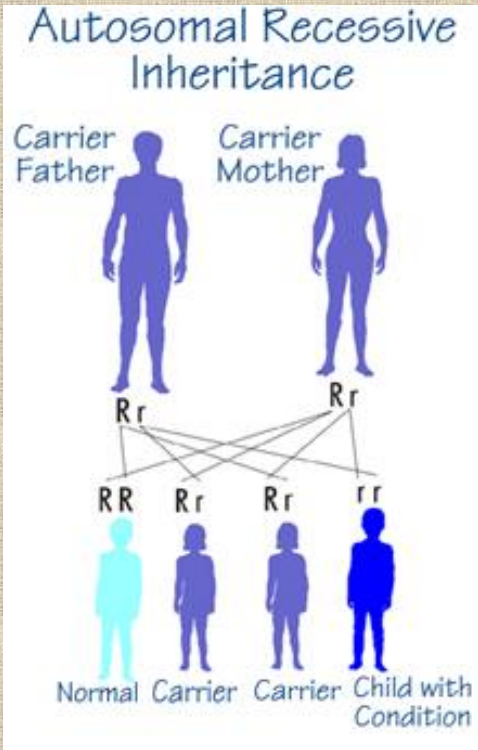
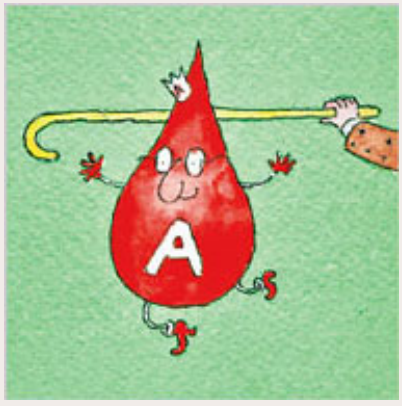
Human Genetics



12. Multiple Alleles -genes with 3+ alleles

a) ABO blood group
b) A/B codominant, O is recessive
c) O universal donor
d) AB universal recipient
e) Defined by protein/antigen
   Type O, none, AB-both
f) AB antibodies
   B has antibody A, etc.



Codominance in blood groups

Phenotype (blood group)	Genotypes	Antibodies present in blood serum	Reacts (clumps) when red blood cells from groups below are added to serum from groups at left?			
			O	A	B	AB
O	*ii*	Anti-A Anti-B	No	Yes	Yes	Yes
A	$I^A I^A$ or $I^A i$	Anti-B	No	No	Yes	Yes
B	$I^B I^B$ or $I^B i$	Anti-A	No	Yes	No	Yes
AB	$I^A I^B$	—	No	No	No	No

Q. What genotypes are responsible for producing the following blood group phenotypes?

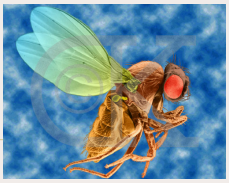
Type O: ii

Type A: I^Ai, I^AI^A

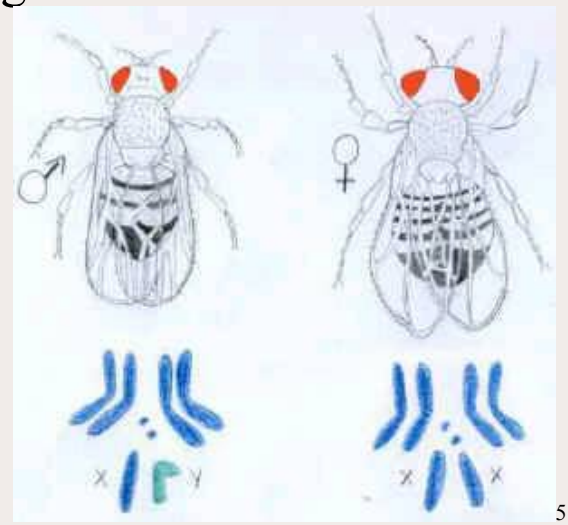
Type B: I^Bi, I^BI^B

Type AB: I^AI^B

13. Sex Chromosomes vs sex linked



a. Fruit Fly- Morgan discovered sex chromosomes



13. continued

B. sex determined by male in humans

C. Gestationally males express SRY gene on Y chromosome (anything Y is Y linked)

D. X chromosome- X linked- "problems" mostly seen in males

E. Sex-Influenced- different phenotypes due to gender- pattern baldness

14. Linked genes

A. Genes that tend to be inherited together
B. Found on the same chromosome in close proximity
C. Chromosome map- shows linear order of genes
D. % of crossing over is proportional to the distance between 2 genes
E. One map unit = 1%

15. Mutations- heritable changes in DNA

A. Gene- involve a few nucleotides
B. Chromosome- more than one gene
C. Mutagen- chemical or physical agent that promotes mutation
D. Carcinogen- mutagen that causes cancer

C.Nondisjunction-chromosomes

D. Deletion or addition

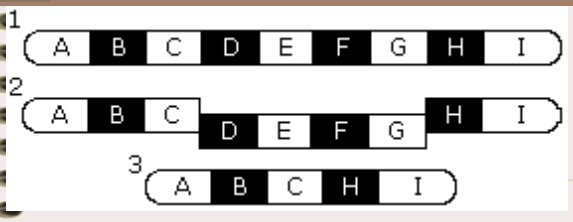
16. Chromosome mutations p. 372-376

a. Germ cell- affect _____

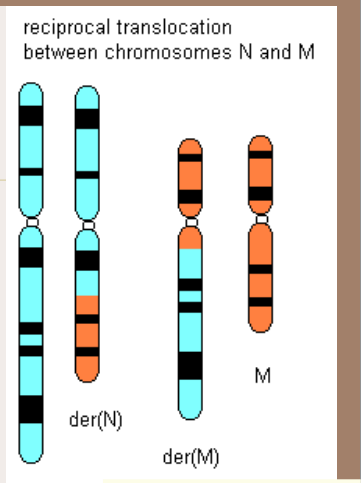
b. Somatic - affect _____

E. Translocations-

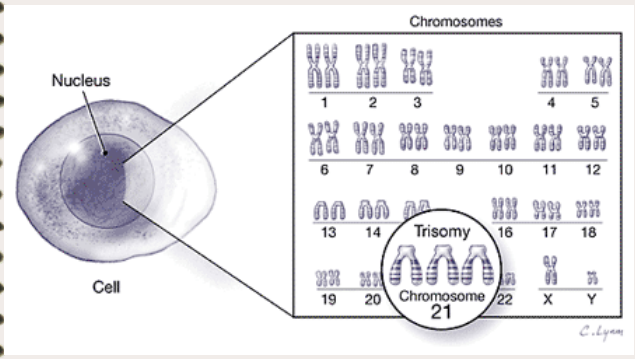
F. Inversion-



Deletion



Translocation



Trisomy 21 Down's Syndrome

A. Point mutation

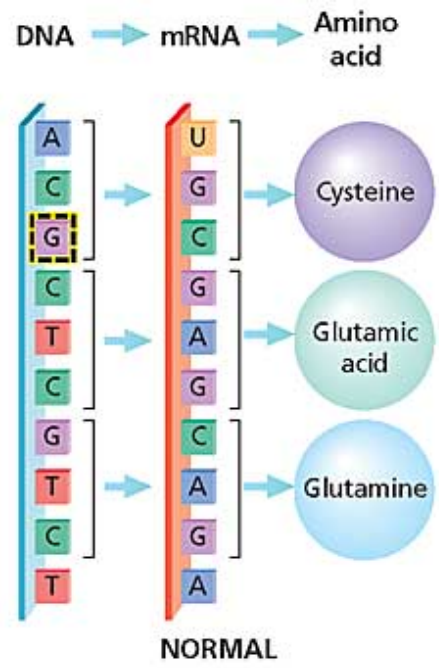
B. Frameshift-

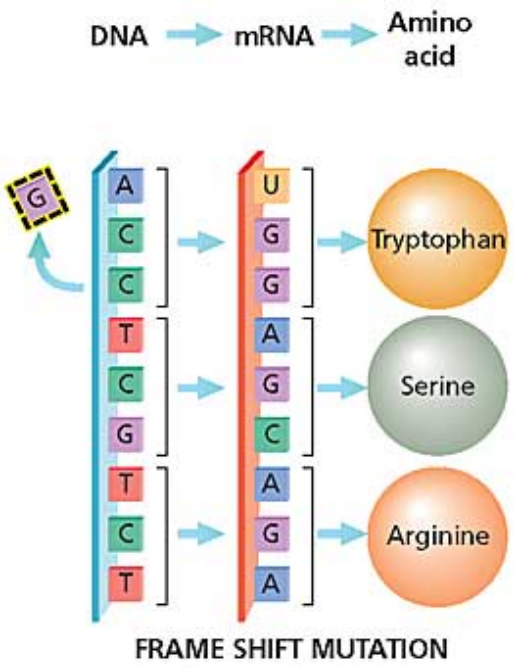
17.

Gene mutations
page 373

C. Substitution

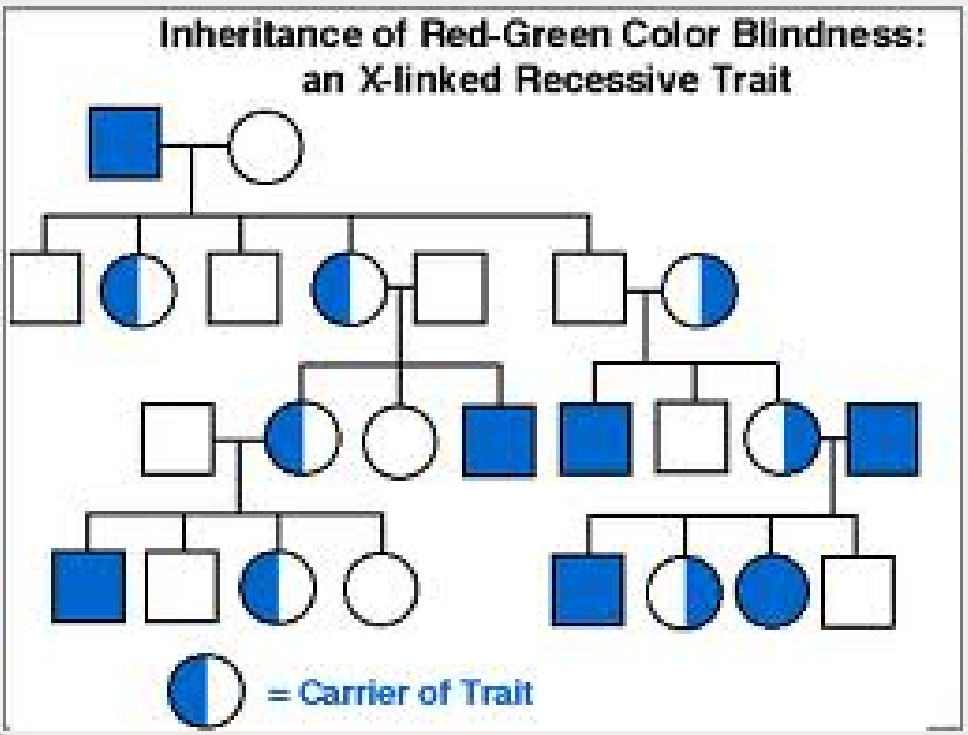
D. Insertion/ deletion





18. Pedigree- diagram that shows how a gene is inherited over generations

a. Circles-F, Squares-M
b. Filled in= affected, 1/2 filled=carrier of a recessive trait
c. Horizontal line=mating, vertical lines indicate offspring
d. Recessive if trait does not appear in parents but DOES appear in kids (dominant if it's in every generation)
e. Autosomal if both genders are affected (sex linked if predominantly one gender)



19. Polygenic trait

A. <u>trait</u> influenced by many genes (weight, skin color, eye color, intelligence)

In nature: Corn cob length, bill/beak length

They show a wide range of phenotypes!

C.Nondisjunction- chromosomes do not come apart during meiosis, trisomy occurs

16.

Chromosome mutations p. 372-376

a. Germ cell- affect offspring

b. Somatic - affect organism

D. Deletion or addition -portion deleted/added

E. Translocations- portion(s) break off and go somewhere else.

F. Inversion- portion breaks off then reattaches in reverse orientation

A. Point mutation change in 1 nucleotide

17.

Gene mutations affect the gene

B. Frameshift- addition or loss means all codons will be a problem

C. Substitution- one nucleotide is substituted for another

D. insertion nucleotide is added to a gene