Honors Biology, Unit 6 Study Guide

Terminology

Genetics	Study of biological inheritance patterns and variation
Gene	Piece of DNA that instructs a cell to make a certain protein (structural or functional)
Allele	One of many forms of a gene, two alleles per gene (eg B, b)
Dominant	Allele expressed when 2 different alleles (Bb) OR 2 dominant alleles are present (BB)
Recessive	Allele only expressed if 2 copies of recessive are present (bb)
Homozygous	Two of the same allele (eg BB, bb)
Heterozygous	Two different alleles (eg Bb)
Genotype	Genetic makeup of a specific set of genes (eg BB, Bb, bb)
Phenotype	Physical characteristics of offspring
Genome	Collection of all the organism's genetic material
Carrier	Heterozygous for disease; carries allele for disorder but doesn't express symptom
Pedigree	Genetic map where phenotypes used to infer genotypes

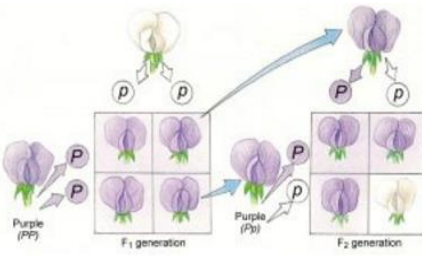
Mendel's Experiments

Before experiment – 3 decisions:	**After experiment - 3 key conclusions:**
Control over breeding: allowed no pollen from other flowers	**Traits are inherited as discrete units:** traits don't blend, independently expressed (eg pea plants can be green OR yellow)
Use of purebred (homozygous) plants	**Law of segregation: organisms inherit two copies of each gene, one from each parent**
Observation of "either-or" traits: only two forms of each gene	**Law of segregation: organisms donate one copy of each gene in their gamete** (2 copies of each gene separate during gamete form)

Punnett Squares

Genotypes	Geno Ratio	Pheno Ratio
1GG, 2Gg, 1gg	1:2:1	3:1
2GG, 2gg	1:1	1:1
4GG	4:0	4:0
4gg	0:4	0:4

Pea plants	reproduce quickly; control how they mate
P (parental) generation	Crossed purebred white (pp) w/ purebred purple (PP)
F1 (1st filial)	All purple flowers (Pp)
F2 (2nd filial)	trait for white hidden

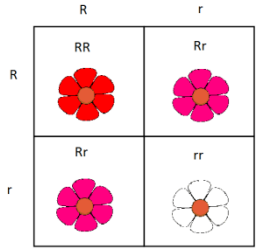


Monohybrid cross

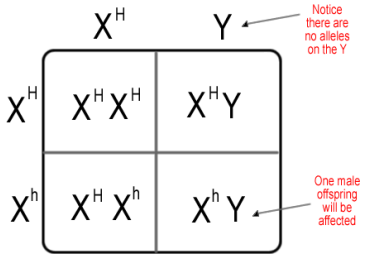
Cross: Aa x Aa

	A	a
A	AA	Aa
a	Aa	aa

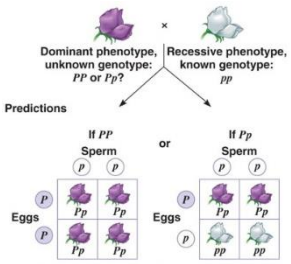
Incomplete dominance



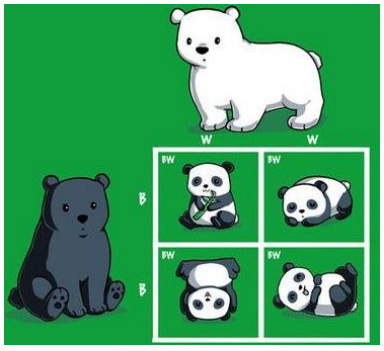
Sex-linked



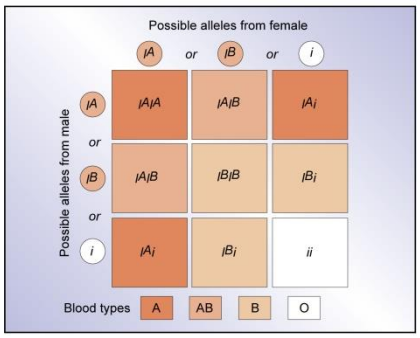
Test Cross



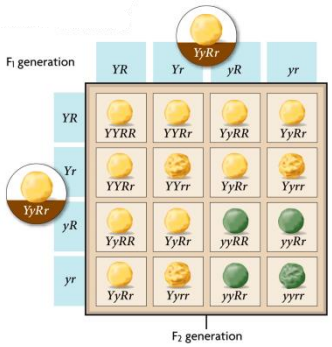
Codominance

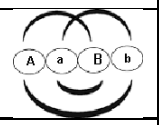


Multiple alleles



Dihybrid

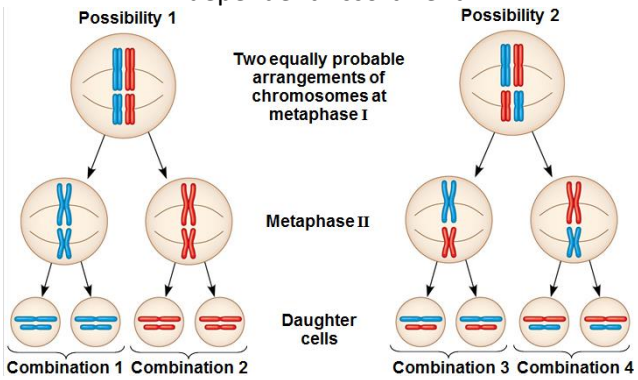


Incomplete dominance	Traits blend: eg red+blue=purple
Codominance	Both traits are expressed: eg a roan cow has both red and white hairs
Sex-linked (see Sex-Linked Traits)	Most sex-linked genes are only on the X chromosome (X^H, X^h) So only females (XX) can be carriers; males (XY) express whatever is on their X
Multiple alleles	Expresses a range of dominance: eg blood types A (I^AI^A or I^Ai), B(I^BI^B or I^Bi), AB(I^AI^B), O (ii); A and B are codominant, O is recessive
Dihybrid cross (see Law of Independent Assortment)	2 traits; pheno ratio **9:3:3:1** **Make gametes by FOILing:** TtBb = TB, Tb, tB, tb; PPGG = PG 
Test cross	Cross btwn a organism with a recessive phenotype, known genotype and a dominant phenotype, unknown genotype (PP or Pp)

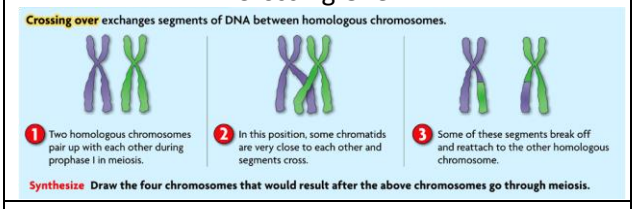
Sexual Reproduction Creates Unique Gene Combinations

Independent assortment	**Law of independent assortment:** allele pairs separate independently of each other during meiosis
Random fertilization of gametes	70 trillion chromosome combos possible
Crossing over	Exchange of chromosome segments btwn homologous chromosomes in Prophase I or Meiosis I, results new gene combo
Linked genes (see gene linkage)	Genes located on same chromosome inherited together • Closer together = higher chance of inheriting together • If far apart, crossing over may separate them

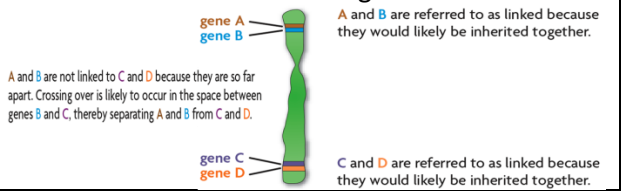
Independent Assortment



Crossing Over



Gene Linkage



Chromosomes and Phenotype

Autosomal traits	**Recessive disorders:** must have 2 copies of recessive allele to have disorder • often appears in offspring of carriers • eg cystic fibrosis: deadly, affects sweat glands and mucus glands
	dominant disorders: less common, eg Huntington's Disease: damages nervous system and usually appears during adulthood, after having children
Sex-linked traits	• Many genes are only contained on larger X chromosome (eg X^H, X^h) • Females (XX) can be carriers for traits because they have two X chromosomes • Males (XY) who has gene for disorder on X chromosome will have disorder, because no second X to mask effects • X Chromosome inactivation: 1 of 2 X chromosomes in females is randomly turned off, creating patchwork of two types of cells
Polygenic traits	Two or more genes determine trait: eg skin color, human eye color
Epistasis	One powerful gene overshadows all of the others, eg **albinism:** a person has genes for pigments, but recessive albinism gene overpowers and removes pigments
Gene Linkage & Mapping (see Linked Genes)	Thomas Hunt Morgan studied linked genes with fruit flies Linked genes not inherited together every time because of **crossing over** Linkage maps estimate distances between genes, make genetic maps of species

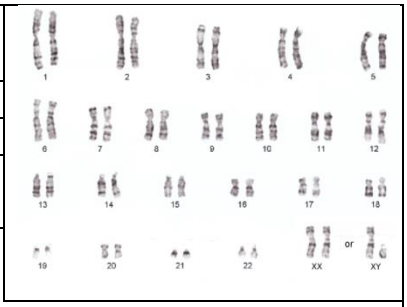
Labs

Shiba Inu Lab: cream dogs are example of epistasis (one gene overpowers the rest)

- Two genes: first is normal gene for fur color (red/sesame/black and tan)
- if recessive form of second gene, overpowers first gene and makes entire dog cream

Karyotype Lab

Karyotype	picture of all chromosomes in a cell; identifies extra, too few, or parts missing of chromosomes
Autosomes	First 22 pairs of chromosomes, 1 from each parent
Sex chromosomes	XX or XY, get either X or Y from dad
Nondisjunction	Cause of Down, Turner syndrome: genes don't split correctly and result in extra/missing chromosomes
Key features to match chromosomes	Size: should be similarly sized Centromere position: where chromatids are joined Banding pattern: Giemsa dye causes dark bands on A & T, so bands should line up

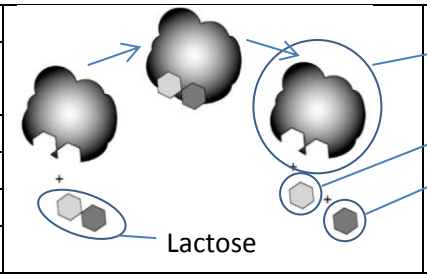


Disease	Cause	Symptoms
Down Syndrome	Extra chromosome 21	
Trisomy 13	Extra chromosome 13	Early death
Cri du Chat	Deletion of chromosome 5	Baby cries sound like cat wails
Turner Syndrome	Missing/incomplete X - X	Girls are shorter, start puberty late
Klinefelter Syndrome	Extra X chromosome - XXY	Guys: less muscles, little facial/body hair, infertile
Williams Syndrome	Missing gene material from chromosome 7	Circulatory system issues, heart defects

Lactose Intolerance

Lactase (enzyme/protein) breaks lactose (sugar) into glucose and galactose
Ethnic groups that didn't raise dairy animals are generally intolerant (Asians, Africans, 75% of world!)
Independent variable: solutions tested (Lactaid, milk, Lactaid+milk, glucose)
Dependent variable: presence of glucose

Enzymes
speed up chemical reactions that would take too long to complete
only fit certain substrate, can't be any reaction
not used up afterwards; they are reusable
Enzyme optimal temp: body temp
This reaction is an example of **catabolism/hydrolysis** →



Epigenetics

Epigenetics	Study of changes in heritable phenotype without a change in genotype Phenotype is affected by more than just gene expression, environment plays a part
Epigenome	second layer of DNA structure that changes during your lifetime made up of chemical tags that turn genes on and off based on outside signals
Active Gene	Loosely wrapped; less methyl; more acetyl
Inactive Gene	Tightly wrapped; more methyl; less acetyl
Reprogramming	During development, epigenetic tags are erased (blank slate) so cells can specialize
Imprinting	Certain epigenetic tags make it through reprogramming & pass unchanged to offspring
Licking Rats	Mothers lick their rats to loosen and activate GR gene in rats, making them calmer
Cancer Cells	Lower level of methylation, activates cell growth genes, chrom. instable, imprint loss
Gene Regulatory Protein	Carry signals: switch genes on/off, recruit enzymes that add/remove tags
Twins	Share genome, environments so epigenome changes: • both twins get disease, disease in genome; one twin gets disease, in epigenome