

Genes, Chromosomes & Human Heredity



Inheritance of Traits
Sex Linked Inheritance
Genetic Disorders
Mutations

EPISTASIS

An interaction between genes such that one gene masks the expression of the phenotype of another gene.

recessive epistasis

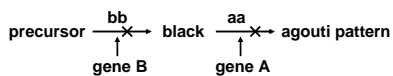
mouse coat color A_ = agouti
aa = black

In a cross thought to be Aa x Aa see 9 agouti : 3 black : 4 albino.

Modified dihybrid ratio implies 2 genes involved
(9:4:3 is typical recessive epistatic ratio for 2 genes)

Can explain results if cross is actually AaBb x AaBb

9 A_ B_	agouti	
3 A_ bb	albino	
3 aa B_	black	b is epistatic to A or a.
1 aabb	albino	



dominant epistasis

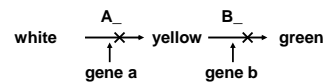
squash color A_ = white
aa = yellow

In a cross thought to be Aa x Aa see 12 white : 3 yellow : 1 green.

Modified dihybrid ratio implies 2 genes involved
(12:3:1 is typical dominant epistatic ratio for 2 genes)

Can explain results if cross is actually AaBb x AaBb

9 A_ B_	white	
3 A_ bb	white	A is epistatic to B or b.
3 aa B_	yellow	
1 aabb	green	



INCOMPLETE DOMINANCE

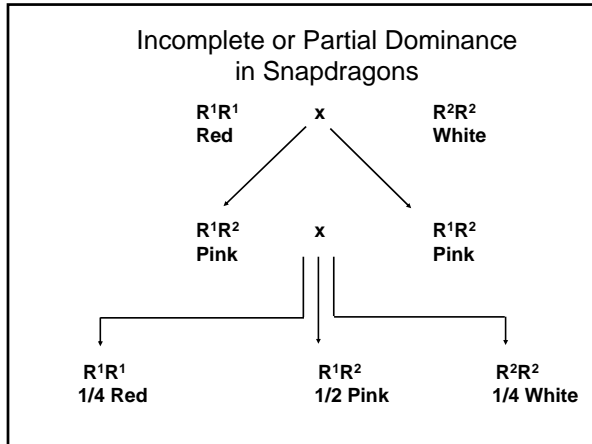
Where a heterozygote has a phenotype intermediate between the corresponding homozygote phenotypes.

INCOMPLETE DOMINANCE

Flower Color in Snapdragons




Pink
White
Red



Pleiotropy

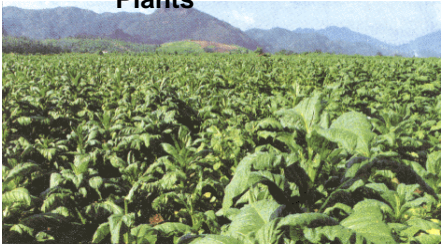
condition in which a single mutation simultaneously affects several characters

- **White eye gene in Drosophila**
 - eye color white
 - flight muscles defective
 - **Albinism in humans**
 - lack of pigment
 - eye sight can be affected
 - hearing can be affected
- 
- http://www.albinism.org/publications/what_is_albinism.html

POLYGENIC INHERITANCE

http://www.tenset.co.uk/ba/gen_poly.html


Kolreuter's Tobacco Plants



Polygenic Inheritance

Interactions between two or more genes, each with an additive effect on the character.

Fruit Flies



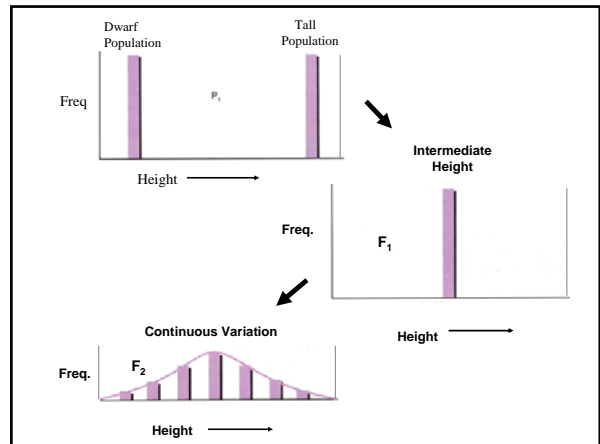
An Explanation of Kolreuter's Results

•Residual = 2 ft
 •A = B = 1ft added each
 •a = b = 0ft added each

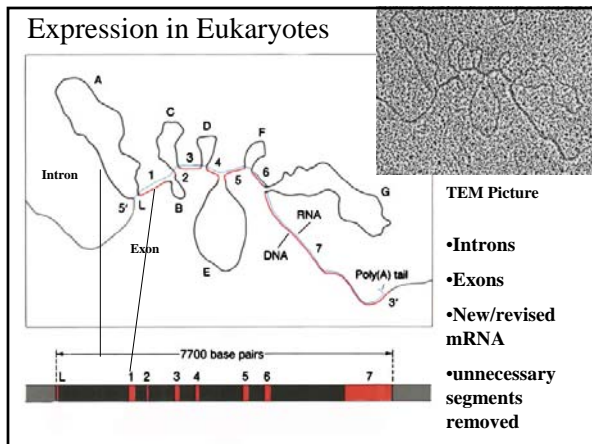
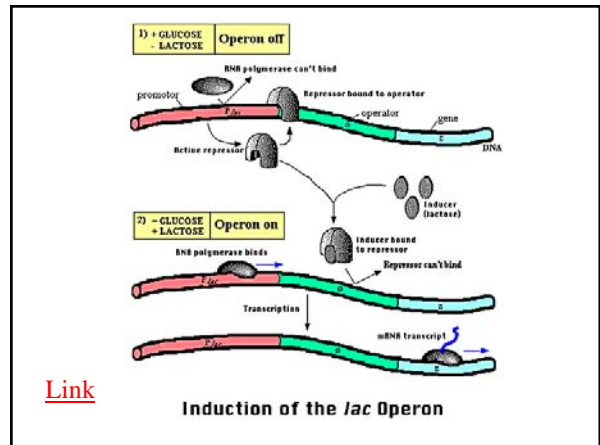
Parents	AABB 6 feet	X	aabb 2 feet	
F₁	AaBb	X	AaBb	
F₂	AB	Ab	aB	ab
AB	AABB	AABb	AaBB	AaBb
Ab	AABb	AAbb	AaBb	Aabb
aB	AaBB	AaBb	aaBB	aaBb
ab	AaBb	Aabb	aaBb	aabb

Summary Polygenic Inheritance F₂ Results

Genotypes	Phenotypes	Frequency	
AABB	6 feet tall	1	1
AABb	5 feet tall	2	4
AaBB	5 feet tall	2	
AAbb	4 feet tall	1	6
AaBb	4 feet tall	4	
aaBB	4 feet tall	1	
Aabb	3 feet tall	2	4
aaBb	3 feet tall	2	
aabb	2 feet tall	1	1



- ### Gene Expression in Prokaryotes (ch. 10)
- Within its tiny cell, the bacterium *E. coli* contains all the genetic information it needs to metabolize, grow, and reproduce.
 - It can synthesize every organic molecule it needs from glucose and a number of inorganic ions.
 - Refer to Pg. 216 in text book



The Inheritance of Human Traits

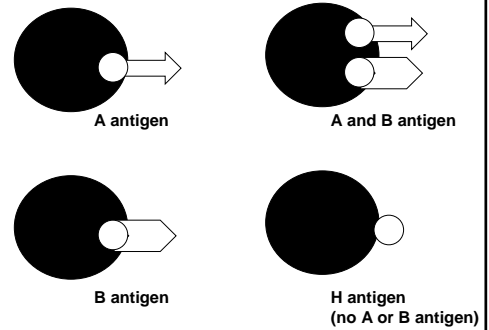
Blood Groups
 Huntington Disease
 Sickle Cell Anemia
 Polygenic Traits

Multiple Alleles & Codominance

codominance - where the heterozygote shows the phenotype of both alleles equally

- **ABO Blood Groups**
- **Discoverer -- Landsteiner (1900-1901)**
Group A, B, O, AB
- **Genetic Explanation -- Bernstein (1924)**
Series of multiple alleles
 $I^A, I^B, I^O (i)$

Red blood cell antigens



Genotype	Phenotype	Frequency	Antigen	Antibody in Serum
$I^A I^A$ $I^A I^O$	Blood Group A	41%	A	Anti-B
$I^B I^B$ $I^B I^O$	Blood Group B	10%	B	Anti-A
$I^O I^O$	Blood Group O	45%	Neither	Anti-A Anti-B
$I^A I^B$	Blood Group AB	4%	Both	Neither

Note: Caucasian frequencies given. Frequencies vary between populations.

- **Huntington Disease** located on gene 4
 - Painful loss of muscle control
 - loss of mental function
 - Death
- **Sickle Cell Anemia**
 - Bent & twisted red blood cells
 - Deprived of oxygen
 - Ridged and get stuck in capillaries
 - Damage to cells and tissues
 - Could result in death

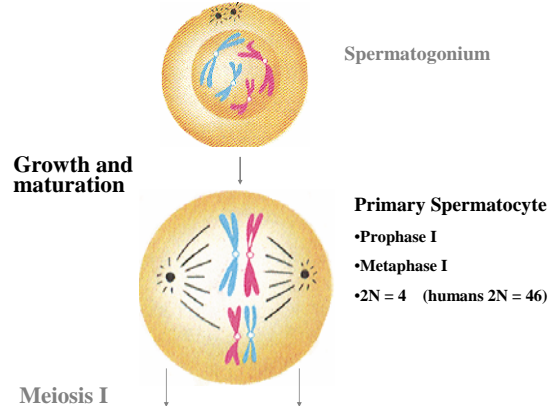
Sex-Linked Inheritance

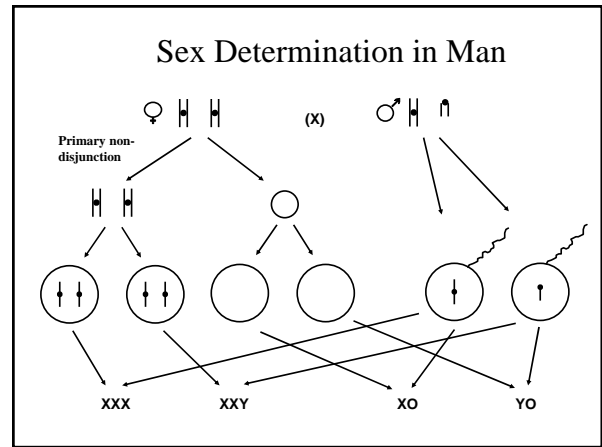
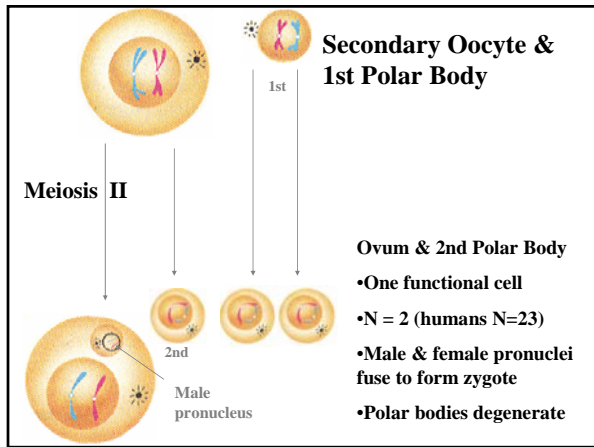
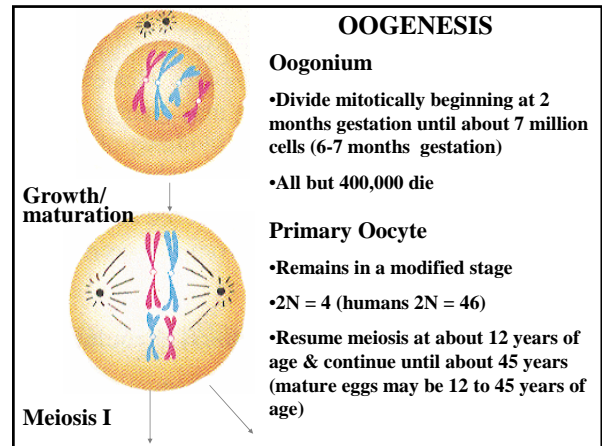
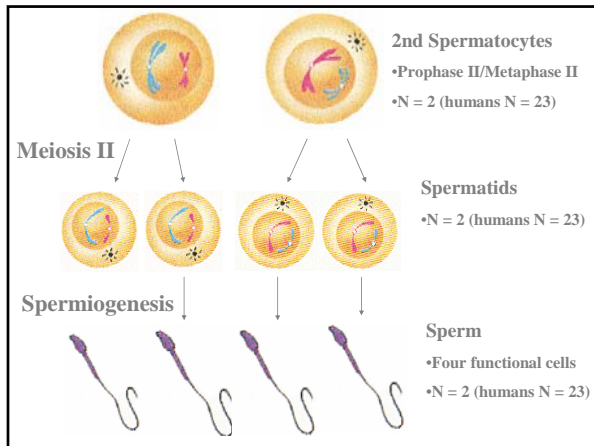
Gamete Formation

Homo sapiens



SPERMATOGENESIS





Aneuploid Offspring

XXX -- Trisomy-X -♀
 XXY -- Klinefelter -♂
 XO -- Turner-♀
 YO -- Dies

XXX Syndrome

- ☐ 47, XXX female
- ☐ 1/1200 female live births
- ☐ Variable phenotype, frequently appear normal, sterile, may show mental retardation. Under-developed secondary sex characteristics
- ☐ Many institutionalized

Klinefelter Syndrome



- ☐ 47, XXY male
- ☐ 1/500 male live births
- ☐ Male genitalia, testes undeveloped, fail to produce sperm
- ☐ Female secondary-sex characteristics

Turner Syndrome



- ☐ 45, XO female
- ☐ 1/3000 female live births
- ☐ Ovaries rudimentary, sterile
- ☐ Short stature, shield-like chest, webbed neck
- ☐ Normal intelligence

XYY Condition

- ☐ 47, XYY male
- ☐ 1/1000 male live births
- ☐ Above-average height, 2% in maximum security prisons are XYY, personality disorders, subnormal intelligence
- ☐ Fertile
- ☐ Many XYY males socially normal

Human Sex-Determination Mechanism

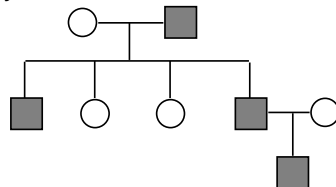
♂	♀
XY	XO
XXY	XX
XYY	XXX
XXYY	XXXX
XXXY	XXXXX
XXXXY	

CONCLUSIONS:

- ☐ Presence of Y is male-determining
- ☐ Lack of Y is female-determining
- ☐ Y carries testis-determining factor (TDF)

Y-Chromosome Linkage in Man : Holandric Inheritance

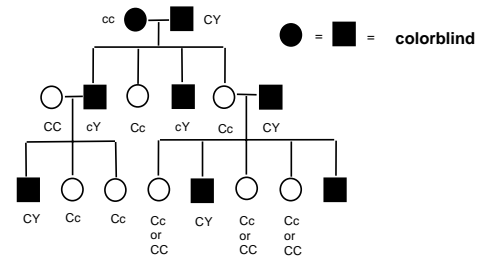
Hairy Pinna



- ☐ Always inherited father to son
- ☐ Gene found only on Y-chromosome

Inheritance of Sex-Linked Traits

Color Blindness



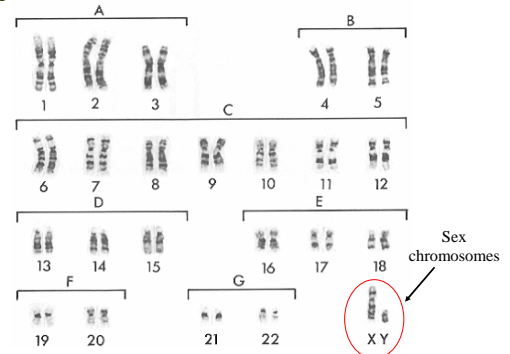
colorblindness is a sex-linked recessive trait

SEX-INFLUENCED TRAITS

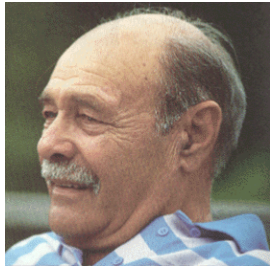
- **sex chromosome** - a chromosome whose presence or absence is correlated with the sex of the bearer, or, a chromosome that plays a role in sex determination
- **autosome** - any chromosome that is not a sex chromosome
- Caused by genes located on autosomes, not on sex chromosomes

Human Karyotype

22 pairs of autosomes and 1 pair of sex chromosomes

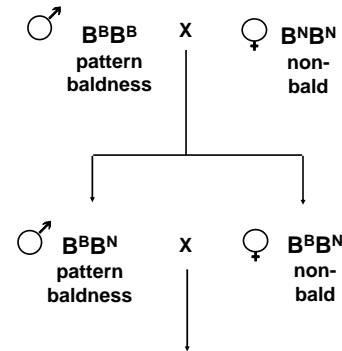


Male Pattern Baldness



Sex-influenced trait: Autosomal trait expressed as a dominant in one sex and as a recessive in the other.

Sex-Influenced Trait Inheritance Pattern

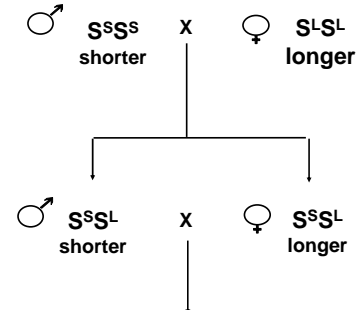


Pattern Baldness F₂ Summary

Genotype	Phenotype	
	Females	Males
1 B ^B B ^B	Bald	Bald
2 B ^B B ^N	Non-Bald	Bald
1 B ^N B ^N	Non-Bald	Non-Bald

Sex Influenced Trait

2nd Finger Shorter than 4th



2nd Finger Shorter than 4th

F₂ Summary

	<u>Genotypes</u>	<u>Phenotypes</u>	
		<u>males</u>	<u>females</u>
1	S ^S S ^S	Shorter	Shorter
2	S ^S S ^L	Shorter	Longer
1	S ^L S ^L	Longer	Longer

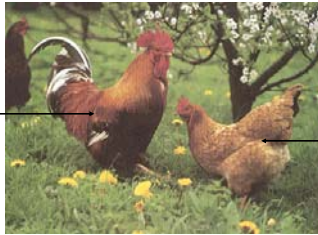
Sex Limited Traits

Autosomal trait expressed in one sex only

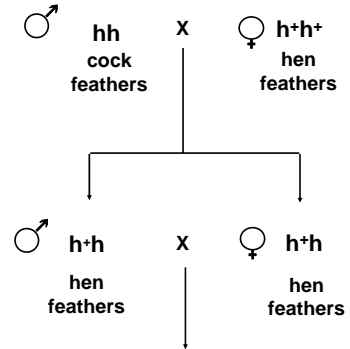


Cock and Hen Feathering in Chickens

Hamburgh rooster (cock feathering)

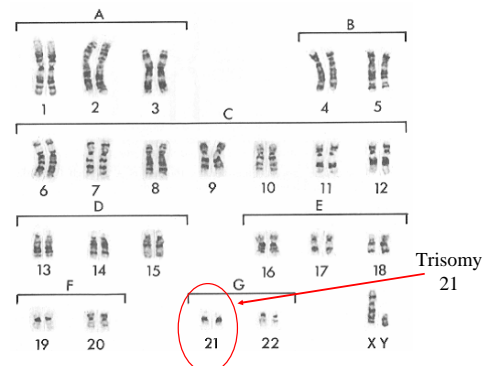


Sebright hen (hen feathering)



Genetic Disorders

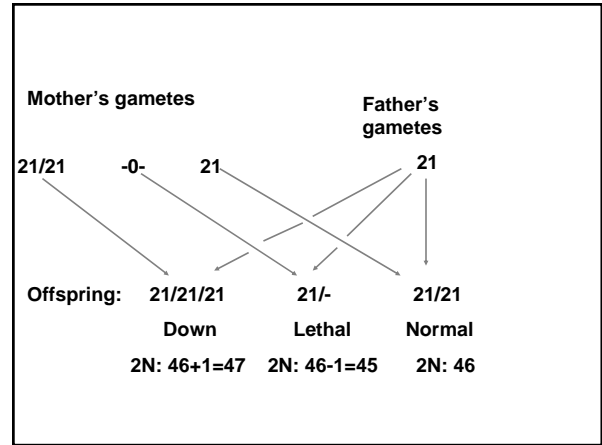
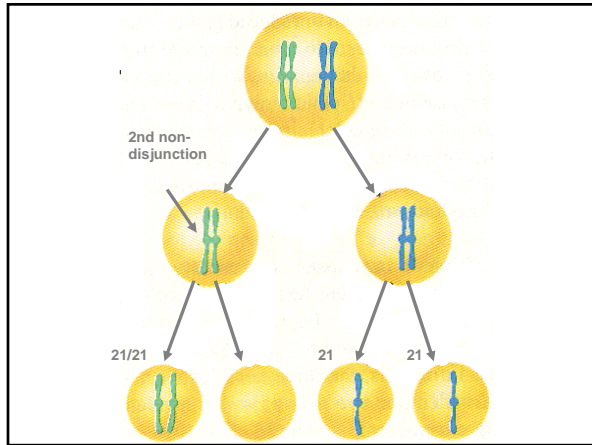
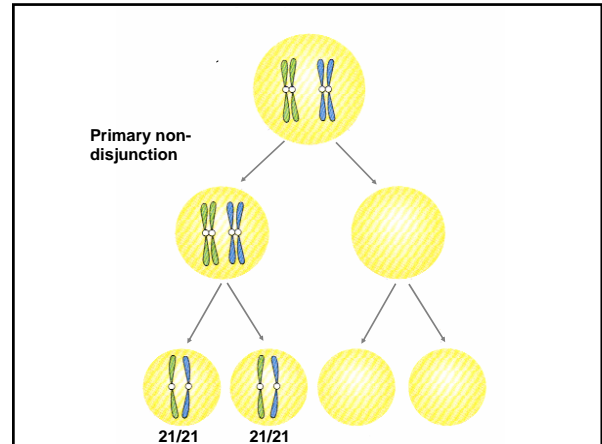
Human Karyotype



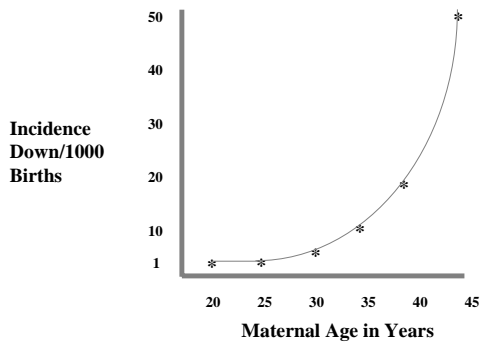
DOWNS SYNDROME PHENOTYPE



- Epicanthic fold in eye corner
- IQ seldom above 70
- Life expectancy short, few survive to 50
- Have simian crease
- May have heart defects
- May have small heads
- May have furrowed tongues



Correlation Down Syndrome and Maternal Age



The End