## Sex Linked Traits

X-linkage

## Colorblindness Test http://enchroma.com/test/instructions/

Note:
Set your screen to the brightest setting!


## - - Sex Determination

o Each human body cell (somatic cell) has 22 pairs of autosomes and one pair of sex chromosomes.
o $X X=$ female, $X Y=$ male

- Biological sex is determined at fertilization
- Male sperm cell contains either an $X$ or a $Y$ chromosome
- Female egg cell contains an $X$ chromosome.
- $\quad$ Sex Linkage
- Thomas Hunt Morgan's work with Drosophila (fruit flies) demonstrated that genes for certain traits are located on the $X$ chromosome.
- Why fruit flies?
- Easy to breed
- New generation every 2 weeks
- Only 4 pair of chromosomes

o X-linked disorders are expressed more frequently in males than in females because males only have 1 X chromosome





## X-linked diseases:

1. Hemophilia

- blood clotting disorder
- lack of clotting factor proteins



## X-linked diseases:

2. Colorblindness

- inability to perceive colors in a normal fashion
- most common form is red-green colorblindness



## 3. Adrenoleukodystrophy (ALD)

- breakdown or loss of myelin, the fatty covering surrounding nerve cells in the brain

o X-linked genotypes: Ex. Colorblindness Females:

normal<br>X X<br>or $X^{N} X^{N}$<br>carrier<br>$X^{C} X$<br>affected<br>$X^{C} X^{C}$<br>or $X^{n} X^{n}$

Males:
normal
XY
or $X^{N} Y$
NO male
carriers
affected $X^{C} Y$
or $X^{n} Y$

## Sex Linked Dominance Y-Linkage Mitochondrial Inheritance

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- Most sex linked traits are recessive
- Sex-linked dominance is a rare inheritance pattern
- A single abnormal gene on the $X$ chromosome can cause a sex-linked dominant disease
o There are no "carriers"
olf the father has the abnormal X gene:
-he has the disease
(because it is
dominant)
- ALL of his daughters will inherit the disease
- NONE of his sons will have the disease

X-linked dominant, affected father


X-linked dominant, affected mother
o If the mother has the abnormal X gene:

- she has the disease
- HALF of her children (daughters and sons) will inherit the disease

o Exł Hypertrichosis: excessive hair growth - AKA Werewolf syndrome


Video - Hairiest Girl in the World


Video: hypertrichosis (Larry Gomez)

- Few genes are located on the $Y$ chromosome (it's small)
o present only in males
o disorder would be passed on to all of a man's sons but never to daughters
- Y chromosome infertility
- Azoospermia


## 50 million base pairs

Short stature homeo box, Y-linked
Short stature
Leri-weill dyschondrosteosis
Langer mesomelic dysplasia
Interleukin-3 receptor, Y chromosomal
Sex-determining region $Y$ (testis-determining)
Gonadal dysgenesis, XY type
Protocadherin 11, Y-linked
Azoospermia factors
Male infertility due to spermatogenic failure Growth control, Y-chromosome influenced
Chromodomain proteins
Retinitis pigmentosa, Y-linked

## Mitochondrial Inheritance

- Mitochondria are organelles cellular respiration (energy release)
- They have their own DNA
o Transmission is from mother's egg cell to ALL offspring
o Sons and daughters are equally effected by mutations


| Mitochondrial Inheritance |
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| Ex. Leber's Hereditary Optic Neuropathy (LHON) | - rare condition, can cause sudden painless loss of central vision

