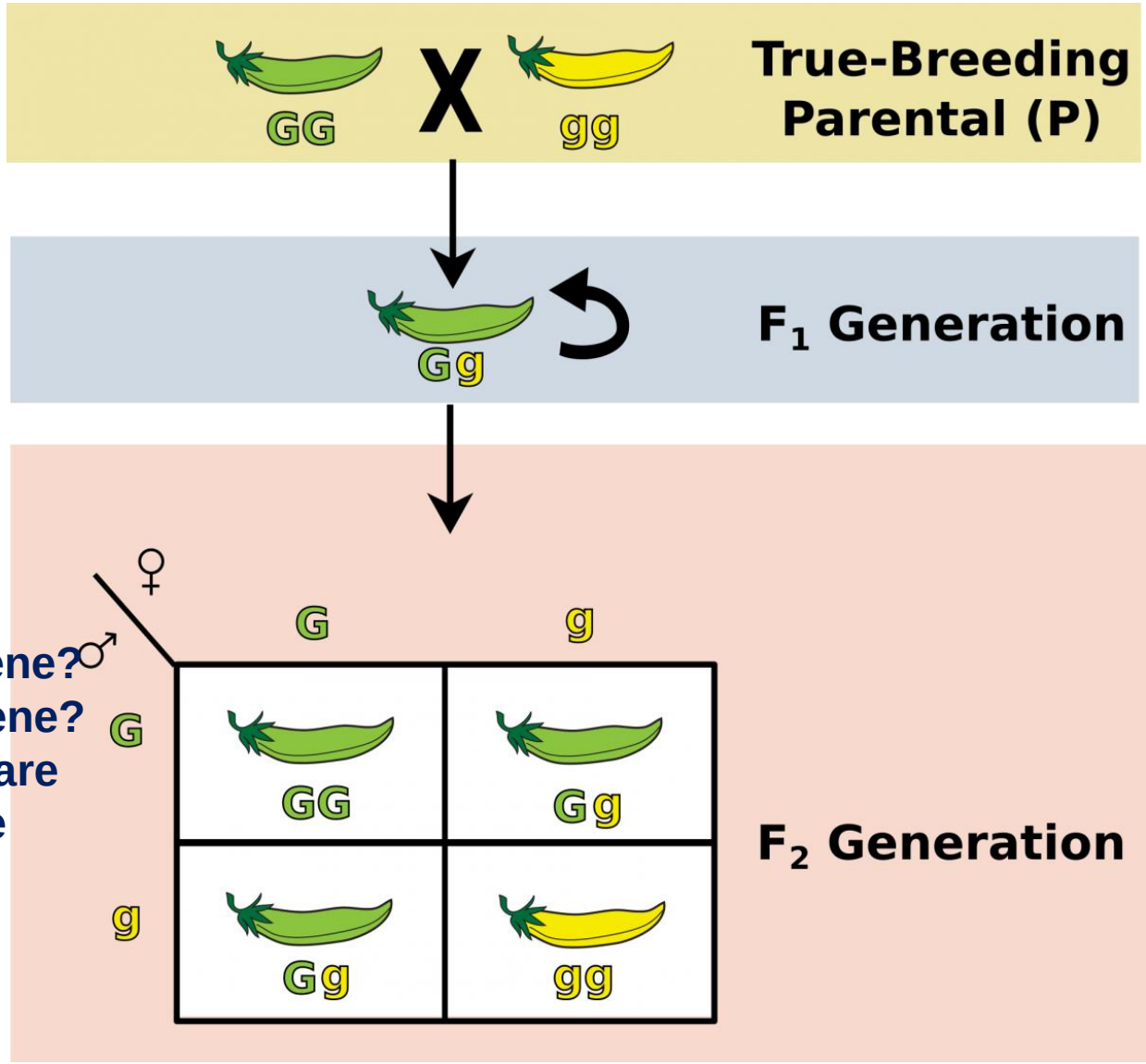
1. The **Punnett square** is a diagram that is used to predict an outcome of a particular cross or breeding experiment.
2. It is named after Reginald C. **Punnett**, who devised the approach.
3. The diagram is used by biologists to determine the probability of an offspring having a particular genotype.
4. **Homozygous (RR) or (rr)**
5. **Heterozygous (Rr)**



Q1. How many Homozygous in F2?
Q2. How many Heterozygous in F2?
Q3. What are they for each Q1 & Q2?

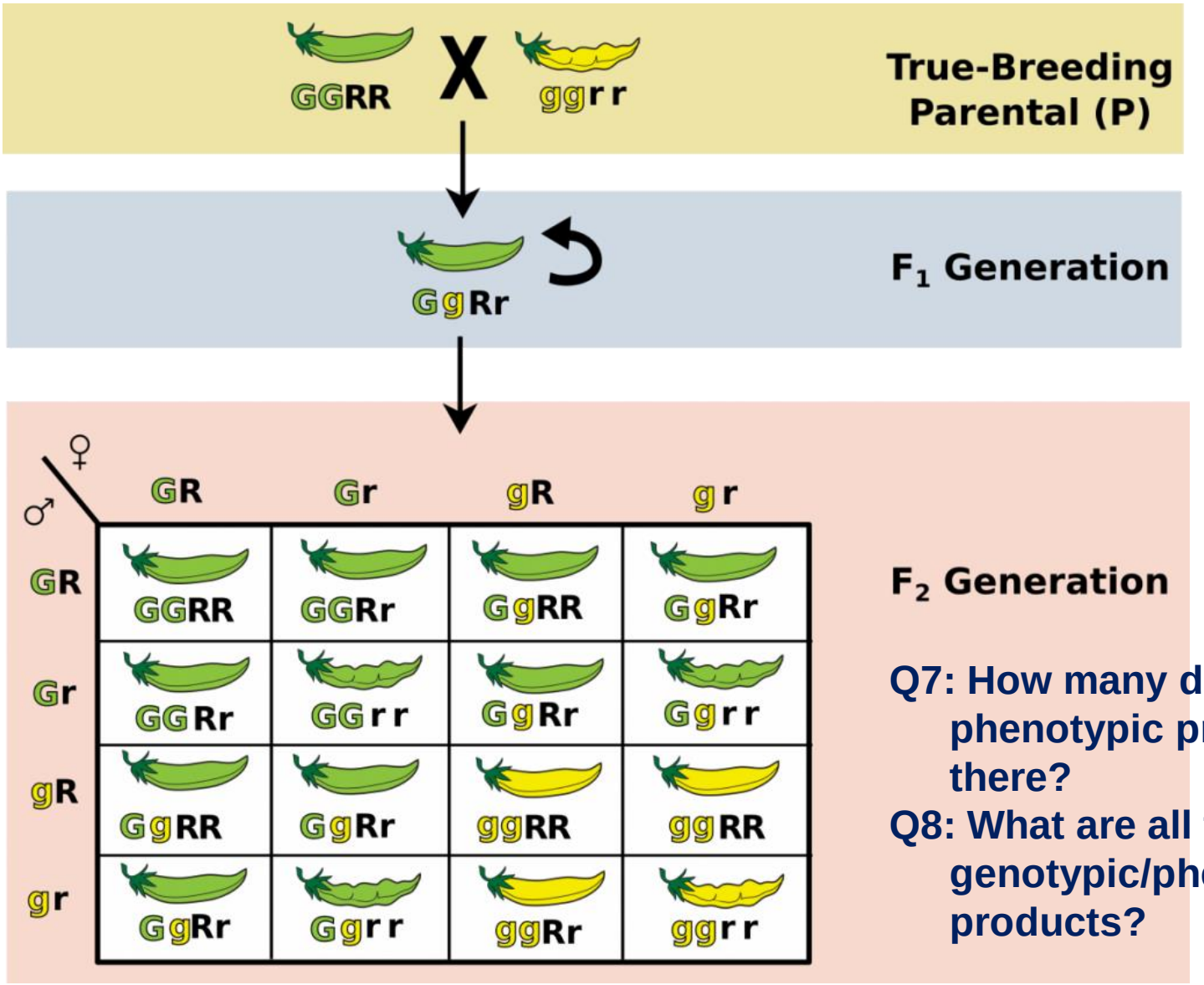
Punnett Square: OneTrait Cross

Dominant (G) versus
Recessive (g) genes



Q4: What is a dominant gene?
Q5: What is a recessive gene?
Q6: In this example, what are the dominant/recessive genes?

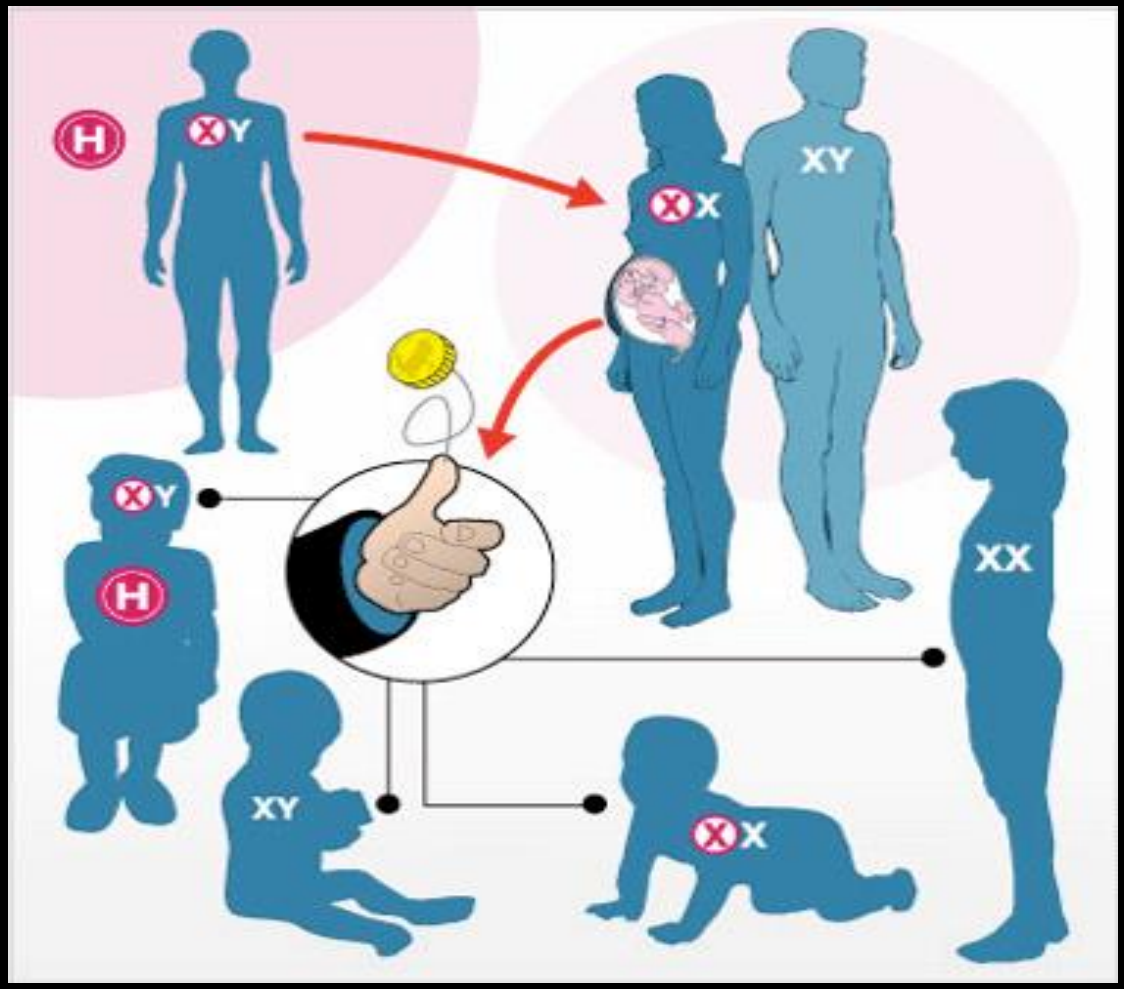
Punnett Square: The Two Trait Cross (Dihybrid Cross)



Q7: How many different phenotypic products are there?

Q8: What are all the different genotypic/phenotypic products?

Chromosomal Inheritance



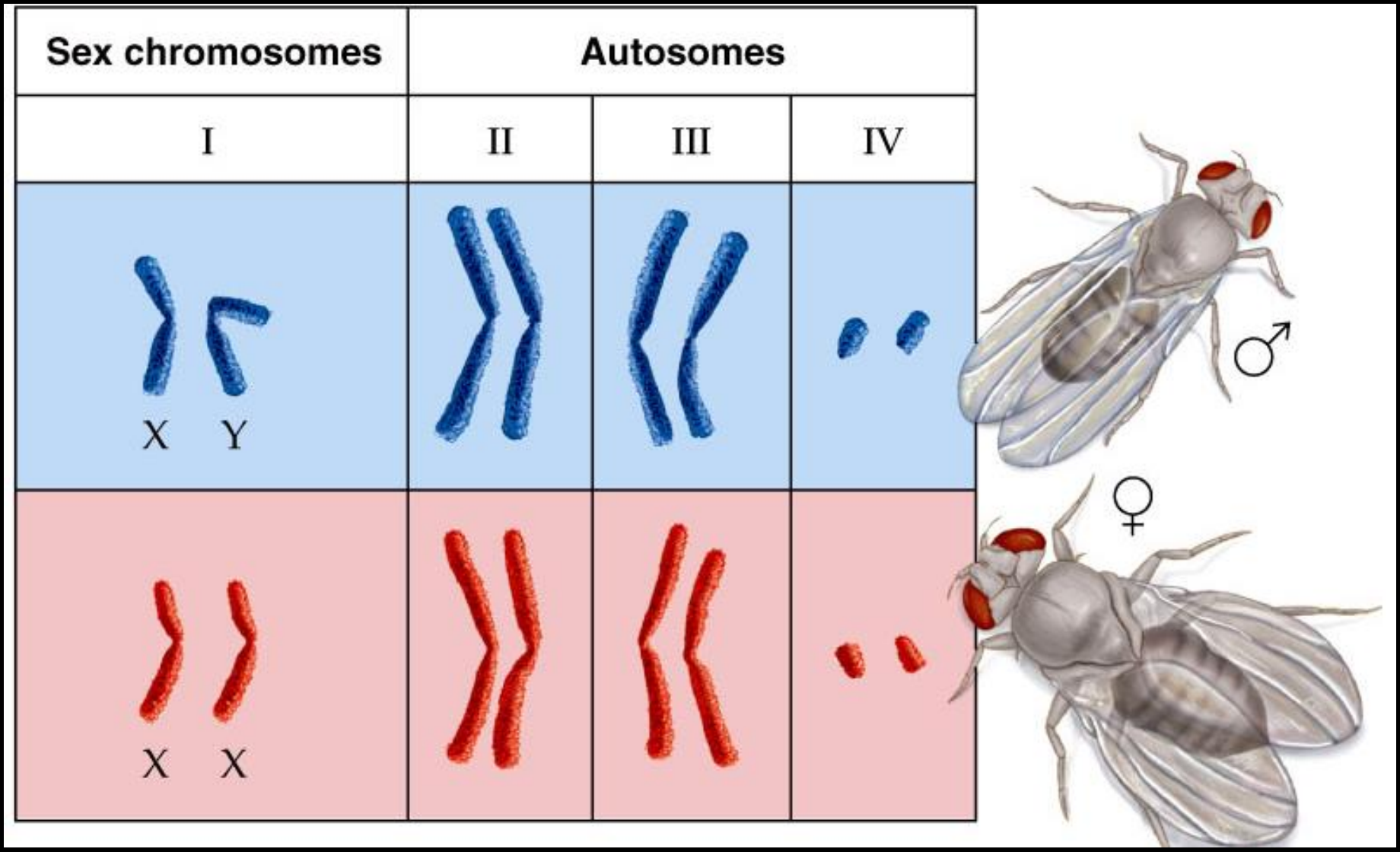
Chromosomes

- …are the physical basis of inheritance
- **Locus** – the physical location of a gene on a chromosome (plural = loci)
- Mendel's laws of segregation and independent assortment are because CHROMOSOMES segregate and assort independently in meiosis

Chromosomal Inheritance

- Humans are diploid (2 chromosomes of each type)
  - Humans have 23 different kinds of chromosomes
  - Arranged in 23 pairs of homologous chromosomes
  - Total of 46 chromosomes (23 pairs) per cell
- One of the chromosome pairs determines the sex of an individual (The **sex chromosomes**)
- The other 22 pairs of chromosomes are **autosomes**
- Autosomal chromosomes are numbered from smallest (#1) to largest (#22)
- The sex chromosomes are numbered as the 23rd pair

Drosophila Chromosomes



Sex Determination in Humans

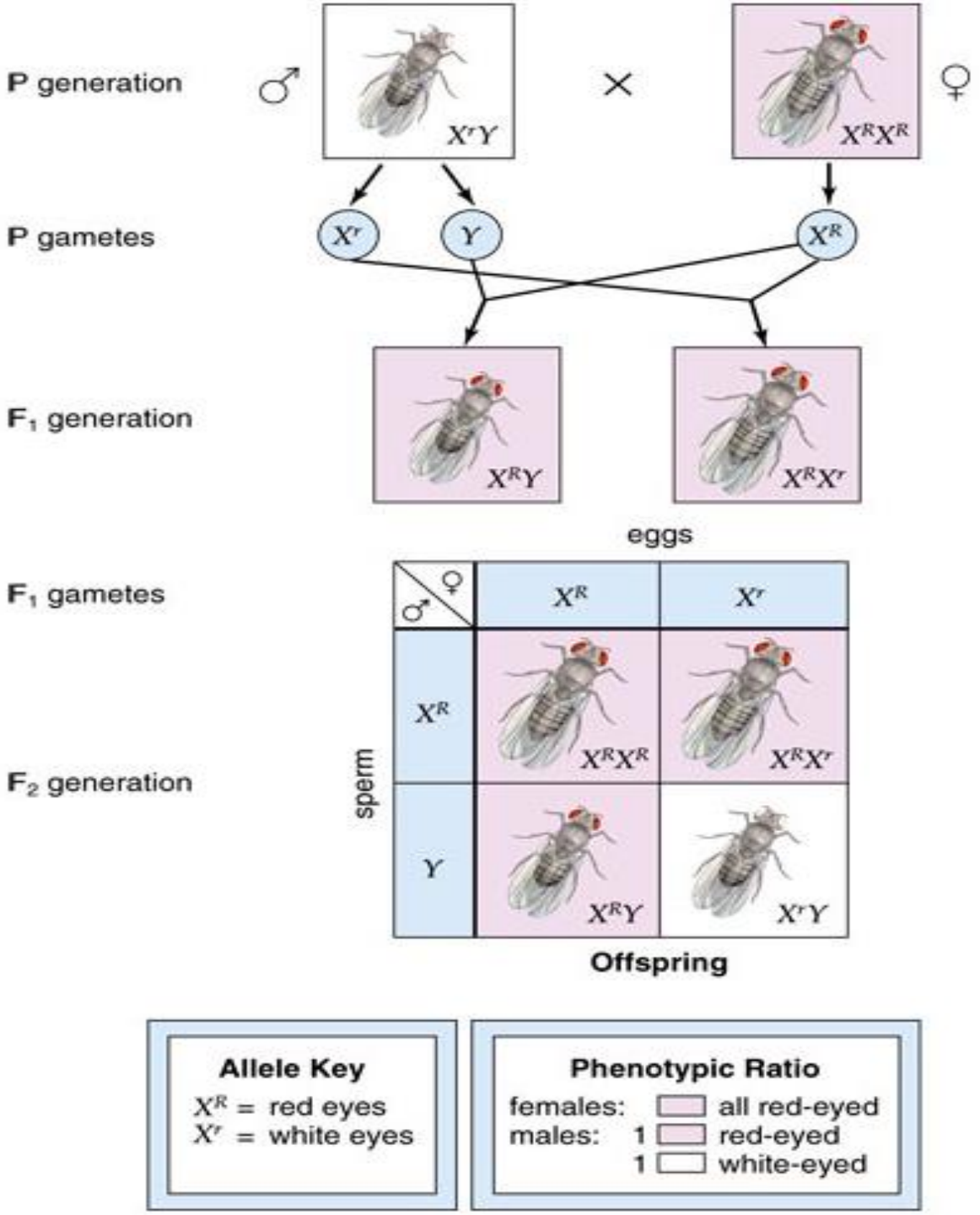
- Sex is determined in humans by allocation of chromosomes at fertilization
- Both sperm and egg carry one of each of the 22 autosomes
- The egg always carries the X chromosome as number 23
- The sperm may carry either and X or Y
  - If the **sperm donates an X** in fertilization, the zygote will be **female**
  - If the **sperm donates a Y** in fertilization, the zygote will be **male**
  - Therefore, the **sex of all humans is determined by the sperm** donated by their father

♂ \ ♀	X	X
X	XX	XX
Y	XY	XY

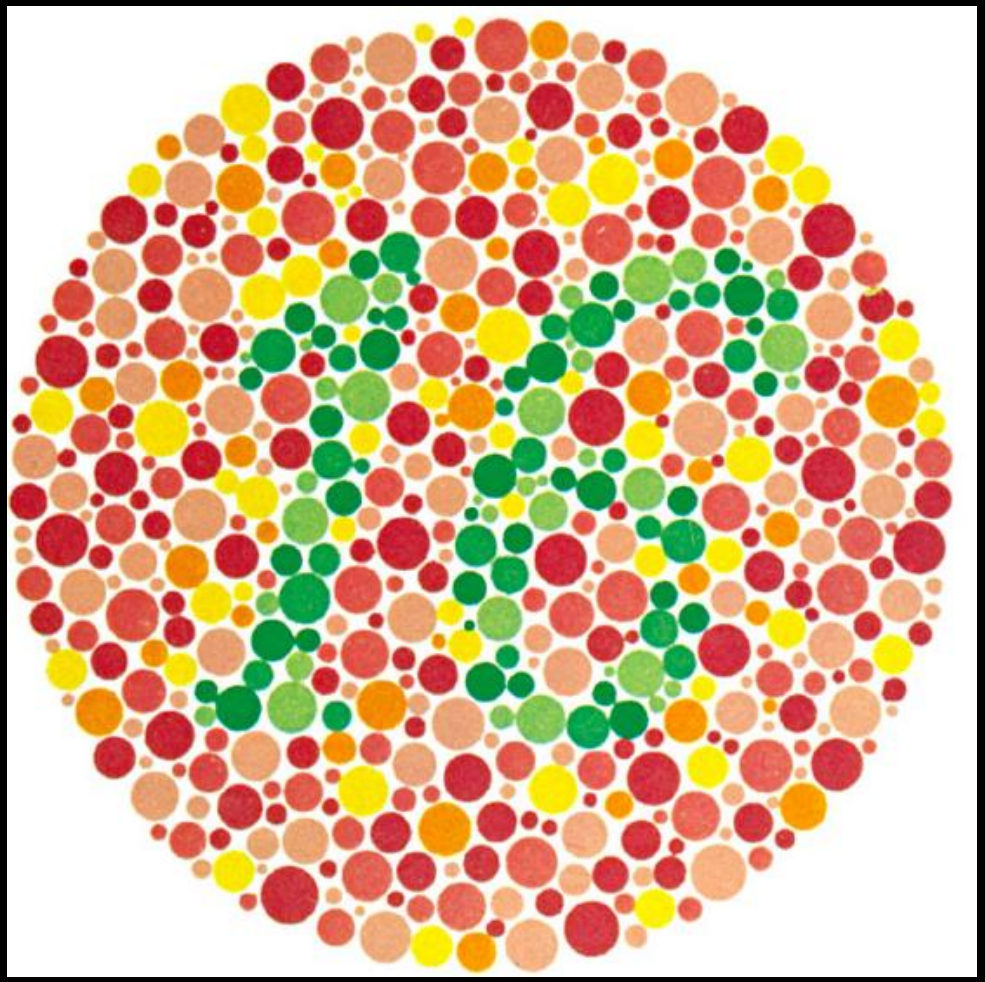
X-Linked Alleles

- Genes carried on autosomes are said to be autosomally linked
- Genes carried on the female sex chromosome (X) are said to be X-linked (or sex-linked)
- X-linked genes have a different pattern of inheritance than autosomal genes have
  - The Y chromosome is blank for these genes
  - Recessive alleles on X chromosome:
    - Follow familiar dominant/recessive rules in females (XX)
    - Are always expressed in males (XY), whether dominant or recessive
    - Males said to be **hemizygous** for X-linked genes

X-Linked Inheritance

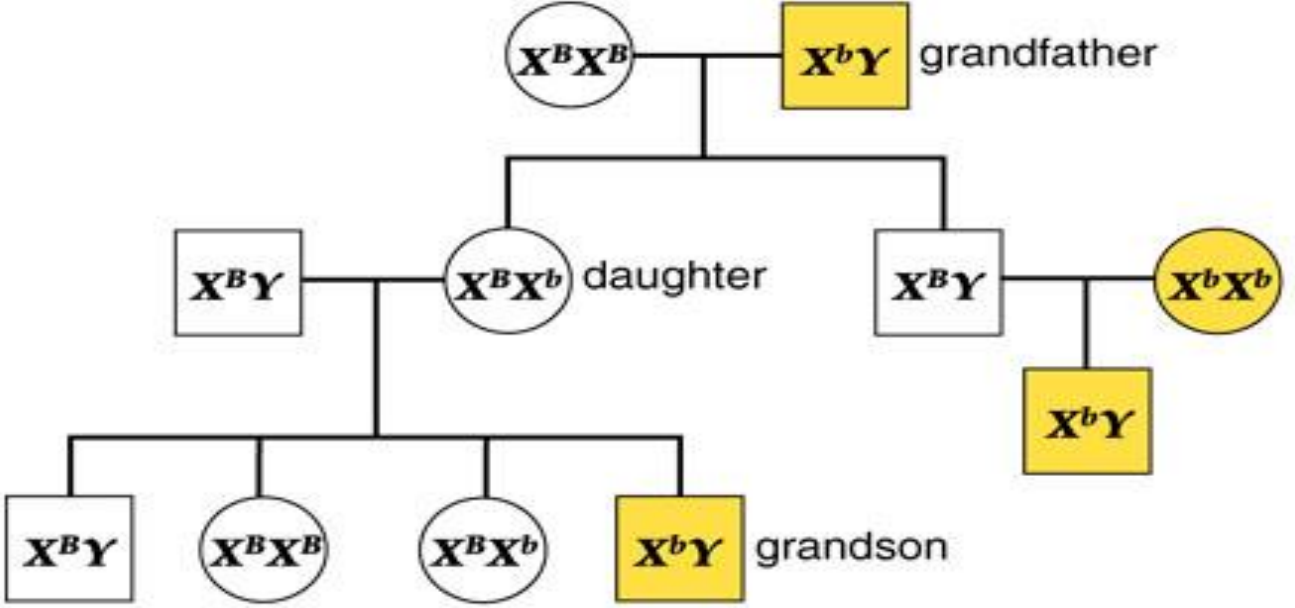


Human X-Linked Disorders: Red-Green Color Blindness



Red-Green Colorblindness Chart

X-Linked Recessive Pedigree



Key

X^BX^B = Unaffected female
X^BX^b = Carrier female
X^bX^b = Color-blind female
X^BY = Unaffected male
X^bY = Color-blind male

X-linked Recessive Disorders

- More males than females are affected.
- An affected son can have parents who have the normal phenotype.
- For a female to have the characteristic, her father must also have it. Her mother must have it or be a carrier.
- The characteristic often skips a generation from the grandfather to the grandson.
- If a woman has the characteristic, all of her sons will have it.

Human X-Linked Disorders: **Muscular Dystrophy**

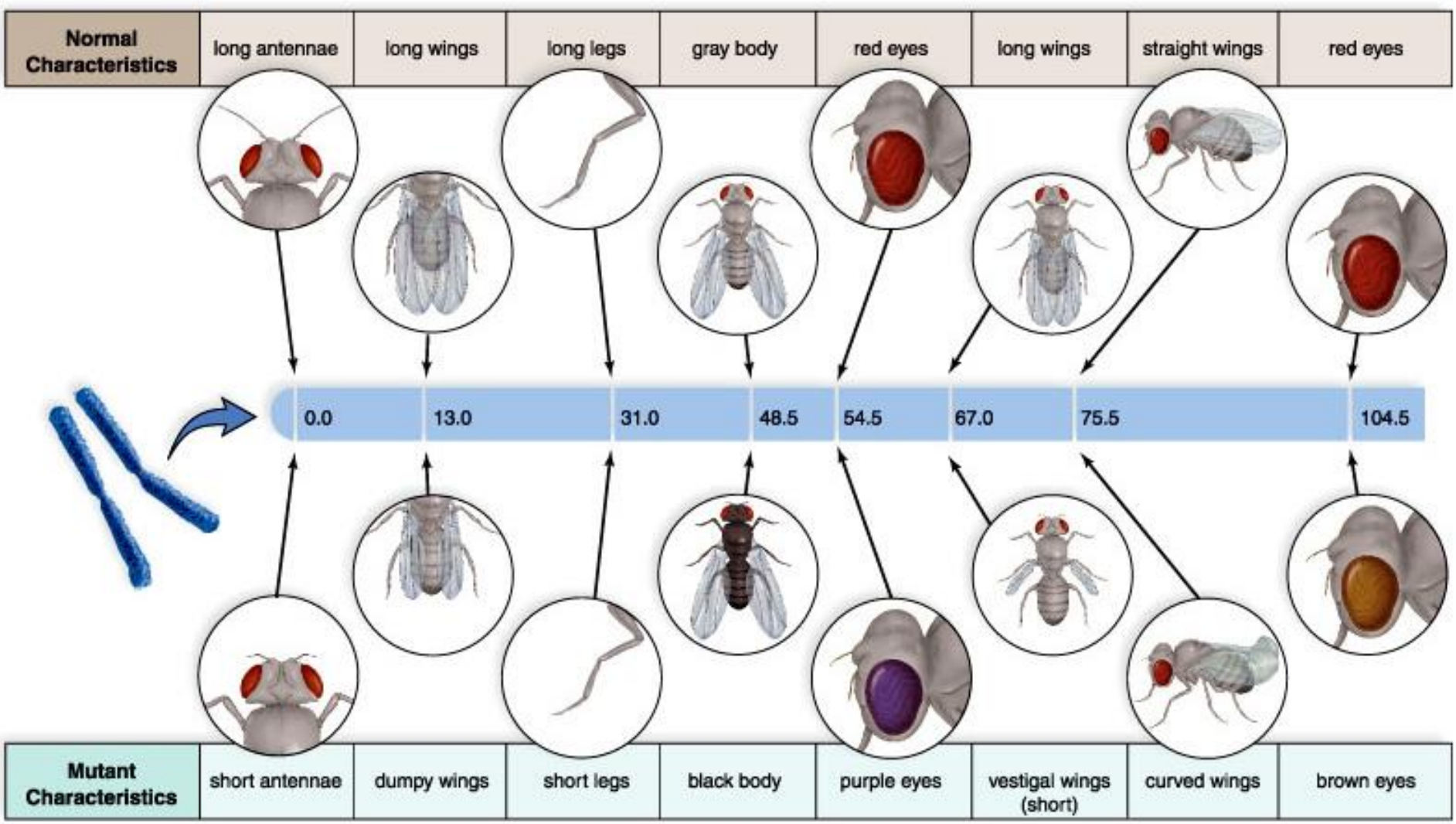
- Muscle cells operate by release and rapid sequestering of calcium
- Protein dystrophin required to keep calcium sequestered
- Dystrophin production depends on X-linked gene
- A defective allele (when unopposed) causes absence of dystrophin
  - Allows calcium to leak into muscle cells
  - Causes muscular dystrophy
- All sufferers male
  - Defective gene always unopposed in males
  - Males die before fathering potentially homozygous recessive daughters

Gene Linkage

- When several genes of interest exist on the same chromosome

- Such genes form a **linkage group**
  - Tend to be inherited as a block
  - If all genes on **same chromosome**:
    - Gametes of parent likely to have exact allele combination as gamete of either grandparent
    - **Independent assortment does not apply**
  - If all genes on separate chromosomes:
    - Allele combinations of grandparent gametes will be shuffled in parental gametes
    - Independent assortment working

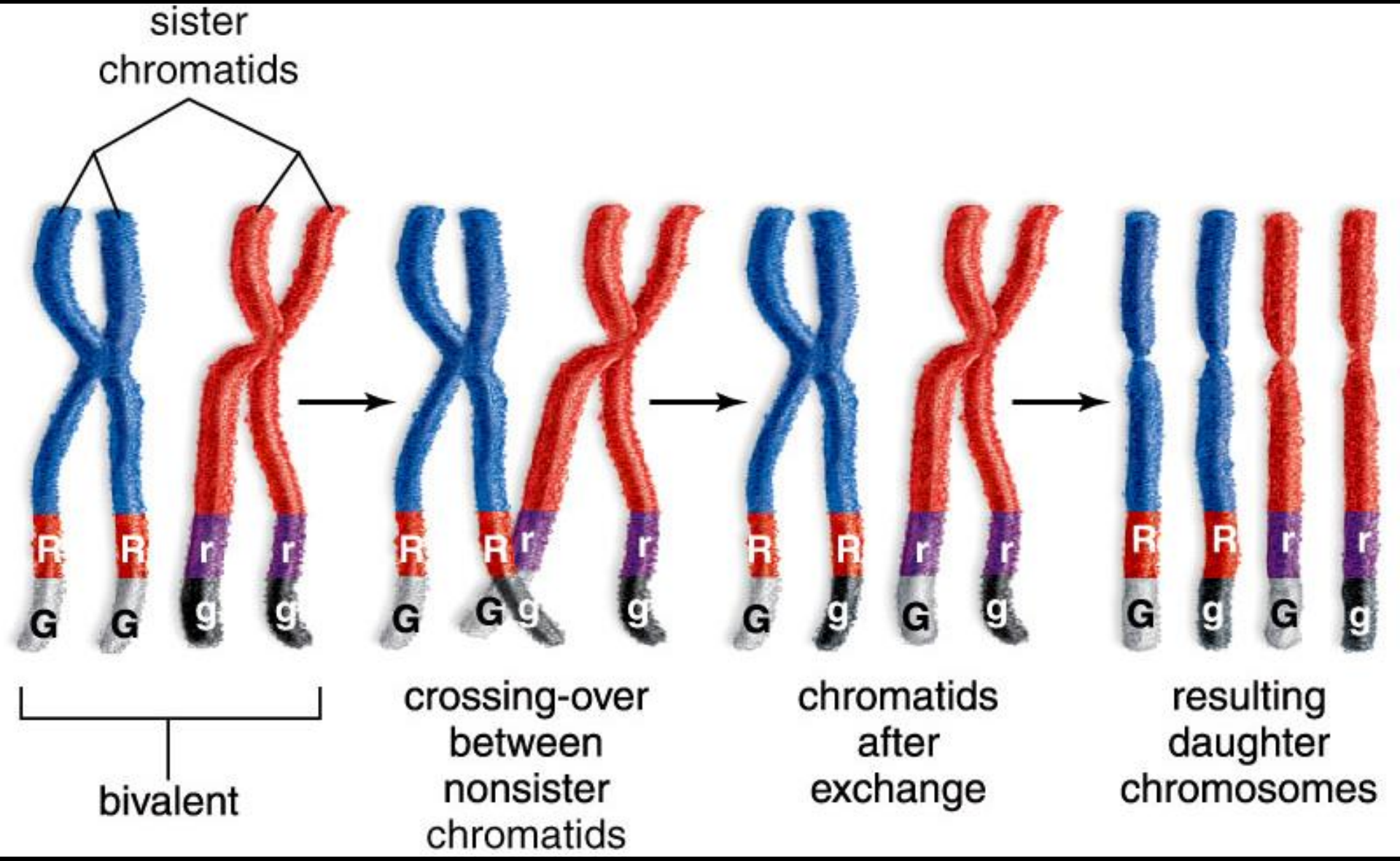
Linkage Groups



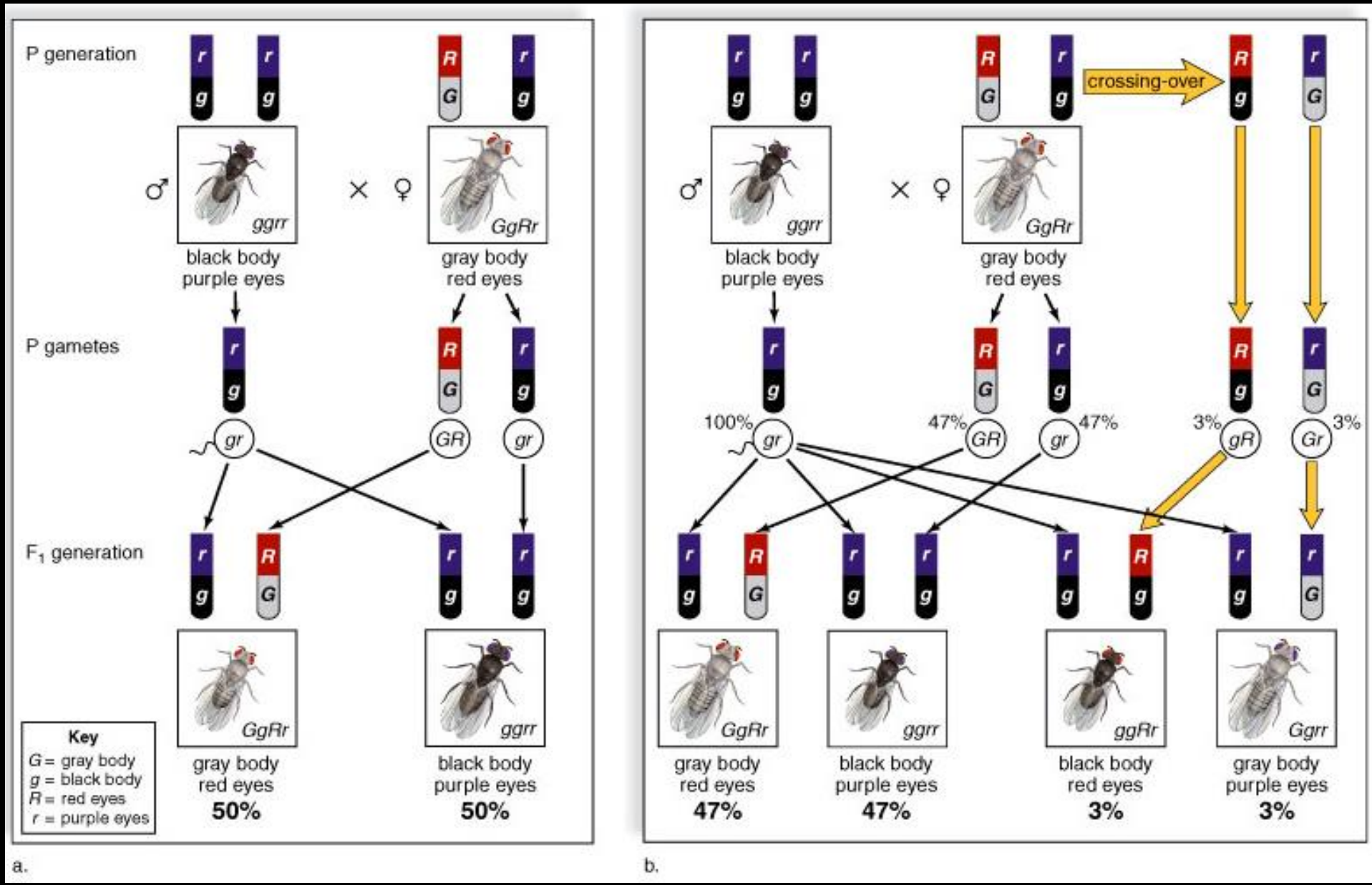
Constructing a Chromosome Map

- Crossing-over can disrupt a blocked allele pattern on a chromosome
- Affected by distance between genetic loci
- Consider three genes on one chromosome:
  - If one at one end, a second at the other and the third in the middle
    - Crossing over very likely to occur between loci
    - Allelic patterns of grandparents will likely to be disrupted in parental gametes with all allelic combinations possible
  - If the three genetic loci occur in close sequence on the chromosome
    - Crossing over very UNlikely to occur between loci
    - Allelic patterns of grandparents will likely to be preserved in parental gametes
- Rate at which allelic patterns are disrupted by crossing over:
  - Indicates distance between loci
  - Can be used to develop linkage map or genetic map of chromosome

Crossing Over



Complete vs. Incomplete Linkage



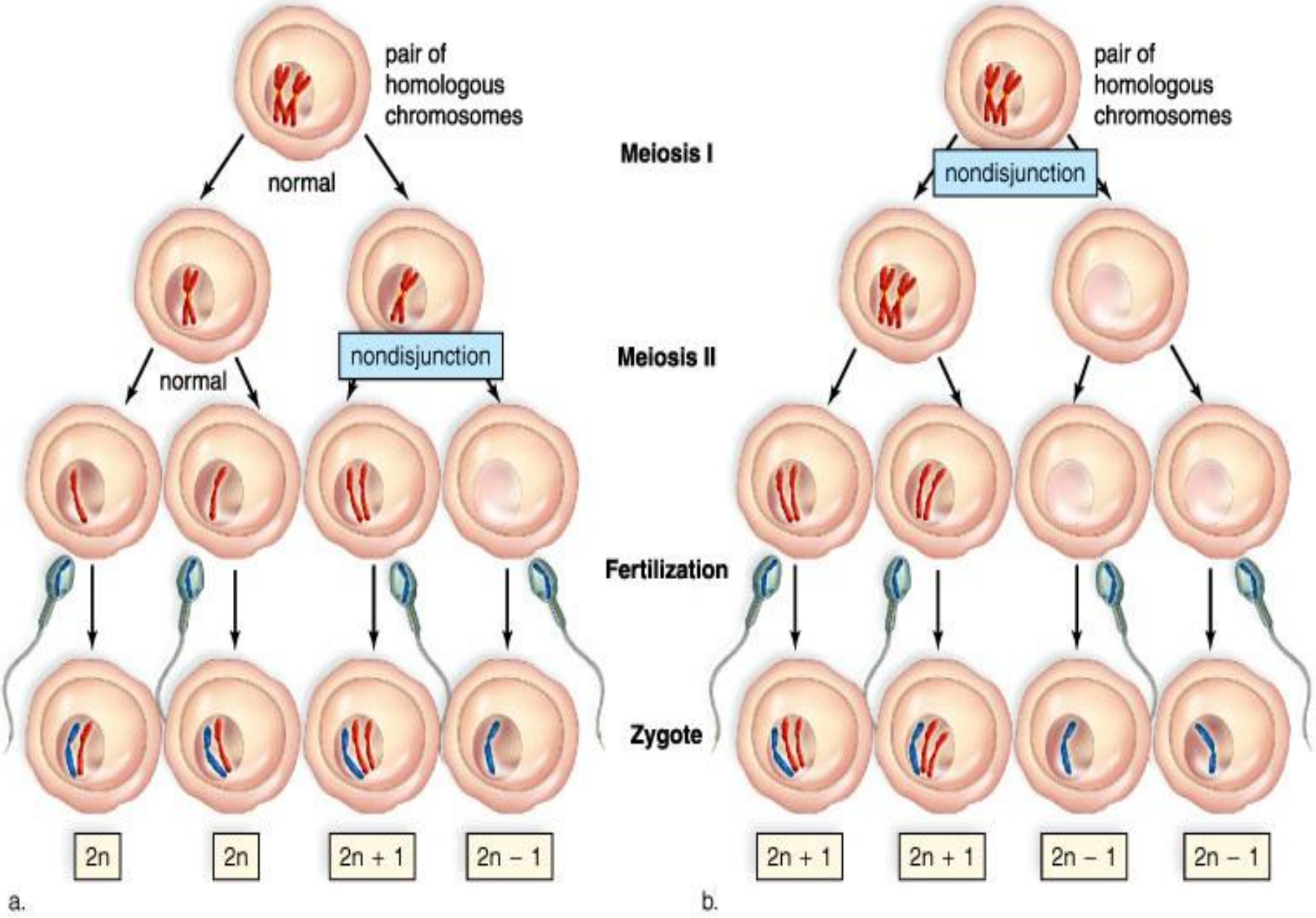
Chromosome Number: Aneuploidy

- **Monosomy** (2*n* - 1)
  - Diploid individual has only one of a particular chromosome
  - Caused by failure of synapsed chromosomes to separate at Anaphase (nondisjunction)
- **Trisomy** (2*n* + 1) occurs when an individual has three of a particular type of chromosome
  - Diploid individual has three of a particular chromosome
  - Also caused by nondisjunction
  - This usually produces one monosomic daughter cell and one trisomic daughter cell in meiosis I
  - Down syndrome is trisomy 21

Chromosome Number: Polyploidy

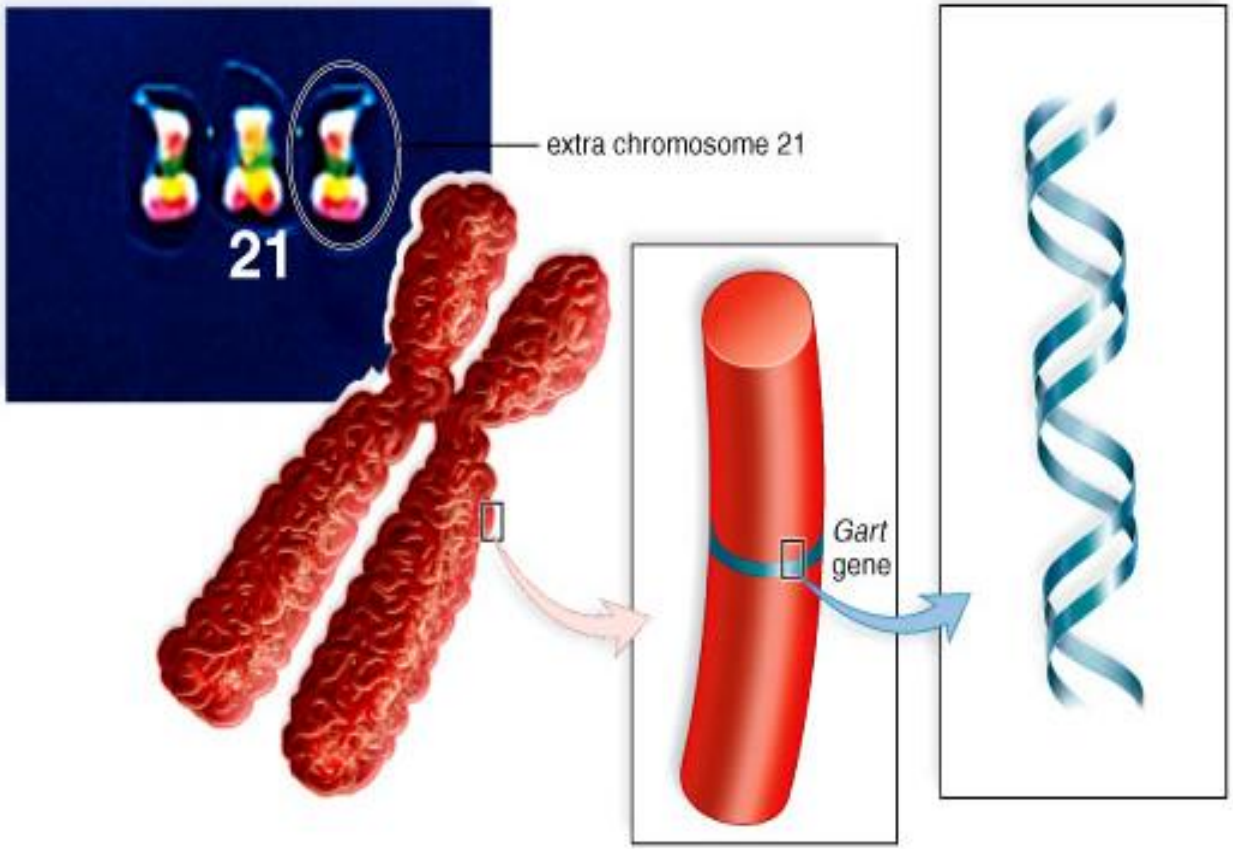
- **Polyploidy**
  - Occurs when eukaryotes have **more than *2n*** chromosomes
  - Named according to number of complete sets of chromosomes
  - Major method of speciation in **plants**
    - Diploid egg of one species joins with diploid pollen of another species
    - Result is new tetraploid species that is self-fertile but isolated from both “parent” species
    - Some estimate 47% of flowering plants are polyploids
  - Often **lethal** in higher animals

Nondisjunction



Trisomy 21

a.

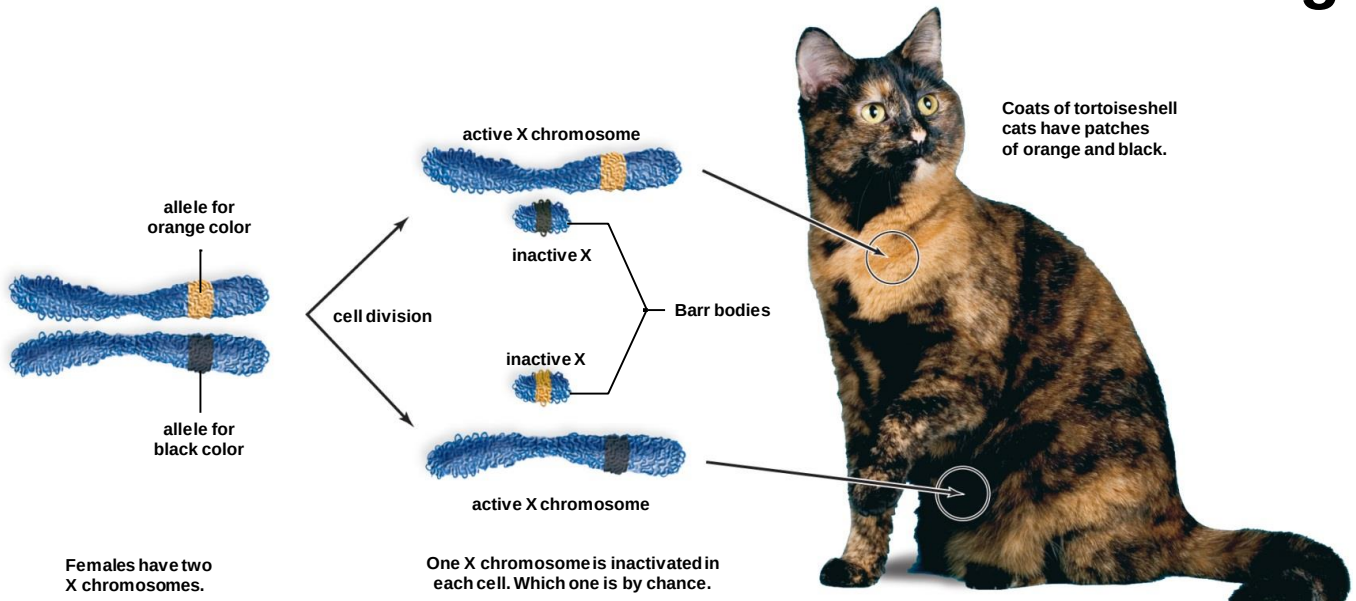


b.

Chromosome Number:
Abnormal Sex Chromosome Number

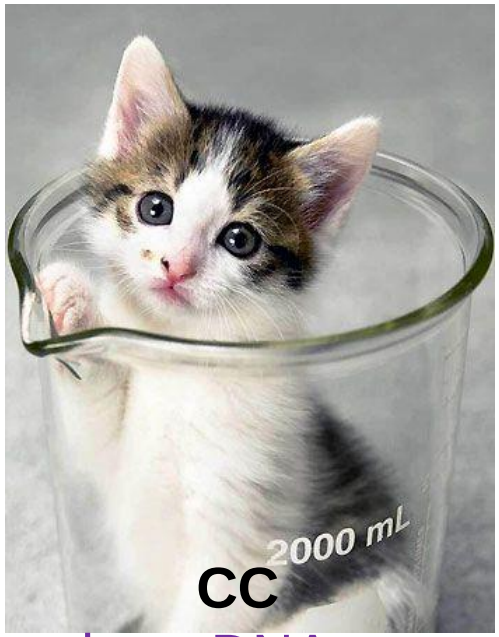
- Result of inheriting too many or too few X or Y chromosomes
- Caused by nondisjunction during oogenesis or spermatogenesis
- = Aneuploidy of sex chromosomes:
  - XXY = Klinefelter Syndrome - affects male physical and cognitive development
  - XO = Turner Syndrome - affects female physical and sometimes cognitive development
  - XXX =Poly-X females - totally normal
  - XYY = Jacob's Syndrome - mostly normal

Chromatin Structure: Gene regulation



Epigenetics -heritable changes in gene expression or cellular phenotype caused by mechanisms other than changes in the underlying DNA sequence

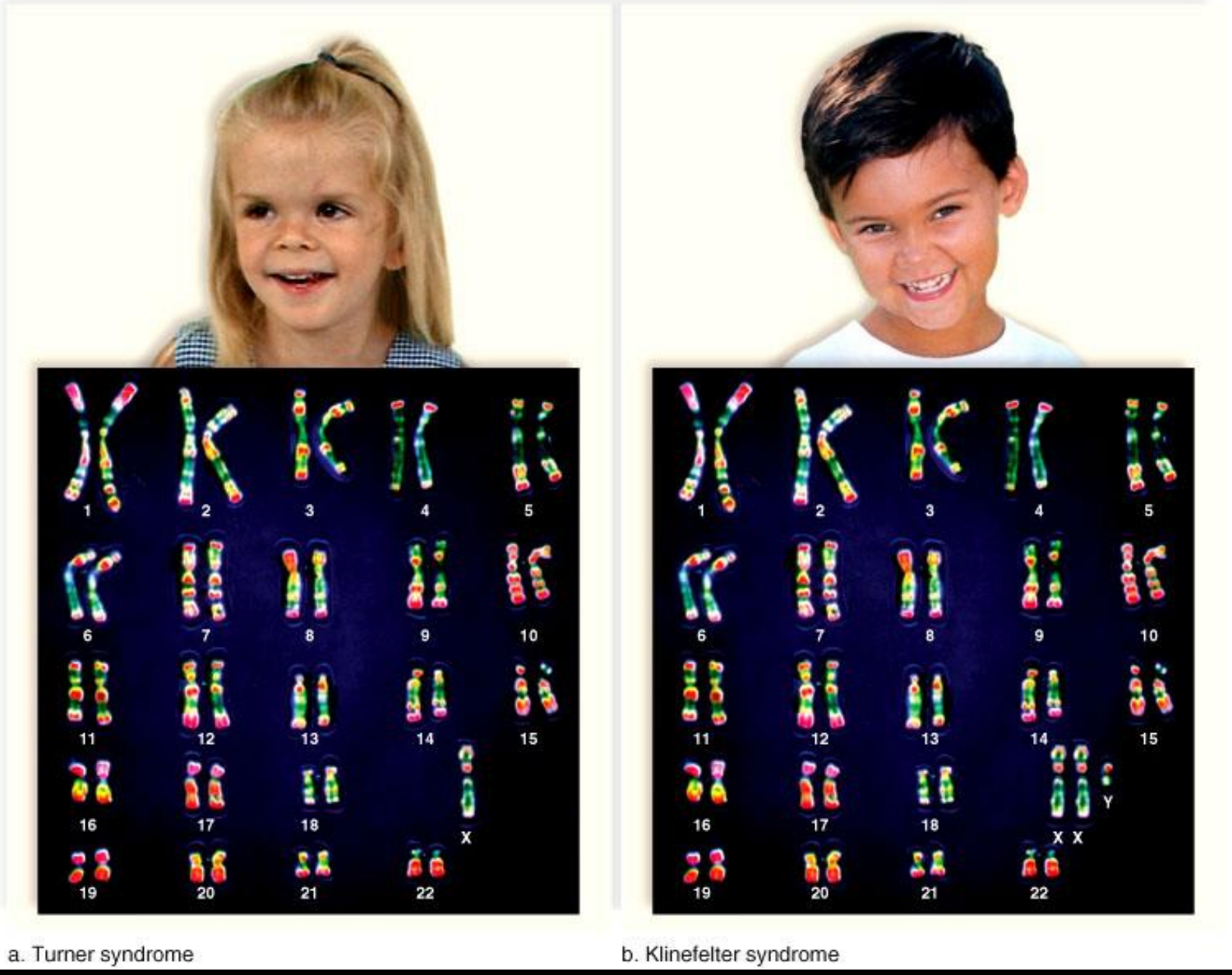
Rainbow



Can male cats be Calico?

Cute cats care about DNA compaction

Turner and Klinefelter Syndromes



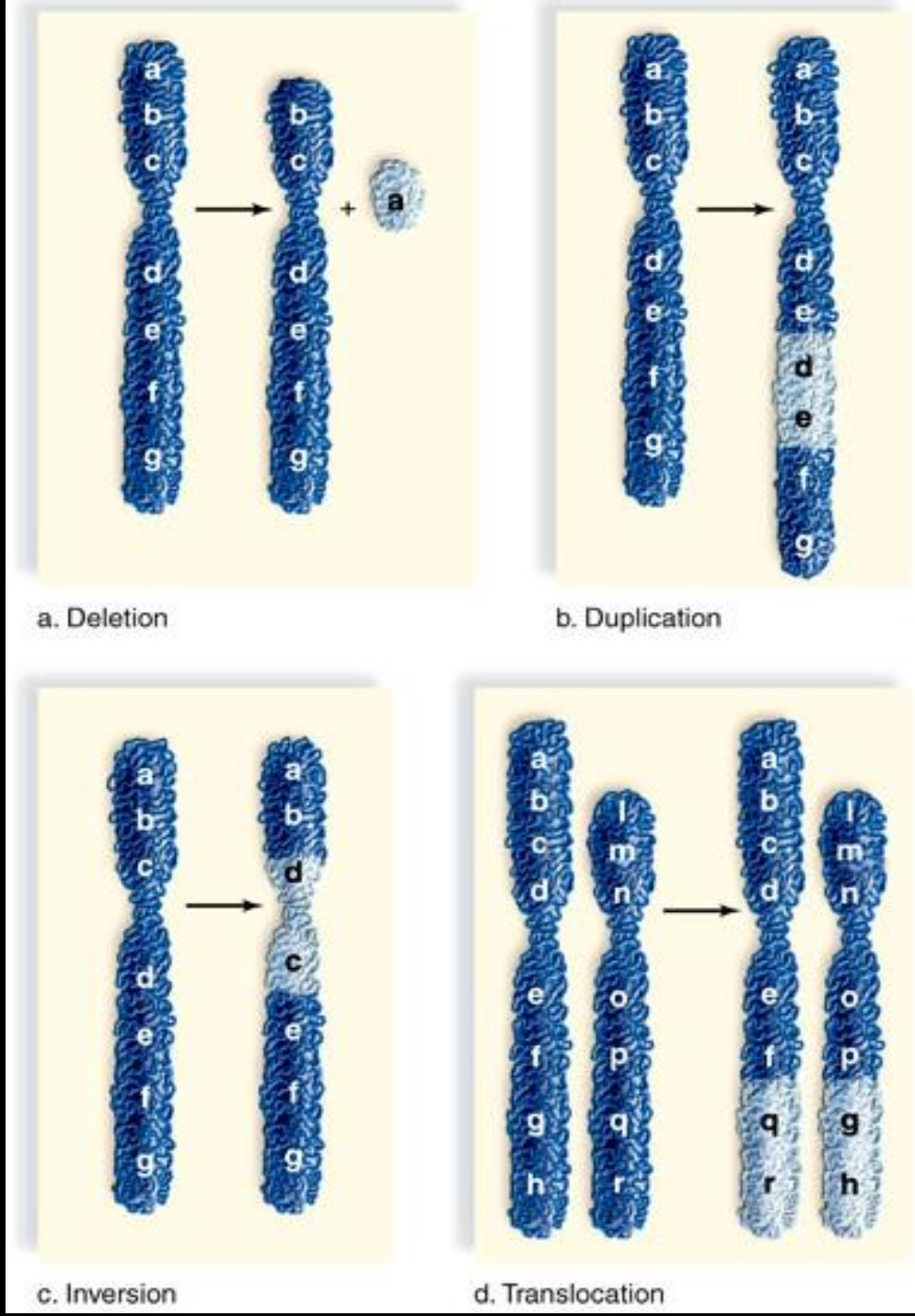
a. Turner syndrome

b. Klinefelter syndrome

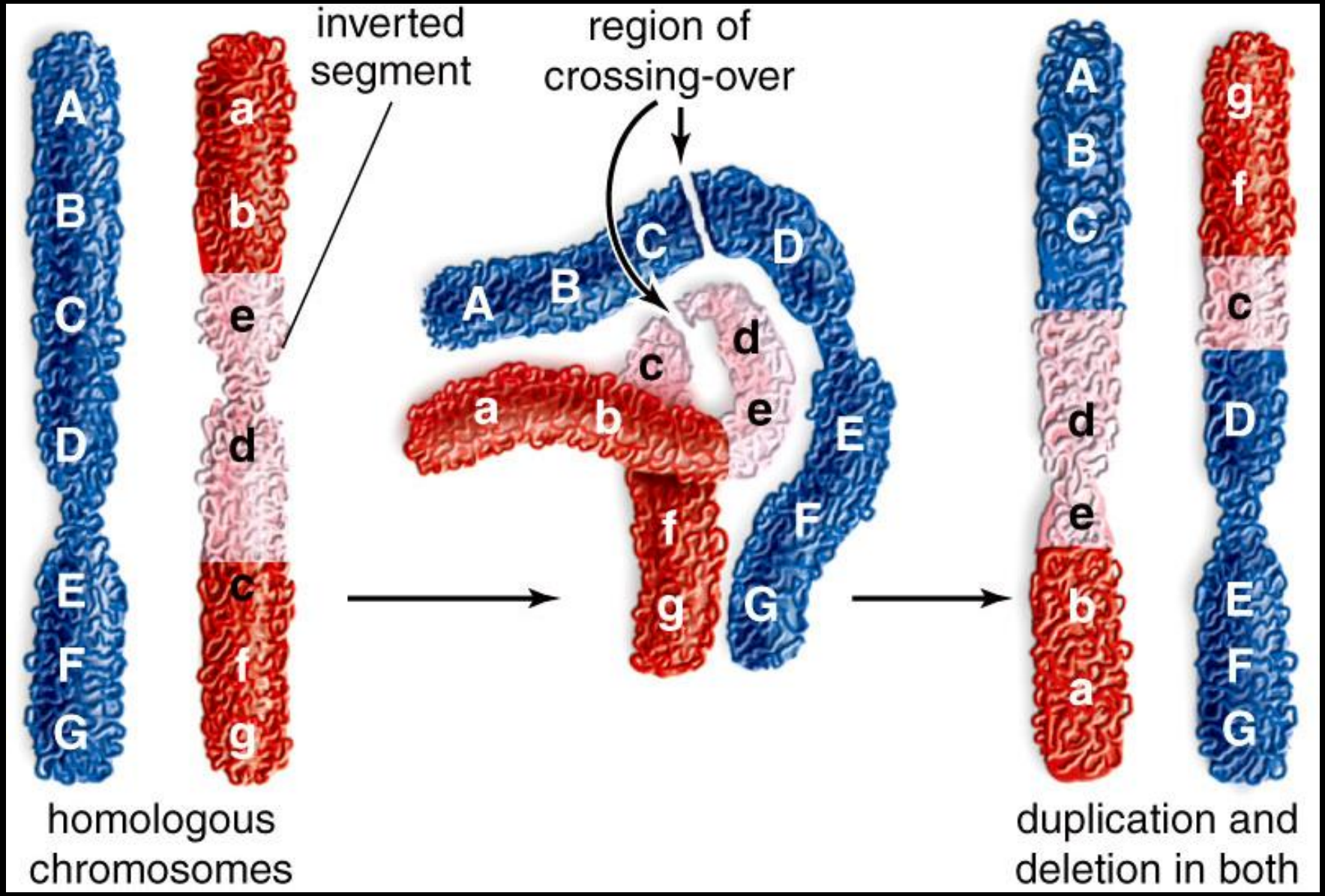
Altered Chromosome structure

- Physical breaks and damage to chromosomes can occur
- **Deletion** – part of a chromosome is completely LOST
- **Duplication** – " " " " accidentally repeated
- **Inversion** – " " " " accidentally placed BACKWARDS
- **Translocation** – " " " breaks off and attaches to another
- **Reciprocal translocation** – is a mutual "double switch" of chromosome fragments

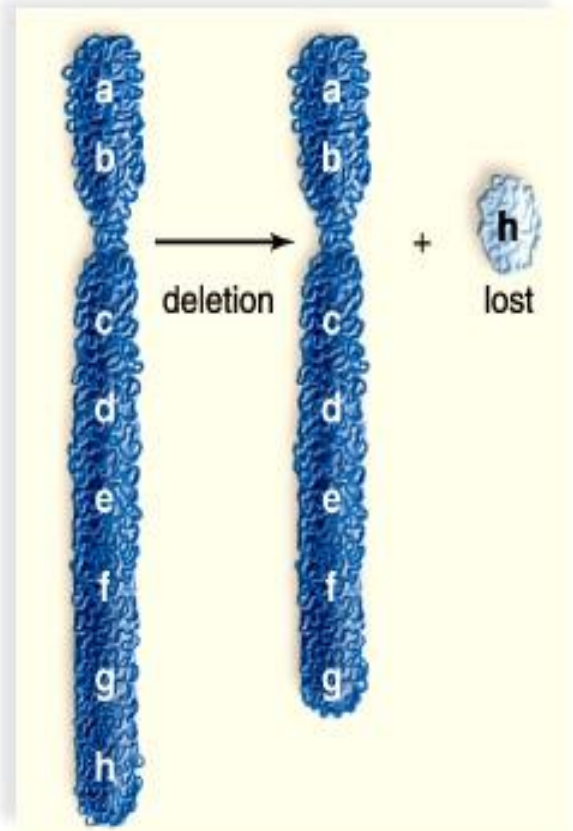
Altered Chromosome structure



Inversion Leading to Duplication and Deletion



Williams Syndrome – Chromosome 7q



a.

b.

Williams syndrome is a developmental disorder that affects many parts of the body. This condition is characterized by mild to moderate intellectual disability or learning problems, unique personality characteristics, distinctive facial features, and heart and blood vessel (cardiovascular) problems.

Questions?