**Mendel Notes 2019**

**Concept: Mendel used the scientific approach to identify two laws of inheritance**

Gregor Mendel

Modern genetics began in the mid-1800s in an abbey garden, where a monk named Gregor Mendel documented inheritance in peas. He used experimental method, used quantitative analysis, collected data & counted them, and was an excellent example of scientific method

Mendel bred pea plants, he cross-pollinated plants. Used true breeding parents (P)

P = parental

 Mendel raised seed & then observed traits (F1)

F = filial

He allowed offspring to self-pollinate & observed next generation (F2)

What did Mendel’s findings mean?

Traits come in alternative versions. Example: purple vs. white flower color

-Alternative forms of a trait are called \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

-different alleles vary in the sequence of nucleotides at the specific locus of a gene

some difference in sequence of A, T, C, G

Traits are inherited as discrete units

For each characteristic, an organism inherits 2 alleles, 1 from each parent

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ organism

-inherits 2 sets of chromosomes, 1 from each parent

-homologous chromosomes

-like having 2 editions of encyclopedia

-Encyclopedia Britannica

-Encyclopedia Americana

Some traits mask others

-purple & white flower colors are separate traits that do not blend

purple x white ≠ light purple

purple masked white

-\_\_\_\_\_\_\_\_\_\_\_\_ allele masks other alleles by producing a functional protein which affects characteristic

-\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ allele have no noticeable effect as it makes a malfunctioning protein

Genotype vs phenotype

Difference between how an organism “looks” & its genetics

-phenotype is the description of an organism’s trait

-genotype is the description of an organism’s genetic makeup

Making crosses: we can represent alleles as letters (P or p). True breeding purple-flower peas 🡪 PP, True-breeding white-flower peas 🡪 pp

Genotypes:

Homozygous = same alleles = PP, pp

Heterozygous = different alleles = Pp

Phenotype vs genotype

2 organisms can have the same phenotype but have different genotypes (PP, Pp)

Test cross

Breed the dominant phenotype-with the unknown genotype-with a homozygous recessive (pp) to determine the identity of the unknown allele

Mendel’s 1st law of heredity (Law of Segregation)

 -during meiosis, alleles segregate (homologous chromosomes separate)

 -each allele for a trait is packaged into a separate gamete

Which stage of meiosis creates the law of segregation? Metaphase 1

Monohybrid cross:

Some of Mendel’s experiments followed the inheritance of single characters

-Examples: flower color, seed color, \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ crosses

Dihybrid cross:

Other of Mendel’s experiments followed the inheritance of 2 different characters. Example: seed color \_\_\_\_\_ seed shape. These are dihybrid crosses.

If genes are on different chromosomes…how do they assort in the gametes? together or independently?

Mendel’s 2nd law of heredity

Law of independent \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ in which different loci (genes) separate into gametes independently

-non-homologous chromosomes align independently

-classes of gametes produced in equal amounts

-YR = Yr = yR = yr

-only true for genes on separate chromosomes or on same chromosome but so far apart that crossing over happens frequently

Which stage of meiosis creates the law of independent assortment? Metaphase 1. If genes are on same chromosome & close together will usually be inherited together. They rarely crossover separately and they are “linked”

**Concept: Linked genes tend to be inherited together because they are located near each other on the same chromosomes**

-Sometimes genes on the same chromosomes stay together during assortment and move as a group. The group of genes is considered linked and tends to be inherited together. For example, the genes for flower color and pollen shape are linked on the same chromosomes and show up together. Since linked genes are found on the same chromosome, they cannot segregate independently, this violates the law of independent assortment.

-Lets pretend that height and color genes are linked. A heterozygote for both traits still have two alleles for height (T or t) and two alleles for color (G and g). However, because height and color are located on the same chromosome, the allele for height and the allele for color are physically linked. For example, maybe the heterozygote has one chromosome with Tg and one chromosome with tG. When gametes formed, the T and g will travel together, and the t and G will travel together and be packaged into a gamete together. So, in the unlinked dihybrid shown earlier there were four possible gamete combinations (TG, Tg, tG, tg), but now there are only two (Tg and tG). The only way to physically separate linked alleles is by crossing over. If a crossover even occurs between the linked genes, then recombinant gametes can occur.

-If the genes were unlinked, then the four gametes (TG, Tg, tG, tg) would be equally likely. However, if certain combinations of alleles are found more often in offspring, then this is a sign of possible linkage.

-A linkage map is a genetic map put together using crossover frequencies. Another unit of measurement, the map unit (also known as a centigram), is used to geographically relate genes on the basis of the frequencies. One map unit is equal to a 1 percent crossover frequency. A linkage map does not provide the exact location of genes, it gives only the relative location.

-Imagine that you want to determine the relative location of four genes: A, B, C, and D. You know that A crosses over with C 20 percent of the time, B crosses over with C 15 percent of the time, A crosses over with D 10 percent of the time, and D crosses over with B 5 percent of the time. From this information you can determine the sequence. Gene A must be 20 units from gene C. Gene B must be 15 units from C, but B could be 5 or 35 units from B, you can determine that B must be 5 units from A as well, if A is also to be 10 units from D. This gives you the sequence of genes as ABDC.

Review: Mendel’s laws of heredity:

Law of segregation

-monohybrid cross; single trait where each allele segregates into separate gametes and is established by Metaphase 1

Law of independent assortment

dihybrid (or more) cross includes 2 or more traits; genes on separate chromosomes assort into gametes independently. Established by Metaphase 1

Extending Mendelian genetics

Mendel worked with a simple system. He used peas are genetically simple. Where most traits are controlled by a single gene Each gene has only 2 alleles, 1 of which is completely dominant to the other

The relationship between genotype & phenotype is rarely that simple

**Concept: The laws of probability govern Mendelian inheritance**

Understanding how to predict offspring of genetic crosses involves familiarity with the basic laws of probability. There are two laws that you will use directly in solving genetic problems.

-The rule of multiplication: When calculating the probability that two or more independent events will occur together in a specific combination, multiply the probabilities of each of the two events. Thus, the probability of a coin landing face up two times in two flips is ½ x ½ = ¼. IF you cross two organisms with the genotypes AABbCc and AbBbCc, the probability of an offspring having the genotype AaBbcc is ½ x ½ x ¼ = 1/16

-The rule of addition: When calculating the probability that any of two or more mutually exclusive events will occur, you need to add together their individual probabilities. For example, if you are tossing a die, what is the probability that it will land on either the side with 4 spots or the side with 5 spots? (1/6 + 1/6 = 2/6=1/3)

**Concept: Inheritance patterns are often more complex than predicted by simple Mendelian genetics**

Incomplete dominance

Heterozygote shows an intermediate \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ phenotype

 -example: RR = red flowers

rr = white flowers

Rr = pink flowers

make 50% less color

Co-dominance

2 alleles affect the phenotype equally & separately. They are \_\_\_\_\_\_\_\_ blended phenotype

-example: ABO blood groups

-3 alleles (IA, IB, i)

-IA & IB alleles are co-dominant to each other

-both antigens are produced

-both IA & IB are dominant to i allele

-produces glycoprotein antigen markers on the surface of red blood cells

Blood Compatibility:

Matching compatible blood groups which is critical for blood transfusions

A person produces antibodies against antigens in foreign blood

-wrong blood type

-donor’s blood has A or B antigen that is foreign to recipient

-antibodies in recipient’s blood bind to foreign molecules

-cause donated blood cells to clump together

-can kill the recipient

Pleiotrophy

Most genes are \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ where one gene affects more than one phenotypic character

-can have wide-ranging effects due to a single gene

-dwarfism (achondroplasia)

-gigantism (acromegaly)

Epistasis

One gene completely masks another gene

-coat color in mice = 2 separate genes

-C,c:
pigment (C) or no pigment (c)

-B,b:
more pigment (black=B) or less (brown=b)

-cc = albino, no matter B allele

9:3:3:1 becomes 9:3:4

2 genes: (E,e) & (B,b): pigment (E) or no pigment (e) and pigment concentration: black (B) to brown (b)

Polygenic inheritance

Some phenotypes determined by additive effects of 2 or more genes on a single character

-phenotypes on a continuum

-human traits include skin color, height, weight, eye color, intelligence, behaviors

**Concept: Sex-linked genes exhibit unique patterns of inheritance**

Sex linked Traits

Genes are on \_\_\_\_\_\_\_\_\_\_\_\_ chromosomes. They are NOT on autosomal chromosomes (Chromosome pairs 1-22)

-first discovered by T.H. Morgan at Columbia U. Drosophila breeding (fruit flies)

Genetics of Sex

In humans & other mammals, there are 2 sex chromosomes: X & Y

-2 X chromosomes and they develop as a female: XX. There is gene redundancy like autosomal chromosomes

-an X & Y chromosome: they develop as a male: XY. There is no redundancy

Genes on sex chromosomes

Y chromosome has few genes other than SRY

-sex-determining region

-master regulator for maleness

-turns on genes for production of male hormones

-many effects = pleiotropy!

X chromosome has other genes/traits beyond sex determination

Mutations can result in hemophilia, Duchenne muscular dystrophy, color-blindness

Sex-linked traits usually mean “\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_”. There are more than 60 dz traced to genes on the X chromosome.

Map of Human Y chromosome: (SRY) Sex determining Region Y

X-linked

-follow the X chromosomes

-males get their X from their mother

-trait is never passed from father to son

Y-linked

-very few genes / traits

-trait is only passed from father to son

-females cannot inherit trait

**Concept: Many human traits follow Mendelian patterns of inheritance**

Pedigrees

A pedigree is a diagram that shows the relationship between parents and offspring across two or more generations. In a typical pedigree circles represent females and squares represent males. White open circles or squares indicate that the individual did not or does not express a particular trait, whereas the shaded ones indicate that the individual expresses or expressed that trait. Through the patterns they reveal, pedigrees can help determine the genome of individuals that comprise them; pedigrees can also help predict the genome of future off spring.

Recessive inherited disorders: (Cystic fibrosis, Tay-Sachs, Sickle Cell)

Dominant inherited disorders: (Huntington’s disease)

**Concept: Mendelian inheritance has it physical basis in the behavior of chromosomes**

The chromosome theory of inheritance states that genes have specific locations (loci) on chromosomes and that it is chromosomes that segregate and assort independently. It is important to connect this physical movement of chromosomes in meiosis to Mendel’s laws of inheritance

X-inactivation

Female mammals inherit 2 X chromosomes. One X becomes inactivated during embryonic development

condenses into compact object = Barr body, which X becomes Barr body is random

patchwork trait = “mosaic”

A Barr body is an X chromosome that is condensed and visible.

Nature vs Nurture

Phenotype is controlled by both environment & genes. Human skin color is influenced by both genetics & environmental conditions

How do we get from DNA to Trait?

Mechanisms of inheritance

What causes the differences in alleles of a trait?

-yellow vs. green color

-smooth vs. wrinkled seeds

-dark vs. light skin

-sickle cell anemia vs. no disease

What causes dominance vs. recessive?

Molecular basis of inheritance

-genes code for polypeptides

-polypeptides are processed into proteins

-proteins function as…enzymes, structural proteins, regulators, hormones, gene activators, gene inhibitors

How does dominance work: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

How does dominance work: Structure:

Heterozygous:

-50% functional structure

-50% proteins malformed

-mutant trait is expressed

-mutant trait is DOMINANT

Homozygous dominant:

 -100% non-functional structure so mutant trait is expressed

Homozygous recessive:

 -100% functional structure so normal trait is expressed

Prevalence of Dominance: Dominant does NOT mean better or more common!