

### **Mendel's Laws of Heredity**

Law of Segregation: During gamete formation, two alleles of a homologous pair of chromosomes segregate from each other so each gamete only contains 1 allele for each gene.

Law of Independent Assortment: Alleles of a gene pair segregate independently of other gene pairs. The segregation of one pair of alleles does not affect the segregation of another pair of alleles.

\*except when genes are linked on a chromosome

### **Genotypes**

homozygous = having two copies of the same allele at a locus

heterozygous = having two different alleles at a locus

### **Phenotypes**

dominant = only needing 1 allele to show the phenotype

recessive = needing 2 alleles to show the phenotype

codominance = phenotypes of both alleles are expressed so heterozygous phenotype is different from either homozygous phenotype

### **Probabilities**

$P(A)$  = probability of drawing an A allele from an individual's genotype

Or Rule  $P(A \text{ or } B) = P(A) + P(B)$  for mutually exclusive events

And Rule  $P(A \text{ and } B) = P(A) \times P(B)$  for independent events

## **Inheritance Patterns**

### Autosomal Dominant

one mutated allele is enough to express the disease

Mendelian diseases are assumed to be rare, so affected individuals are probably heterozygotes

- vertical transmission of disease phenotype, no skipped generations
- equal number of affected males and females
- half the offspring of an affected parent will also be affected

### Autosomal Recessive

two copies of mutated allele are required to express the disease

carriers = individuals with one copy of the mutated allele

obligate carrier = individual who must be a carrier due to observed affected individuals in the pedigree

commonly seen from consanguineous relationships

- clustering of disease in siblings
- disease skips generations
- equal number of affected males and females

### X-linked Recessive

one mutated allele is enough to express the disease in males

affected males have 50% chance of receiving the mutated allele from carrier mothers

all daughters of carrier mothers are obligate carriers

- more affected males than females
- never passed from father to son
- passed from affected grandfather to half of grandsons through carrier mothers

### X-linked Dominant

one mutated allele is enough to express the disease

- half the offspring of affected mother will also be affected
- all daughters of affected fathers will also be affected
- never passed from father to son
- twice as many affected females than affected males

### Mitochondrial

mitochondrial genome is inherited only from mother through ovum

- all children of affected mothers are also affected
- no children of affected males will be affected

### Complex

doesn't follow Mendelian inheritance