HUMAN GENETIC TRAITS

compiled by David B. Fankhauser
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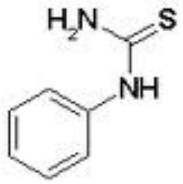
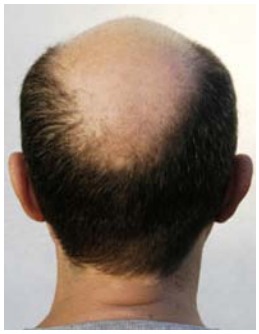
revised 8 April 2016

phenylthiocarbamide:



Genetics text sources used: Griffiths et al,7th p 426; Klug and Cummings, 4th: p 69;Tamarin 4th : p 82-87, Campbells 6th, pp257-266, 9th: 273-304, 10th: 282-290

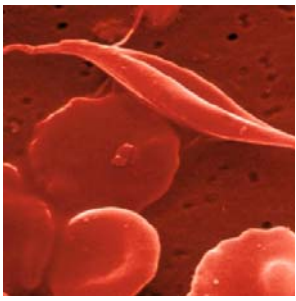
Non-pathological:

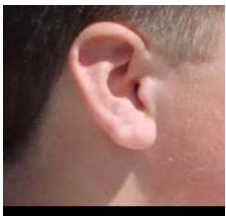


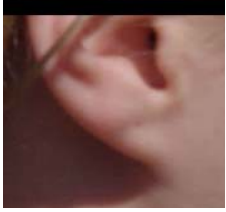
DOMINANT PHENOTYPE	HOMOZYGOUS RECESSIVE PHENOTYPE
Brown eyes	Blue eyes (actually more complex, simplified here)
PTC taster (phenyl thiocarbamide)	PTC non taster
Widow's Peak (p. 282)	Lack Widow's peak
Middigital hair	Hairless mid digits
Tongue roller (questionable inheritance)	Cannot roll tongue
Detached earlobe (image below to right)	Attached earlobe (p. 282)
A and B blood type (codominant) (p. 278)	Type O blood type
Rh positive blood type	Rh negative blood type
Pattern baldness (dominant in males)	Pattern baldness (recessive in ♀)
interlock fingers, L thumb on top	R on top

Pathological: (be familiar with those in bold)

DOMINANT TRAIT	RECESSIVE TRAIT
Achondroplasia (p. 285)	**Albinism** (p. 283)
Brachydactyly (image to left)	Alkaptonuria
Congenital stationary night blindness	Atasia telangiectasia
Ehler-Danlos syndrome	**Color blindness** (sex-linked)
Fascio-scapulo-humeral muscular dystrophy	**Cystic fibrosis**
Huntington disease	Duchenne muscular dystrophy
Hypercholesterolemia	Galactosemia
Marfan Syndrome	**Hemophilia** (sex-linked)
Nail-patella syndrome	Lesch-Nyhan syndrome
Neurofibromatosis	**Phenylketonuria**
Polydactyly	**Sickle-cell anemia** (image to right)
	Tay-Sachs disease







Examples:

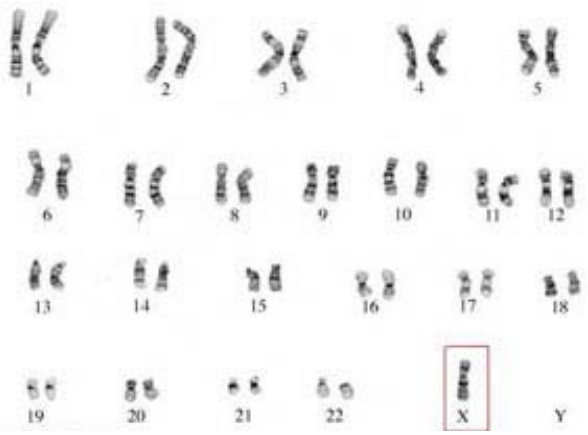
Sickle Cell Anemia	1/400 blacks affected (ss) What % are heterozygous? (roughly sq rt of 1/400 = 1/20)
Phenylketonuria	1/11,000 births
Cystic Fibrosis	1/2500, 1/25 are carriers (dom. protein pumps Cl^- out of cell) usually die 4-5 yrs

TRISOMY: CAUSED BY **NON-DISJUNCTION DURING MEIOSIS**

Sex Chromosome:		freq:	signs: (MM, VX, p. 2148-2150)
Turner's	XO	1/3000	pathognomic: lymphedema of dorsal hands&feet, neck
triple X	XXX	1/1000	little effect, may be sterile, may be retarded
Klinefelter's	XXY	1/700	tall, eunuchoid, sterile, a few are retarded
super male	XYY	1/1000	acne, language dysfunction (1/15 in prisons for violent crimes)

Down's Syndrome: (p 281) all live births 1/700
Increases risk with maternal age after age 45 1/40
49 yrs 1/7

From father in 1/4th to 1/3rd of cases



Blood Type (From Carolina protocol sheet) p 278
ABO example of **codominance**, Rh of classic dominant/recessive alleles

Type	%	Blacks	Am Ind
A:	41	29.5	2.6
B:	10	15.5	0
AB:	4	3.5	0
O	45	51.2	97.4
Rh^+:	85	92	100

SEX LINKAGE: CHROMOSOMAL BASIS FOR SEX

revised 8 Spril 2016
GMSLG 7th: 38-40, Campbell's 6th: 271-279, 7th: 282-290, Sadava: 225-228, Campbell 9th: 286-302, 10th:

Henry VIII kept divorcing or beheading (2) wives because they kept producing daughters...
Whose fault WAS it?
Why is the ratio of male to female births close to 50:50?

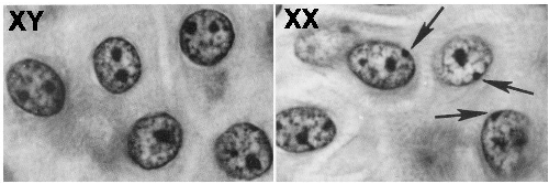
Thomas Hunt Morgan, 1909, used ***Drosophila melanogaster*** for genetic studies:
12 day life cycle, after emerge from pupa, ready to mate 12-14 hrs
Found a single white-eyed male, crossed times red-eyed female: (p 294)

white-eyed male x red-eyed female: (p 295) F_1 = all red.
F_2 = 1/4 white, but were *all* male

Sex determination:	species	male	female
(p 296)	Moths, chickens	ZZ	ZW (heteromorphic)
	Mammals, flies	XY	XX (heteromorphic)
	Hymenoptera	haploid	diploid

Sex-linked (carried on the sex chromosomes, in mammals are **heteromorphic**) vs **autosomal** genes.
crisscross inheritance: daughters like fathers, sons like mother

SEX-LINKED EXAMPLES:
In humans. female = XX
male = XY (a condition called **hemizygous**)



X linked human recessive traits

(show sample pedigrees):	**affected ♂:**
red-green color blind: (p 297)	1/8
hemophilia A, factor XIII missing:	1/2,500
Duchenne's muscular dystrophy: muscle atrophy by 6 yrs, dead by 20:	1/3,500

Calico cats are example, almost always female (1/300 calico is male: XXY, sterile). (p. 298)
Cats carry alleles for coat color on the X chromosome: black or orange.
Calico cat can have black and orange male kittens in same litter. Show cross.

In females, only single X chromosome should function in a cell, inactivate (random) one or other X.
Called **Lyonization** after Barbara Lyon. Inactivated X can be seen: **Barr bodies**

Aneuploidy: incorrect number of chromosomes:

How to tell sex-linked for a rare phenotype:
1) many more males affected
2) None of offspring of affected males affected (usually).
All of his daughters are carriers, half of *their* sons will be affected
3) None of sons of affected male will be affected, will not transmit to offspring

Barr body: