Genes: units of inheritance passed onto offspring, located on chromosomes, and made up of DNA.
DNA belongs to a class of macromolecules called nucleic acid.
Nucleic acid: polymers made up of monomers called nucleotides.
Nucleotides: 3 parts: nitrogen base, pentose sugar (5-C), and a phosphate group.
Nitrogen base connects directly to pentose sugar.
2 types of nucleic acids: deoxyribonucleic acid and ribose nucleic acid. These molecules enable organisms to reproduce their components from one generation to the next.

**Purines**: one of the families of nitrogen bases, they are big with a 6-membered ring fused to a 5-membered ring. Ex: adenine & guanine

**Pyrimidines**: one of the families of nitrogen bases, they are small 6-membered rings. Ex: cytosine and thymine in DNA, where RNA contains uracil and cytosine.

- Adenine binds to thymine → forms 2 bonds between them.
- Cytosine binds to guanine → forms 3 bonds between them.
- In DNA, deoxyribose (pentose sugar) is attached to the nitrogen base.
- In RNA, ribose (pentose sugar) is attached to the nitrogen base.

^the only difference between these 2 is that **deoxyribose lacks** an oxygen atom on its #2 carbon.

Heredity: the transmission of traits from one generation to the next.
Variation: this brings about changes from offspring to siblings.
Genetics: study of heredity and variation.

Humans contain 46 chromosomes in their body/somatic cells and sex chromosomes in their reproductive/sex cells/gametes.
There are about 20,000 genes on the 46 chromosomes that make up the human genome.
Each chromosome consists of a one long DNA molecule; each one has 100’s to 1000’s of genes on it.

**Locus (loci):** a gene’s specific location on the chromosome.
Our genes program the specific traits that emerge as we develop from fertilized eggs to adults.
Genes, or segments of DNA, contain information for all the traits of the body.
Because DNA is too big to exit the nucleus, it relies on mRNA to transcribe or carry DNA’s message out of the nucleus; it then arrives at a ribosome where the message gets translated into the making of a protein/polypeptide.

“Like begets like” refers only to organisms that reproduce asexually.
Asexual reproduction occurs more often with unicellular organisms and SOME multicellular organisms (ex: hydra) by a process known as budding.

Buds are derived by mitosis and not meiosis → offspring are identical to the parent assuming no mutation.
Asexual reproduction: brings about clones of genetically identical individuals.

**Sexual reproduction:** results in greater genetic variations because unique combinations of genes come from two parents; it is a result of gametes (sperm and egg) coming together that were formed during meiosis.
The life cycle of an organism follows the reproductive stages of an organism from generation to generation.
Chromosomes differ by their 1) length 2) location of their centromeres 3) distinctive banding patterns when stained.
Although humans have 46 chromosomes, there are actually 2 of each kind (1 from mom and one from dad), so 23 pairs of chromosomes.

Karyotype: when chromosomes are arranged in pairs starting with the longest.
- Homologues/homologous pairs/tetrads: the chromosomes carry the same traits and carry the same genes at the same position or loci.
- **Alleles**: trait comes in different “versions” (ex: A vs. a). ← Exception: X and Y chromosomes/sex chromosomes determine an individual’s gender.
- Females: homologous chromosomes XX
- Males: Chromosomes XY
- **Autosomes**: the other 22 pairs/44 chromosomes have nothing to do with the gender.
- Each egg or sperm cell contains 22 autosomes and either X or Y for a sum total of 23 chromosomes.
- Sperms and eggs are **haploid** (1n).
- **Zygote** (diploid/2n): 2 haploids (1n) come together through fertilization.
- Once the zygote forms, mitosis occurs to allow the offspring to grow and develop.
- **Gametes (sperms and eggs)**: made in the gonads by meiosis, not mitosis.
- **Mitosis**: makes copies
- **Meiosis**: from 2n → 1n
- Human *somatic cells* are in the 2n (diploid) state and human gametes are 1n (haploid).
- Fungi and some protists (ex: algae) are different, their multicellular body cells and gametes are already in the 1n state, so via mitosis (not meiosis) they ready to fertilize/form a zygote-the 2n state temporarily. But because all their body cells exist normally as multicellular haploids, they need to halve their chromosome number and this is accomplished by meiosis.
- **Alternation of generation**: another life cycle that plants and some algae exhibit, their normal multicellular cells exist in both the 1n and 2n states on the plant.
- **Gametophyte**: when they are a multicellular 1n/haploid plant. Ex: gametophytes of an apple tree are the structures that make the gametes.
- **Sporophyte**: when they are a multicellular 2n/diploid plant. Ex: sporophyte of the apple tree is all other parts of the plants.
- Some parts of the sporophytes undergo meiosis, so therefore they MUST start as 2n.
- After meiosis they have halved to 1n and are called **spores**.
- These 1n multicellular spores now undergo mitosis to produce more just like themselves or 1n gametophytes that are the structures that make gametes.
- Then the gametophytes once again undergo mitosis to produce 1n gametes.
- The gametes then fuse to make a 2n zygote, and then this zygote undergoes mitosis to grow into a 2n multicellular sporophyte.

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A. Animals:
1) Multicellular form= 2n
2) Gametes= 1n

B. Fungi:
1) Multicellular form= 1n
2) Gametes= 1n

C. Plants:
1) Multicellular form= 1n + 2n
2) Gametes= 1n
Meiosis consists of meiosis I and meiosis II. There are 2 cell divisions resulting in 4 different daughter cells. Mitosis yields only 2 identical daughter cells.

Q: In picture 1: Why are there 2 chromosomes? One from mom and one from dad.
Do these chromosomes have the same genes? Yes
Do these chromosomes have the same loci? Yes
Do these chromosomes have the same alleles? Not necessarily
Q: In picture 2: Now the chromosomes have replicated and look like X’s. Where in the cell cycle does this occur? S-phase of interphase.
What is the best term to call these X-shaped structures? Sister chromatids/duplicated/replicated chromosome.
Q: In picture 3, with meiosis a replicated chromosome stays near by its matching replicated chromosome in what stage/s? Prophase I and metaphase I.
What do you call 2 replicated chromosomes next to each other? Tetrads, homologous, homologous pair.
What do you call the process of the 2 X’s coming together? Synapsis.
Since there are 4 chromatids they are called what at this point? Tetrads.
When in meiosis does exchange of genetic material called crossing over occur resulting in chiasmata? Prophase I.
What is the whole point of meiosis I? Separated homologous.
Q: In picture 4: What is the whole point of meiosis II? Separated sister chromatids.
Are the resulting daughter cells identical or different? Different.
- Offspring of sexually reproducing organisms are not genetically the same for the following 3 reasons:
  1) **Independence assortment**: when homologues line up at the equator of the cell in metaphase I or metaphase II, it is an “independent”/random event for each homologous pair. The number of possible combinations due to the law of Independent assortment is $2^n$ (n= haploid number). So in humans, there are $2^{23} \approx 8$ million possible different gametes due to independent assortment alone.
  2) **Crossing over**: it occurs during prophase I of meiosis when homologues come together in a process called synapsis to form pairs called tetrads. Then nonsister chromatids trade places. The resulting regions are called chiasmata. There is on average 2-3 pairs crossing events per homologous pair.
  3) **Random fertilization**: woman has ~8 million possible different ovules and a man has ~8 million possible different sperm cells. Therefore, if we ignore crossing and just put the results of the Law of Independent assortment to the next level of random fertilization, there are ~64 trillion possible different combinations of chromosomes.

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**REPRODUCTION**

- **Asexual**:
  - Asexual reproduction is the creation of new individuals by the process of mitosis. This creates identical offspring rapidly.
  - It is common with many invertebrates.
  - 3 types of asexual reproduction:
    1) **Fission**: division of a parent into 2 genetically identical individuals. Ex: bacteria and sea anemone.
    2) **Budding**: new individuals pinch off from an existing one to form a colony. Ex: hydra, yeast, corals.
    3) **Fragmentation**: breaking off of the body into smaller fragments that then develop into complete adults through regeneration. Ex: some species of seastars (planaria) can grow a new animal from an arm.
  - Advantages: animals living in isolation can produce offspring without a mate and can create many offspring in a short amount of time.

- **Sexual**:
  - It is the creation of offspring when 2 haploid gametes come together to form a diploid zygote (fertilized egg).
  - Gametes are created during meiosis.
  - Female gamete is the **ovum/oocyte** (unfertilized egg).
  - Male gamete is the **sperm/spermatozoan**.
  - Sexual reproduction increases genetic variation amongst offspring by inheriting genes from two parents.
  - This variability may enhance Darwinian fitness and survival when environmental factors change.
Exception:

- Some can do both sexual and asexual reproduction (Ex: Daphnia).
- Under favorable conditions, Daphnia reproduce by a process called parthenogenesis/ virgin birth.
- Parthenogenesis: mother organisms forms genetically identical daughters/ clones that do not need to be fertilized → allows their numbers to increase rapidly making use of short growing periods.
- Under unfavorable conditions, Daphnia give birth to males and sexual females so that mating can occur and some can survive.
- Example of parthenogenesis: bees, ants, birds (turkey), fish, amphibians, lizards.
- In bees, male drones develop by parthenogenesis, while the female sterile workers and fertile queen develop from fertilized eggs.

Most animals’ reproductive cycles are linked to the changing seasons → allows animals to conserve energy and reproduce when conditions are favorable.

- This is true even of animals in stale habitats like the ocean.
- Reproductive cycles are controlled by both hormones and environmental cues ex: temperature, rainfall, day length, and lunar cycles.
- Sexual reproduction is difficult for sessile (organisms that do not move) and burrowing animals that may never encounter a member of the opposite sex.
- Hermaphroditism: having both male and female reproductive systems.
- Even though hermaphrodites have male and female reproductive parts, most still must mate with another member of the same species. Ex: annelids.
- Sequential hermaphroditism: when an individual changes its sex/gonads during its lifetime.
- Protoandrous: going from a male to a female
- Protogynous: going from female to a male.
- Sex reversal is associated with age and size. Ex: some fish species the largest and the oldest individual can change from a female to a male if there are not enough males around.
- Fertilization: joining of sperm and egg.
- Environmental cues like temperature, day length, and pheromones or non-environmental cues like courtship behavior, influence gamete release.
- External fertilization: eggs and sperm meet in the environment, almost always occurs in water, produce large numbers of zygotes and few survive.
- Internal fertilization: sperms are deposited in or near the female's reproductive tract, produce fewer zygotes with high parental care, hence greater survival.
- Gonads: organs that produce gametes—ovaries in females and testes in males.
- Cloaca: many non-mammalian vertebrates (ex: fish and birds) have a single opening for both excretion of waste and reproduction.
- Most mammals have two separate openings.
- Spermatogenesis: production of mature sperm cells.
- The structure of a sperm cell fits its function.
- The head region contains a 1n nucleus that is tipped with enzymes called acrosome.
- Acrosome: enzymes that help the sperm penetrate the egg.
- Behind the head are many mitochondria that provide energy (ATP) for movement of the tail/ flagellum.
- The head/ chromosomes enter the ovum. Other parts just fall off.
- Oogenesis: development of ova (unfertilized egg cells).
- All of the 400,000 follicles a woman will ever have are formed before birth, but they are not “done yet”.
- Follicle: 1 egg surrounded by layers called follicle cells that protect and nourish the developing egg.
- At birth, all of the 400,000 eggs a woman will ever have are diploid (2n) and in G1 of interphase of meiosis.
- Birth → puberty: each of the eggs in the ovary ALL reach prophase I of meiosis and are essentially “stuck” there for a while.
- With puberty: usually a single primary oocyte completes meiosis I each month → occurs when FSH hormone stimulates one follicle (the “chosen one”) to go through meiosis I each month. At the end of meiosis I, the secondary oocyte is formed and this secondary oocyte is then released from the ovary with ovulation to go wait in the fallopian tube/ oviduct.
- This meiotic division is unequal—the secondary oocyte gets almost all the cytoplasm and organelles, and the other cell called 1st polar body is much smaller.
- If a sperm reaches/ penetrates the secondary oocyte, it will then undergo the meiosis II to produce the haploid ovum/ egg.
- The 2nd polar body separates from the ovum, while the haploid ovum and sperm's nuclei fuse= fertilization.

- When the secondary oocyte leaves the ovary during ovulation, it leaves behind follicular tissue → this tissue develops into the corpus luteum in the ovary.
- The corpus luteum secretes hormones estrogen and progesterone that maintain the uterine lining during pregnancy.
- If the secondary oocyte does not get fertilized, the corpus luteum disintegrates and the woman has her period, then a new follicle matures during the next cycle.
- Androgen (steroid hormones): the principle sex hormones of males
  - Testosterone: most important, produces both primary sex characteristics that are associated with reproductive system (sperm production and development of the reproductive structures) and secondary sex characteristics that are not directly related to the reproductive system (deepening of the voice, hair, growth, and increase muscle growth).

- FSH (follicle stimulating hormone): involved in the development of growing follicle.
- LH (luteinizing hormone) is also involved.
- Both hormones are secreted by the anterior pituitary and stimulate the release of the hormone estrogen and progesterone from the ovaries.
- As the follicle grows larger with the aid of FSH, it develops receptor sites for LH.
- While both LH and FSH increase rapidly by positive feedback, it is actually LH that induces ovulation of the secondary oocyte.
- After ovulation, LH stimulates the remaining follicular tissue left behind when the secondary oocyte was released to form the corpus luteum.
- LH then triggers the corpus luteum to secrete the hormone estrogen and progesterone. When the corpus luteum disintegrates, these 2 hormones decrease their levels → induces menstruation.
- If a woman is pregnant, these 2 hormones remain intact.
- Estrogen: responsible for secondary sex characteristics like high water retention, breast development, and Ca++ metabolism.
- **Menopause**: women cease to ovulate/menstruate → happens because the ovaries lose their responsiveness to the hormones FSH and LH which then cause the decline of estrogen.
- **Osteoporosis** (os = bone, porosis = hole): estrogen is linked to Ca++ metabolism. Ex: older women tend to have this.
- **Gestation/ Pregnancy**: condition of carrying one or more embryos.
- **Conception**: fertilization of an egg by a sperm.
- **Cleavage** (mitosis): division of zygotes about 24 hours after fertilization. It continues until it forms a solid ball of cells called a morula → blastula (hollow ball) then implants into the endometrium of the uterus.
- **Gastrulation**: the cells invaginate/push inwards to form a gastrula.
- **Gastrula**: commence the differentiation of body structures.

- **Placenta**: disk-shaped organ that contains both embryonic and maternal blood vessels, exchanges materials between the mother and embryo like nutrients, respiratory gases, and disposes of metabolic waste from the embryo.
- The exchange of these materials occurs via the arteries and veins of the embryo's umbilical cord.
- **Organogenesis**: development of organs occurs during the first trimester of pregnancy (1st three months).
- The embryo is now considered a fetus because all the major structures of the adult are present in rudimentary form.
- **Human chorionic gonadotropin (HCG)**: secreted by the embryo during the 1st trimester of pregnancy. It maintains the secretion of progesterone and estrogen by the corpus luteum and keeps the endometrium lining intact.
- Pregnancy test detects HCG in urine.
- During the 2nd trimester, HCG drops, the corpus luteum deteriorates, and the placenta secretes its own progesterone to maintain the lining.
- 3rd trimester: estrogen levels are the highest → triggers the release of oxytocin receptors on the uterus. Oxytocin causes powerful contractions of the smooth muscles of the uterus. Prostaglandins (local regulators) are secreted to enhance contractions and cause pain.
- After birth, lactation occurs in mammals.
- The anterior pituitary produces the hormone prolactin that stimulates milk production in the mammary glands and is controlled by the hormone oxytocin.
• **Birth control pills**: a combination of synthetic progesterone and estrogen, act through negative feedback by stopping the releasing of FSH and LH thus preventing ovulation.

• **Zygote division**: happens after fertilization.
  1) **Cleavage**: zygote divides to form solid morula→ blastula (hollow/fluid-filled).
  2) **Gastrulation**: 3 germ layers form in the embryo.
  3) **Organogenesis**: generates rudimentary organs from which adult structures will grow.

• **Cleavage**:
  - Rapid cell division that follows fertilization.
  - The cell cycle during cleavage includes only the S-phase of Interphase (no G₁ or G₂) where DNA gets copied, the M-phase (mitosis), and cytokinesis.
  - Embryo does not enlarge during cleavage, it simply partition the cytoplasm of the zygote into many smaller cells called blastomeres.
  - Echinoderms (ex: sea stars) and chordates (ex: humans) are **deuterostomes**= 2nd mouth. The cleavage is radial meaning that the upper four blastomere cells are aligned directly over the lower four blastomeres.
  - Mollusks (ex: clams), annelids (ex: earthworms), and arthropods (ex: crustaceans) are **protostomes**= 1st mouth. The cleavage is spiral where the blastomere cells of the upper tier sit in the grooves between the cells of the lower tier.

• **Gastrulation**:
  - Morula→ blastula→ gastrula
  - It is the rearrangement of the cells of the blastula.
  - **Invagination**: cells at one pole of the blastula move inward, it transforms the hollow single layered blastula into a 3-layered gastrula.
  - **Blastopore**: point where the cells invaginate.

  - The 3 germ layers:
    1) **Ectoderm**: outside layer of the gastrula. Becomes epidermis of skin, nervous system.
    2) **Endoderm**: lines the embryonic digestive tract. Becomes lining of digestive tract (linings)
    3) **Mesoderm**: partially fills the space between the endo and ectoderms. Becomes skeletal system, muscular system, dermis of the skin (most of the body system).

  - Eventually the blastopore will from another opening on the opposite end of the developing embryo.
  - In protostomes: the mouth develops from the blastopore.
  - In deuterostomes: the anus develops from the blastopore.
Organogenesis:

- Three kinds of morphogenetic (body shape or look) changes occur with organogenesis:
  1) Folds
  2) Splits
  3) Condensation/clustering of cells

- Neural tube and notochord: 1st organs to take shape in chordates.
- Neural tube → Central nervous system
- Notochord → backbone. The notochord forms when the mesoderm condenses.
- If a pregnant mother does not get enough folic acid, her baby will have defected neural tube (CNS defects ex: spina bifida).

MENDEL

- Before Mendel, people believed in blending hypothesis: analogous to how blue and yellow paints blend to make green.
- Mendel introduced the particulate hypothesis: deals with discrete units called factors or genes passed on from parents to offspring that retain their separate identity.

Perfect flower: both female and male.

- The pea plant was ideal to work with and Mendel’s results were so accurate because:
  1) Many varieties: purple vs. white flowers, yellow vs. green seeds, etc.
  2) Easy to control the fertilization: Mendel removed the male stamen so the plants could not self-fertilize.
  3) Chose traits that were "either-or": ex: purple or white flowers, no intermediate.
  4) Started with true breeding/purebred varieties: all the parentals were homozygous.

  ➢ Hybridization: mating or crossing of 2 varieties. Monozygotic is for a single character, like flower color. Dihybrid is for two characters, like flower color and seed shape.

  5) Followed the traits for at least 3 generations: P (parental), F\textsubscript{1} (first filial (offspring/daughters/sons), which then self-pollinate, and the F\textsubscript{2} generation.
  6) Use quantitative analysis.
  7) Large number of offspring/sample size → increase probability.
  8) Short life cycle and easy to maintain/grow peas.

- Mendel’s experiments showed that the blending model of inheritance was incorrect.
- Purple x white flowers in the parents produces all purple flowers, not pale purple flowers → purple flower color (P) is dominant to white (p).

\[
G = \text{genotype ratio}; \ P = \text{phenotype ratio} \rightarrow \]

\[
P \text{ Generation (true-breeding parents)} \]
\[
P P \times pp \rightarrow 100\% \ Pp \]

\[
F_1 \text{ Generation (hybrids)} \]
\[
P \text{ purple flowers} \rightarrow \text{All plants had purple flowers} \]

\[
F_2 \text{ Generation Ratio 3:1} \]
\[
\begin{array}{c}
705 \text{ plants had purple flowers} \\
224 \text{ plants had white flowers} \\
\end{array}
\]

\[
G = 1:2:1 \\
P = 3:1
\]
Mendel observed the same pattern of inheritance with six other characters (7 totals different).

Mendel explains this 3:1 ratio of inheritance with 4 related ideas:

1) Alternate versions of genes account for variation. Ex: purple vs. white flower color. The alternate versions of genes are called alleles.

2) An organism inherits 2 alleles, one from mom and one from dad. Thus a genetic locus is represented twice in a diploid organism like humans. These homologous loci may be identical alleles (homozygous) or may be different (heterozygous).

3) If the 2 alleles differ, the dominant allele is expressed in the organism’s phenotype, while the recessive is not seen.

4) Law of Segregation: the 2 alleles present in a cell separate or segregate during gamete production. The separation of alleles during meiosis accounts for the haploid number in sperm and eggs.

- Phenotype: how an organism looks, like purple vs. white.
- Genotype: genetic makeup, like PP or Pp or pp.
- Test cross: the breeding of an unknown genotype with a homozygous recessive. If a pea plant has purple colored flower, we can’t tell its genotype. It could be PP or Pp. To determine a pea plant’s genotype, it is necessary to cross the mystery purple plant with a white homozygous recessive plant (pp). If all the offspring have purple flowers the unknown purple plant’s genotype is PP. If half are purple and half are white, the unknown purple plant’s genotype is Pp.

By performing dihybrid crosses, Mendel developed his 2nd law called the Law of Independent Assortment.

- Law of Independent Assortment: Mendel questioned whether 2 characters, like seed shape and seed color were inherited as a package or independently. He discovered that in a dihybrid the two alleles for a character (like seed shape) assort independently of the two alleles for another character (like seed color). This produces four gametes: RY, Ry, rY, ry.

- Monohybrid = law of segregation. (mono=1, segregation=1 word)
- Dihybrid = law of independent assortment. (di= 2 independent assortment= 2 words).

- Probability is closer to predicted results when the sample size is large.

- Dominant does not mean a trait is more common in a population. Ex: polydactyl- having extra fingers or toes is dominant to the allele for 5 digits. If the baby is born with PP or Pp (P=polydactyl, p=normal), polydactyl is exhibited.

- An allele is not called dominant because it subdues a recessive allele.

- Alleles are variations in a gene’s nucleotide sequence (A,T,C,G).
Round vs. wrinkled pea seed shape comes about due to the presence or absence of an enzyme that converts simple sugars to starch. Round (R) is dominant, so if a pea seed's alleles are RR or Rr the enzyme is made and the seeds are round. If the pea seed's alleles are rr, the enzyme is not made to convert the sugar to starch so the seed stores simple sugars. Then water enters the seed by osmosis causing it to swell and wrinkled when it dries.

**Incomplete dominance:** when the F1 hybrids have an appearance in between the 2 parentals. Ex: when you cross red snapdragons with white snapdragons you get pink snapdragons.

**Codominance:** when both alleles are expressed in the phenotype. Ex: three different human blood groups: M,N, or MN. These groupings are based upon molecules located on the surface of red blood cells. So an “M” individual’s genotype is MM, and “MN” individual’s genotype is MN, and an “N” individual’s genotype is NN. Notice the “MN” individual’s phenotype expresses both types of molecules on RBC’s and hence is codominant. Ex: black and white checkered chicken.

**Multiple alleles:** exist in populations with more than 2 allelic forms. The ABO blood group is an example of multiple alleles (three different alleles of one gene). A,B,AB, and O blood types represent two carbs that either are present or absent on to the surface of RBC’S- the A substance and the B substance.

**Blood type AB is an example of codominant, where both alleles are expressed.**

**Pleiotropy:** a gene affects an organism’s phenotype in many ways. Ex: sickle cell-anemia caused by a recessive gene. This causes RBC’s to change their shape, resulting in many symptoms.

**Epistasis:** when a gene at one locus affects the phenotypic expression of a gene at another locus. This is seen with fur color in mice. Black fur is dominant to brown. So if a mouse has BB or Bb it is black. However, a second gene locus determines whether pigment is present or not. In order to have any color/ pigment a mouse must have the dominant form of the pigment allele (CC or Cc). If the pigment genotype is cc, then the mouse is albino regardless of the genotype at the black/brown locus.

**Polygenic inheritance:** human skin color and heights - when a character varies along a continuum. It is defined as the additive effect or accumulative effect of two or more genes on a single phenotypic character. Ex: skin color each dominant allele contributes to dark skin color and each recessive allele to light skin. So AABBCC person would be very dark having 6 “units” of darkness, a aabbcc person would be very light, and a AaBbCc person would be intermediate. Because polygenic inheritance is quantitative the genotypes AaBbCc and AABBcc each contribute 3 “units” to skin darkness. (the figure is on the next page)

- Phenotype is determined not only by genes but also by the environment.
- Nature (gene) and nurture (environmental factors) both influence all traits.
• Nature is the genotype.
• Nurture can be such things as nutrition, exercise, or exposure to sunlight in the case of skin color being affected by the environment.
• The nurture side of this debate is broadest for polygenic traits like skin color and height.
• With hydrangea flowers, the same genetic (nature) varieties produce a range of colors from pink (acidic) to white (neutral) to blue (basic) based upon the environmental factor of soil pH.
• Pedigree: a method to track a trait across generations to understand the past and to predict the future.

![Pedigree Example]

- When allele causes a genetic disorder there is either a malfunction of a protein or no protein at all.
- With recessive disorders heterozygous individuals have a “normal” phenotype because one copy of the normal allele produces some of the specific protein.
- Heterozygotes that are phenotypically normal are called carriers.
- A recessively inherited disorder shows up only with homozygous recessive individuals.
- Genetic disorders not evenly distributed among all groups of humans. This is because of past conditions when the world was less industrialized and people were more geographically isolated.
• **Cystic fibrosis**: most common recessive lethal genetic disease in the U.S. This disorder affects Cl⁻ transport between cells and the interstitial fluid. The protein channels that transport chloride in and out of the cell are defective or absent in people with cystic fibrosis. This causes Cl⁻ to build up in the extracellular fluid that leads to mucus build up around organs and air pathways, as well as bacterial infections.

![Cystic Fibrosis Diagram]

• **Tay-sachs disease**: recessive disorder where a dysfunctional enzyme fails to break down lipids in the brain → causes seizures, blindness, degeneration of motor and mental performance.
• **Sickle cell anemia**: recessive disorder common amongst African Americans. The substitution of one amino acid in the hemoglobin protein of RBC’s causes RBC’s to sickle. As a result, sickled cells hold less oxygen. While this is a recessive disorder its alleles are incomplete dominance, so some individuals who are heterozygous for sickle cell disease suffer some
symptoms of the disease due to a fraction of their RBC’s being sickled. About 1/10 African Americans are carriers of this allele (Ss). The reason for this high number is because in certain environments like Africa, heterozygotes have an advantage over homozygous dominant having a single copy of the sickle cell allele increases resistance to malaria.

- Most harmful alleles are recessive.
- **Achondroplasia**: dominant disorders; a form of dwarfism is expressed in individuals with AA or Aa genotypes. Therefore, 99.99% of the population is aa.
- Many lethal dominant alleles result due to mutation. Other can escape notice early in life and as a result, be passed onto offspring. Ex: Huntington’s disease: dominant disorder that doesn’t show phenotypic symptoms until age 35~45; this disorder causes degeneration of the nervous system.

- **Amniocentesis**: involves inserting a needle into the amniotic fluid surrounding the fetus around the 14th-16th week of pregnancy. This fluid is either A) tested with chemicals for certain genetic disorders or B) the cells from the fluid are grown in a labototry or within weeks a karyotype can identify chromosomal defects.
- **Chorionic villus sampling (CVS)**: fetal testing where a doctor suctions out fetal tissue from the placenta around 8th-10th week of pregnancy. This tissue is then karyotyped immediately, giving results within 24 hours.
- **CVS and amniocentesis are invasive- to check for abnormalities.**
- **Ultrasound**: uses sound waves to produce an image of the fetus (non-invasive).
- **Fetoscopy**: a tube with a viewing scope inserted into the uterus.

**CHROMOSOMES**

- Chromosomal theory of inheritance: Mendel called something that was being passed between generations “factors”. Until 1902, scientists noted the parallels between Mendel’s factors, now called genes and chromosomes.

- **T.H. Morgan**: 1st to associate the gene with the chromosome in his work with *Drosophila melanogaster*. He chose fruit flies because:
  - A) single mating produces hundreds of offspring
  - B) A new generation can be bred every 2 weeks
  - C) Fruit flies has only 3 pairs of autosome, 1 pair of sex chromosomes, which makes them relatively simple to study.

- After a year breeding, a mutation occurs—a white eyed fly, instead of the usual color of red.

- **Wild type**: the normal phenotype for a character/ the most common in natural populations.

- **Mutant phenotype**: alternate to the wild type.

- Morgan introduced new symbols to identify these traits; he didn’t use capital and lowercase letters for the gens. For a given character the gen takes its symbol from the mutant. So in this case, he chose w, then used the superscript + to identify the wild-type trait. ➔ Homozygous red-eyed fly would be written as w+w++. Morgan mated the white-eyed male mutant Xw+Y with a homozygous red-eyed female Xw−Xw−, all the offspring had red eyes. When Morgan bred the F1 flies, he observed the normal 3:1 phenotypic ratio with the F2’s. But the white-eyed trait only showed up in the males. All the females had red eyes, where 50% the males had red eyes, 50% the males had white eyes.

- **Sex-linked traits**: genes located on the X-chromosomes.
- Each chromosome has hundreds-thousands of genes.
- Genes located on the same chromosome are inherited together in genetic crosses because the chromosome is passed along as a unit.
- Genes on the same chromosome are said to be linked. ➔ This doesn’t obey Mendel’s Law of independent assortment.
- With linked genes (body color& wing size), the alleles are located on the same chromosome and are inherited together. With body color, gray is normal and black is mutant. With wing size, wild type (long) is normal and vestigial (short) is mutant.
- In a test cross, if these genes were separated one would expect a phenotypic ratio of 1:1:1:1. But the results were many of the parental genotypes and few
recombinants. These recombinants came about due to crossing over during prophase I.

- **Genetic Map**: an ordered list of loci along a chromosome, constructed by Sturtevant (one of Morgan’s student).
- Then by associating frequencies of recombinants along the chromosomes, Sturtevant constructed a linkage map. He found that recombinant frequencies corresponded to the distance between genes on a chromosome.
- The further apart 2 genes are on a chromosome, the greater the probability that a crossover will occur and thus, the greater the recombination frequency.
- This distance is expressed in map units. One map unit = 1% chance of recombination.
- Some genes on a chromosome are so far apart from each other that crossovers occur very often. They are 50 map units apart and thus are not considered linked because their results are indistinguishable from genes located on different chromosomes.
- With Humans, we know females are XX and males are XY. So with each birth, it is the male who determines the gender of the offspring because the females’ eggs all carry an X chromosome. This is NOT the case with all animals.
- Sex-linked/ x-linked characters: have many genes unrelated to sex.
- For a female to express a recessive sex-linked allele, she has to be homozygous recessive (X<sup>a</sup>X<sup>a</sup>).
- Males only have one locus → use the term hemizygous.
- Any male receiving the recessive allele from his mother X<sup>a</sup>Y expresses the trait. This is why many more males than females have sex-linked recessive disorders.
- In order for a female to get a sex-linked recessive disorder → her mom has to be a carrier and her dad must have the recessive disorder. This chance of this happening is very slim.
- **Red-green blindness**: mild recessive sex-linked disorder.
- **Duchenne Muscular Dystrophy**: X-linked recessive disorder affecting 1 in 3,500 males, this leads to the weakening of muscles and loss of coordination because a key muscle protein (dystrophin) is missing.
- **Hemophilia**: X-linked recessive trait due to the absence of a protein for blood clotting. Hemophiliacs bleed excessively when injured even with minor cuts or bruises.
- Although female mammals inherit two X chromosomes, one from each parent, one of these 2 becomes almost completely inactive during embryonic development.
- **Barr body**: inactive X in each female cell condenses into a compact object; it lies along the inside of the nuclear membrane; most of the genes are not expressed, although some remain active; these Barr body chromosomes are reactivated in the ovaries.
- X chromosome is inactivated in a cell is random and differs from one cell to the next.
- Females consist of a mosaic of two types of cells: those with the active X derived from the mother and those with the active X derived from the further.
- ^can be seen in humans where a woman is heterozygous (X<sup>S</sup>X<sup>s</sup>) for the development of sweat glands. So she has patches of normal skin and patches of skin lacking sweat glands. Barr bodies come about because methyl groups (−CH<sub>3</sub>) inactivate one of the X chromosomes.
- This X-inactivation explains why calico cats are almost always female. One X chromosome can have an allele for orange color; the other X chromosome can have an allele for black color. These 2 colors are codominant sex-linked. ∴ a female can inherit both alleles, while a male gets only one color. Another set of alleles NOT on the X chromosomes are involved in patchy vs. white coloring. So when Barr bodies form randomly in different cells of a female cat heterozygous for coat color, the typical calico pattern forms.
- Sometimes during anaphase I or anaphase II chromosomes are not separated as expected → causes nondisjunction.

- Gametes often have one extra or one less chromosome than normal.

- **Aneuploidy**: with fertilization the offspring will have an abnormal chromosome number.

- In most cases of aneuploidy, the zygote dies.

- **Nondisjunction** can also occur during mitosis. If this occurs during development of the embryo it will affect a large number of cells.

- **Polyploidy**: some organisms don’t inherit one extra or one less chromosome, but instead inherit an entire set.

- **Triploidy** (3N): an organism gets one extra set and can arise if a sperm fertilizes an egg that is 2N.

- Polyploidy is common in the plants kingdom, but rare with animals.

- **Nonreciprocal translocation**: chromosome segment is given to a nonhomologous chromosome without receiving and return.

- **Down syndrome/ trisomy 21**: result of an extra chromosome 21. Each body cell has a total of 47 chromosomes. Symptoms: facial features, short stature, heart defects, respiratory infections, mental retardation, and some are sterile; they are also prone to leukemia and Alzheimer’s both of which the genes are located on chromosome 21. The frequency of Down’s syndrome increases with the age of the mother, where women older than 35 are most susceptible.

- **Klinefelter syndrome**: an extra X chromosome in a male XXY. Man is sterile; breast development is common and is of normal intelligence.

- **Jacob Syndrome/ Super Male**: males with an extra Y chromosome XYY. Man tends to be taller than average.

- **Trisomy XXX**: females are healthy, cannot be distinguished from XX females except via karyotype.

- **Turner’s syndrome/monosomy X**: only viable monosomy in human, females having this are XO, have underdeveloped sex organs, sterile, short, and of normal intelligence.

- **Cri du chat**: due to a deletion in chromosome 5 → mental retardation, small head, unusual facial features, and a cry that resembles a distressed cat.

- **Leukemia**: due to a reciprocal translocation, in which a portion of chromosome 22 and chromosome 9 switch places → affects the cells that give rise to abnormal WBC.
- **Prader-Willi**: disease inherited from dad → causes obese, insatiable appetite, mental retardation, shorter, smaller hands & feet.
- **Angelman**: disease inherited from mom → causes uncontrollable laughter, jerky movement, motor and mental symptoms.
- Both of these disorders are caused by a deletion of chromosome 15.
- **Genomic imprinting**: it seems the parental origin of the chromosome is imprinted on the chromosomes. This is true for both the autosomes and sex chromosomes. But these imprints are erased before the offspring produces its own gametes. It can also explain fragile X syndrome.
- **Fragile X syndrome**: tip of an X chromosome is hanging by a thin thread of DNA → causes mental retardation and is more common when the abnormal X chromosome is inherited from the mom, so male offspring are most affected.
- Genes/chromosomes/DNA are found primarily in the nucleus.
- Animals have extracellular DNA in mitochondria; plants have extracellular DNA in chloroplast and mitochondria.
- Some disorders associated with mitochondrial DNA affect ATP.
- Reductions in the amount of ATP a cell can make leads to energy deprivation especially to the nervous system and muscle.
- While plants also have mitochondrial DNA, yellow and white patchiness on leaves is determined by DNA in chloroplast. So whether a leaf is spotted or striped results from genes from the ovule and not the pollen.