mendel

worked w pea plants

his theory us one of particulate inheritance	inherited characteristics are carried by genes

nondisjunction

error in meiosis where homologous chromosomes fail to separate as they should

one gamete receives two of the same type of chromosome and other receives no copy

remaining chromosomes may be unaffected and normal

if either aberrant gamete unites w normal gamete during fertilization, resulting zygote will have abnormal # of chrom

aneuploidy	any abnormal number of chromosomes
trisomy	if chromosome is present in triplet
trisomy 21 (Down syndrome)	extra chromosome 21

cancer cells grown in culture almost always have extra chromosomes

organism in which cells have extra set of chromosomes is triploid (3n)

4n tetraploid

strawberries r octoploid

polyploidy is COMMON in PLANTS

results in platens of abnormally large size

in some cases in responsible for evolution of new species

crossover and linkage mapping

chiasma	physical bridge around point of exchange

result of crossover is recombination

crossover and recombination are major sources of variation in sexually reproducing organisms

one map unit distance on chromosome is distance within which recombination occurs 1 percent of the time

crossover/recombination frequency

recombination frequency	number of recombinants / total #offspring X 100

can find recom frequency fro linked genes this way

is expressed as a percent

dihybrid cross

genotype (AaBb x AaBb)	9:3:3:1

linked genes

is OPP to ia

if height is linked to seed color, genes will **not** segregate independently

on SAME chromosome

genes that are adjacent and close to each other on same chromosome tend to move as unit and do NOT segregate

genotype for two traits is dihybrid

humans have 46 chromosomes, so have 46 linkage groups

mendels second law: law of segregation

during formation of gametes, two traits carried by each parent separate

are **not** linked

ex: monohybrid cross

trait not identified in either parent appears in F1 generation (recessive when 2 hetero)

intro

people used to think that inheritance was blended, a mixture of fluids that passed from parents to children

probability

prob can predict average outcome

absolute certainty is 1

multiply	prob of two independent events, multiply chance of one by chance of other

ex: chance of a couple having two boys (1/2 x 1/2)

add	more than one arrangement of events producing the specified outcome is possible

order matters

ex: couple having children, one boy one girl in either order

boy and then girl is 1/2 x 1/2 = 1/4

girl then boy is 1/4 too

1/4 + 1/4 = 1/2

mendels third law: law of independent assortment

applies when cross is carried out bet two individuals hybrid for two or more traits

that are NOT on the same chromosome

dihybrid cross

ex: height segregates independently from seed color

only factor that det how these alleles segregate or assort is how homologous pairs line up in metaphase 1 which is **random**

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mendels first law: law of dominance

only dominant trait shows

recessive is hidden

incomplete dominance

BLENDING

neither trait is dominant

genotype is cap letters

ex: red Japanese flower crossed w white one

produces pink offspring

chromosomal abberations

deletion	fragment lacking a centromere is lost during cell division
inversion	chromosomal fragment reattaches to its go chromosome but in reverse orientation
transl-ocation	fragment of chromosome becomes attached to a non homologous chrom
polyploidy	when cell or organism has extra **SETS?** of chromosomes

X inactivation

early in development of the embryo of female mammal, one of the X chromosomes is inactivated in every somatic (body cell)

embryo	an unborn or unhatched offspring in the process of development

inactivation occurs randomly

results in embryo that is a genetic mosaic (some cells have one X activated, some have other)

all cells of female mammals are NOT identical

inactivated chromosome condenses into dark spot of chromatin can be seen at the outer edge of nucleus of all somatic cells in female

^ Barr body

X inactivation (cont)

ex female calico cats (pg 141)

another ex of x chrom inactivation is when certain x linked recessive mutation prevents the development of sweat glands

heterozygous for some does NOT mean carrier

^ has patches of normal skin and patches of skin lacking sweat glands

polygenic inheritance

blending of several sep genes that vary along a continuum

bell shaped curve

ex: skin color, hair color, height

epistasis

two separate genes control one trait

one gene MASKS the expression of the other gene

the gene that MASKS is epistatic to the gene it masks

gene interactions

pleiotropy	ability of one single gene to affect an organism in several or many ways

ex is autosomal recessive disease cystic fibrosis

characterized by abnormal thickening of mucus that coats certain cells

instead of protecting body, thick mucus builds up in pancreas, lungs, digestive tract

pleiotropic affects: poor absorption of nutrients in the intestine and chronic bronchitis

multiple alleles

most genes in a pop exist in two allelic forms	ex tall or short

multiple alleles is when there are more than two allelic forms of a gene

4 diff blood groups	A, B, AB, O

are set by specific molecules on surface of red blood cells

3 alleles that det those (ABO)

A and B are codominant	I^A and I^B
I stands for immunoglobin	
O is recessive	i

codominance

BOTH traits show

ex: MN blood groups in humans

NOT related to ABO blood groups

3 diff blood groups (M, N, MN)

based in distinct molecules located not he surface of the red blood cell

single gene locus at which two allelic variants are possible

more to it but won't put here

incomplete dominance

BLENDING

neither trait is dominant

genotype is cap letters

ex: red Japanese flower crossed w white one

produces pink offspring

beyond mendelian inheritance

mendelian laws apply to traits set by a **single gene** for which there are only two alleles

now we can do ones w 2 or more genes

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more exceptions to mendelian inheritance

genomic imprinting	variation in phenotype depending on whether a trait is inherited from the mother or the father

occurs in gamete formation

caused by silencing of a particular allele by methylation of DNA

zygote expresses only one allele of the imprinted gene

located on autosomes, not on x chromosome

extran-uclear genes	located in mitochondria and chloroplasts

dna in these organelles is small, circular, carries only a small # of genes

linked to several severe and rare inherited diseases in humans

since products of mito. genes involved w energy production

defects (mutations) in these genes cause weakness and deterioration in muscles

mito. dna is inherited only from mother bc fathers mito. do no not enter egg during fertilization

genes and the environment

environment can alter the expression of genes

in fruit flies, vestigial wings can be altered by temp

when raised in hot environment, can grow wings almost as long as normal wild type wings

many human diseases have a multifactorial basis

is an underlying genetic component w a significant environmental influence

ex: heart disease, diabetes, cancer, alcoholism, schizophrenia, and bipolar disorder

genes and the environment (cont)

also development of intelligence is result of interaction of genetic predisposition and the environment or nurture and nature

penetrance

proportion or percentage of individuals in a group w a given genotype that actually shows the expected phenotype

ex is breast cancer allele who don't get breast cancer (pg 138)

sex linkage

46 chromosomes

44 are autosomes

2 sex chromo-somes	X and Y

few genes carried on Y chromosome

Females XX	can inherit two copies of the sex linked genes

can be carrier

Males (XY)	only inherit one X linked gene

recessive sex linked is more common than dominant sex linked

males suffer w sex linked more than females

ex for recessive six linked traits	color blindness, hemoph-ilia, Duchenne muscular dystrophy

all daughters of affected fathers are carriers

sons CANNOT inherit sex linked traits from father bc son inherits Y chromosome from him

son has 50 % chance of inheriting sex linked from carrier mother

mutations

mutations	any changes in the genome

can occur in somatic cells and be respon-sible for cancer

or during gameto-genesis	affect future offspring
radiation and certain chemicals cause mutations	but when and where is random
two types:	gene and chromosomal
gene mutations	caused by change in DNA sequence

some human genetic disorders caused by both types

gene mutations cannot be seen under a microscope

chromosomal can

karyotype show size, number and shape of chromosomes

can reveal presence of certain abnorm-alities

can be used to scan fro chromosomal abnormalities in developing fetuses

3 conditions that occur from nondisjunction in formation of ovum or sperm

ADD ACTUAL MUTATIONS ANDDDDDDD LINKAGE MAPPPP

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