Ivan's Chapter 7 Study Guide
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Autosomal Genes in Humans

- Autosomes are chromosomes which do not play a role in sex determination
- Your sex is determined by the sex chromosomes that you have
  - XX = Female
  - XY = Male
- Most traits present in sexually reproducing organisms are determined by autosomal genes
  - Ex: Skin color, eye color, hair shape, etc.
- A good deal of genetic/chromosomal disorders are also caused by autosomal genes

Recessive Genetic Disorders

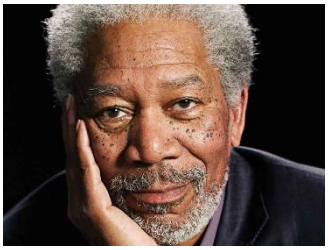
- Most genetic disorders are caused by recessive alleles
  - Both copies of the recessive allele must be present for the disorder to occur
- Heterozygous individuals who have an allele for a disorder but do not show symptoms are called **carriers**
- This shows why having a deep gene pool is important, since healthy dominant alleles can mask harmful recessive ones

Dominant Genetic Disorders

- Less common than recessive disorders
  - Only one copy of the dominant allele has to be present in order for symptoms to show

Sex-Linked Genetic Disorders

- Since men only have one X chromosome, they are more susceptible to X-linked genetic diseases than women
  - Since women have two X chromosomes, the normal one can mask over the faulty one
- Besides having sex-linked genes, the X chromosome also contains other genes which are not related to sex
  - However, the Y chromosome does not contain non-sexual genes
- Scientists believe that the Y chromosome is smaller than the X chromosome since over millions of years of meiosis, genes from the Y chromosome could have transferred over to the X chromosome
  - Since the Y chromosome is smaller than the X chromosome, it contains less genetic information
- Only women can be carriers of sex-linked genetic disorders



Morgan Freeman's autosomes code for traits such as curly hair and dark skin, whereas his sex chromosomes are what makes him a male

- *Sex-Linked Genetic Disorders Cont.*
- **X chromosome inactivation** is a process in which one of the two X chromosomes in a female cell is randomly inactivated
    - One of the X chromosomes coils more tightly than the other to deactivate itself

Hetero. Carrier		
Hetero. Carrier	P	p
P	PP	Pp
p	Pp	pp

The offspring of two hetero carriers for PKU (which is recessive) would have a 50% of becoming carriers and a 25% chance of having the disease

Hetero (Diseased)		
Homo recessive	H	h
h	Hh	hh
h	Hh	hh

The offspring of a Hetero with Huntington's Disease and a healthy homo recessive would have a 50% chance of having the disease and a 50% chance of not

Bald Male		
Normal Hetero Fem	X^b	Y
X^B	$X^B X^b$	$X^B Y$
X^b	$X^b X^b$	$X^b Y$

The offspring of a man with pattern baldness (sex-linked) and a normal hetero woman would have a 25% chance of being normal female carriers, a 25% chance of being bald females, a 25% chance of being normal males, and a 25% of being bald males

Incomplete Dominance

- The phenotype is in-between
- Red flower x white flower = 100% Pink Flowers

	R	R
W	RW	RW
W	RW	RW

Incomplete vs. Codominance

Codominance: Both equally expressed
Red flowers + White flowers =

Incomplete Dominance: alleles blend
Red flowers + White flowers =

Incomplete vs Codominance

Codominance

Both phenotypes are equally present

- Homo. red bull x homo. white cow = 100% Roan Cows

	C^r	C^r
C^w	C_rC_w	C_rC_w
C^w	C_rC_w	C_rC_w

Polygenic Traits

- **Polygenic Traits** are traits which are produced by two or more genes
  - For example, skin color results from four genes which interact
  - Literally means "many [poly] genes [genic]"
- Eye color is determined by a combination of at least three genes
  - The allele for brown eyes dominates over the allele for green eyes which dominates over the allele for blue eyes
  - Brown > Green > Blue

GENE	DOMINANT	RECESSIVE
BEY1	Brown	Blue
BEY2	Brown	Blue
GEY	Green	Blue

The alleles for brown eyes dominate over those for green and blue

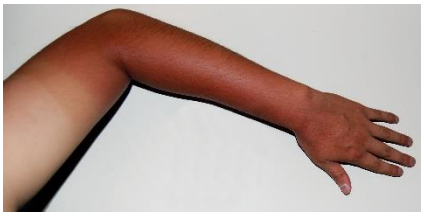
Epistasis

- **Epistatic genes** are genes for polygenic traits which overshadow all of the others
  - A single gene interferes with the other four genes for mouse fur color, causing albinism

Phenotypes, genotypes, and the Environment

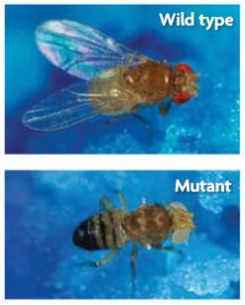
- Height can be influenced by genes
- Height can also be influenced by the environment
  - Ex: Getting more nutrients in the uterus, healthcare, chemicals, etc.
- Hair color is determined by genes, but can also become lighter after prolonged exposure to sunlight
- Skin color is also determined by your genes, but can also darken (tan) after prolonged exposure to sunlight

Despite being born with a paler complexion, this person's skin has darkened due to repeated UV exposure



Genetic Linkage and Fruit Flies

- Thomas Hunt Morgan worked with fruit flies (*Drosophila melanogaster*)
  - Observed easily identifiable characteristics such as eye color, body color, and wing shape
- A **wild type** is the most common natural phenotype of a species
- Morgan bred wild type fruit flies with mutant ones
  - Didn't always follow 9:3:3:1 ratio but still differed in a noticeable pattern
- The crosses showed that some traits seemed to be inherited together
  - Morgan called these "linked traits"
  - Appeared to fall into four groups
    - Corresponds to the four chromosome pairs of a fruit fly
- Morgan concluded that linked genes were on the same chromosome
- Also concluded that chromosomes exchange homologous genes during meiosis



A wild type fruit fly vs a wingless mutant

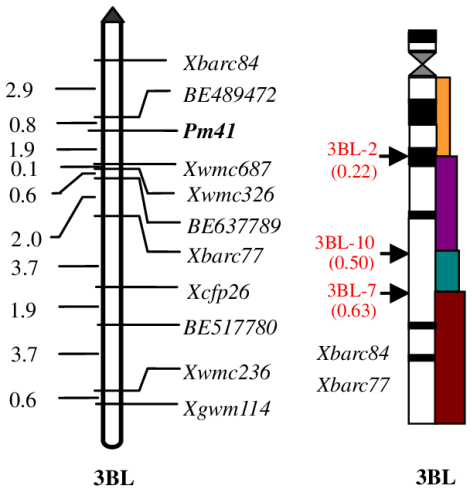
Linkage Maps

- **Linkage maps** are maps of the locations of loci (genes) on the chromosome
  - One map unit = 1 cross-over for each 100 offspring or 1% point
- The probability of two genes being inherited is related to their distance
  - Closer genes are more likely to be inherited together
- Hypothesized by Alfred Sturtevant
  - Identified three linked traits in fruit flies
    - Body color, eye color, and wing size
  - Sturtevant performed many crosses on the flies
- Sturtevant recorded the % of times that the phenotypes didn't appear in the offspring
  - Represented by frequency of cross-overs between chromosomes

Making a Linkage Map

- Suppose that you performed a few crosses and this data was your result;
  - Gene A and B cross over 3% of the time
  - Gene B and C cross over 13.5% of the time
  - Gene A and C cross over 23.5% of the time
- On a linkage map, genes A and B would be 3 units apart since they cross over 3% of the time
- Genes B and C would be 13.5 units apart since they cross over 13.5% of the time
- Genes A and C would be 23.5 units apart since they cross over 23.5% of the time
- From this data, we can conclude that gene B would be between genes A and C

An example of a more complicated linkage map

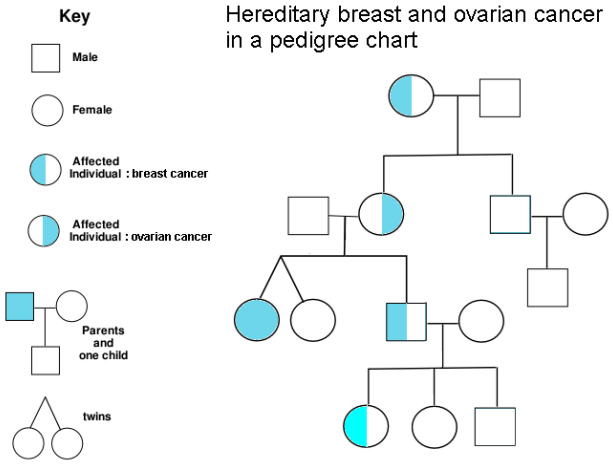


Pedigrees and Tracing Genes

- **Pedigree** charts are used to trace the genotypes/phenotypes which run in a family
  - Determines which family members carried recessive alleles
- Boxes represent males and circles represent females
- A shaded shape indicates that the person shows the trait
- An unshaded shape indicates that the person didn't show the trait
- A half-shaded shape indicates that the person is a carrier
- If a trait occurs in similar numbers among males and females then it is likely an autosomal trait
- However, if it occurs mostly in males then it is likely a gene on the X chromosome

Karyotypes

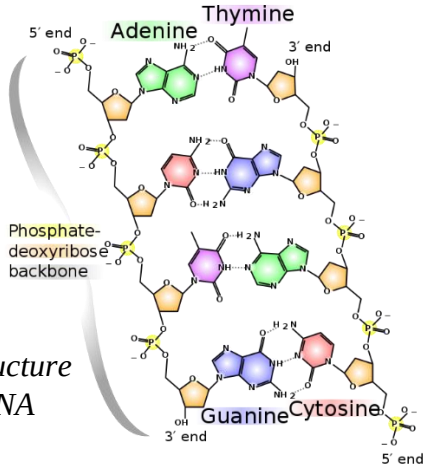
- **Karyotypes** are maps of all the chromosomes in a cell
- Chemicals are used to stain the chromosomes
  - This produces a banded pattern
- Chromosomes are paired by similarities between their size, shape, and bands
  - Cut and pasted according to size (largest => smallest)
- Karyotypes tell us about an organism's…
  - Sex
  - Possible genetic disorders
  - Species



An example of a breast & ovarian cancer pedigree

Genetic Mutations

- Genes give the correct order of amino acids to be assembled into proteins
- Nucleotides (DNA) are made from
  - Bases
    - Adenine, thymine, guanine, and cytosine
  - Sugars
  - Phosphate groups
- Nucleotides are changed when bases are added, removed, or swapped
- Examples of Genetic mutations include PKU, Tay-Sachs, Hemophilia, Cystic Fibrosis, and Huntington's disease



The structure of DNA

Genetic Disorders – PKU (Phenylketonuria)

- Caused by a recessive gene
- Babies are born without the protein needed to synthesize an enzyme needed to degrade the amino acid called phenylalanine
- Phenylalanine builds up in the brain where it can lead to intellectual disability
- Requires a special diet to prevent the intake on phenylalanine
- Incurable but treatable

Genetic Disorders – Tay-Sachs Syndrome

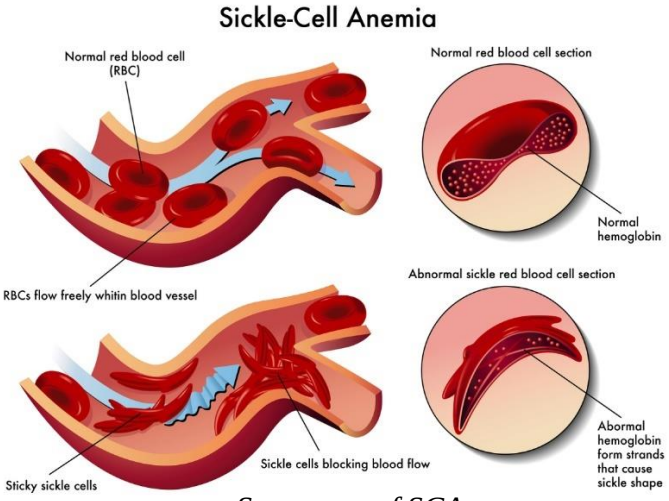
- An enzyme which breaks down lipids in the brain is missing
- The accumulation of these lipids leads to death
- More common in central European Jews
  - Due to central European Jews having a shallow gene pool
- Also caused by a recessive gene
- Since there is no treatment, patients usually die early in childhood

Genetic Disorders – Hemophilia

- Another recessive gene
- Associated with sex chromosomes
- Individuals with hemophilia lack the proteins required to create clots
  - Platelets
- Common in nobility
  - Shallow gene pool

Genetic Disorders – Sickle Cell Anemia

- Caused by a recessive gene which codes for hemoglobin
- Most common in African populations
  - Reduces severity of malaria infections
- Causes blood cells to curve into crescent shapes
  - Mutation to protect against malaria
- Carriers have little symptoms and tend to live normal lives
- Individuals with SCA tend to make it to age 40
- Panama Canal story



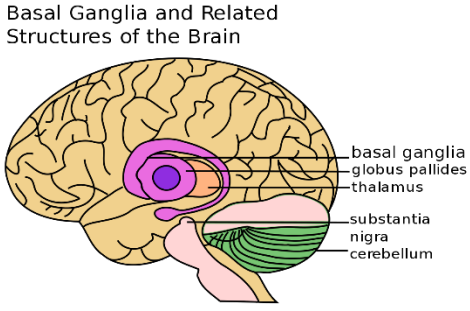
Symptoms of SCA

Genetic Disorders – Cystic Fibrosis

- Caused by a recessive gene on chromosome #7
- Causes a thick mucus to be produced around the lungs, liver, and pancreas
  - Damages these organs
  - Makes breathing difficult
- No cure, often die at a young age

Genetic Disorders – Huntington's Disease

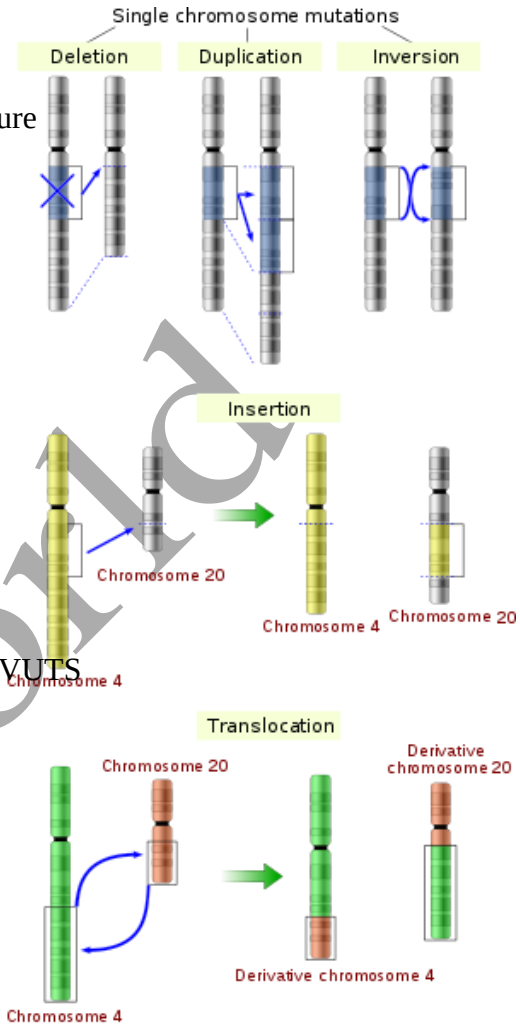
- Caused by a dominant gene
- Brought to the colonies in 1630 by three men whose families had been persecuted in England for witchcraft
  - Displayed strange behavior
- Results in mental and physical deterioration
  - Causes uncontrollable trembling
- Starts at ages 30-40
- Eventually the person dies



Huntington's mainly affects the basal ganglia

Chromosomal Mutations

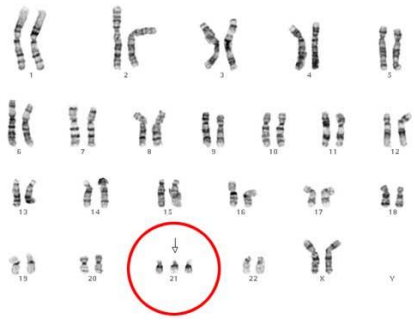
- Chromosomal mutations are changes in a chromosome's structure or the number of chromosomes
- Can be caused by mutagens
  - Agents which alter the genes of an organism
- Can also be caused by improper separation of chromosomes during meiosis
  - This is known as a nondisjunction
  - Gametes receive more or less than appropriate
- Deletions
  - ABCDEFGH => ABCDGH
- Inversion
  - ABCDEFGH => ABDCEFGH
- Duplication
  - ABCDEFGH => ABABCDEFGH
- Translocation
  - [1] ABCDEFGH [2] ZYXWVUTS => [2] ABCZYXWVUTS



A chart of chromosomal mutations

Chromosomal Disorders – Down Syndrome

- Caused by three copies of the 21st chromosome (trisomy 21)
- Caused by a nondisjunction of chromosomes
  - Chromosomes didn't pull apart during meiosis
- Individuals display intellectual disability and physical differences
- Frequency of down syndrome increases with the mother's age

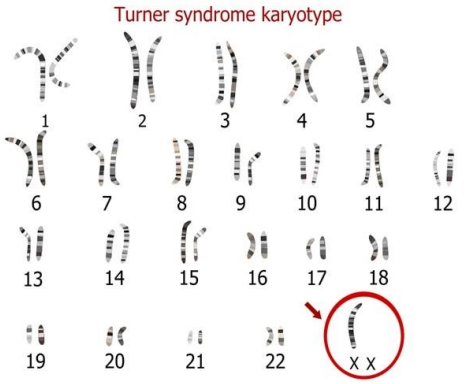


Notice how the karyotype has a third copy of the 21st chromosome

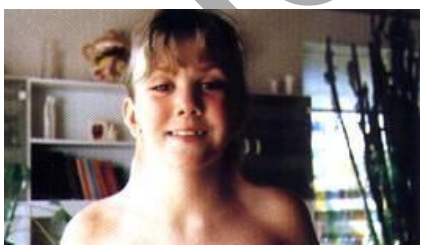
A man with Down Syndrome showing common facial characteristics

Chromosomal Disorders – Turner Syndrome

- Genotype XO = X
  - 45 chromosomes
  - Sterile females
- Sex organs are not mature
- No pubic hair
- Immature ovaries
- Caused by a nondisjunction of chromosomes



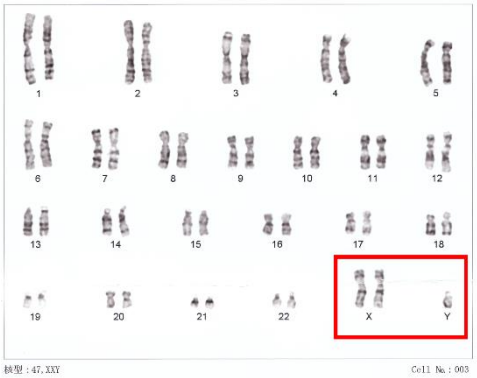
Notice how there is only one X chromosome



A girl with Turner syndrome showing characteristic neck-webbing

Chromosomal Disorders – Klinefelter's Syndrome

- Genotype XXY
  - 47 chromosomes
- Nondisjunction
- Sterile males
- Several physical defects
  - Immature sex organs
  - Taller than normal
  - Weaker muscles
  - Increased breast tissue
- Some degree of cognitive impairment may be present

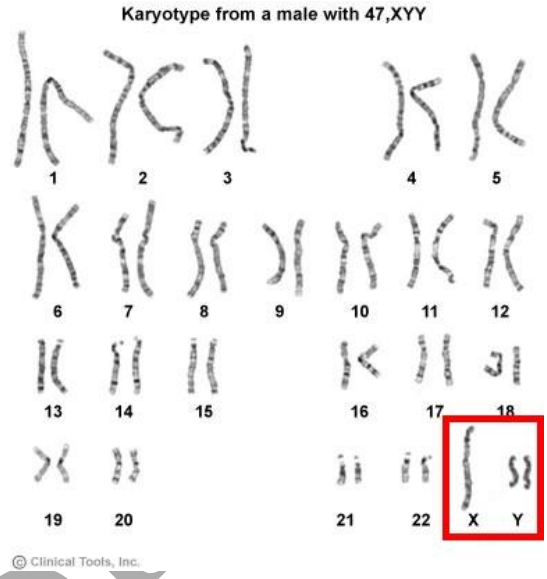


Notice the inclusion of two X chromosomes and a Y chromosome

A man with Klinefelter's

Chromosomal Disorders – XYY "Super male" Syndrome

- Genotype XYY
  - 47 chromosomes
- Nondisjunction
- Sterile males
- Besides being taller than average, most men with the syndrome tend to look normal
- Many are in prisons and mental facilities
  - Increased aggression from higher levels of testosterone
- Don't have abnormal sex organs



Notice the presence of an extra Y chromosome in this man's karyotype

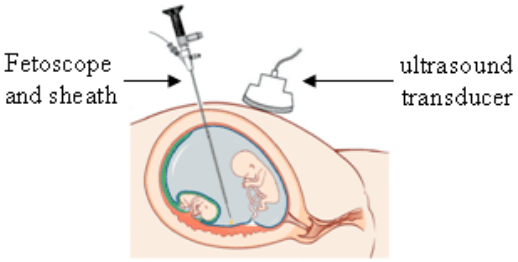
Chromosomal Disorders – Triple-X "Metafemale" Syndrome

- Also known as trisomy X
  - Inclusion of three X chromosomes
    - 47 chromosomes in total
- Often taller than average women
- Occasionally born with increased eye distance and epicanthal folds
- Some experience learning difficulties

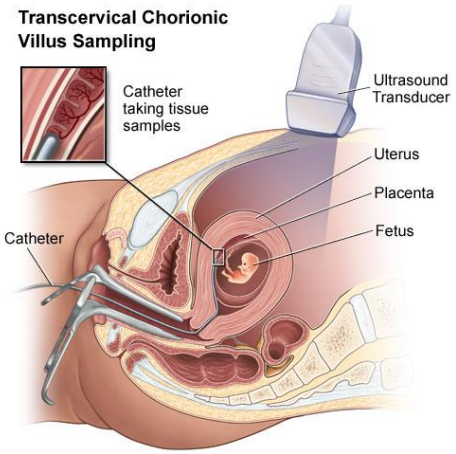
A young girl with increased eye distance as a result of the syndrome

Detecting Chromosomal Abnormalities

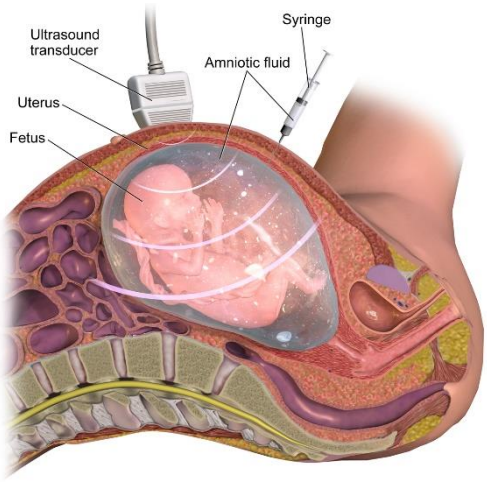
- Ultrasonography
  - Uses sounds above our range of hearing to create an image of the fetus
- Fetoscopy
  - Uses a fetoscope pushed through a hole in the abdomen to capture an image of the fetus
- Chorionic Villus Sampling
  - Involves the removal of a small amount of tissue from the placenta
- Amniocentesis
  - A syringe is pushed through the abdomen and into the amniotic sac where a small sample of amniotic fluid is drawn
- Karyotyping
  - See the section on page 5



A fetoscopy



Chorionic Villus Sampling



Amniocentesis

PRANKED
BOI

The wonderful message which somebody left in my notes while I was away from my computer