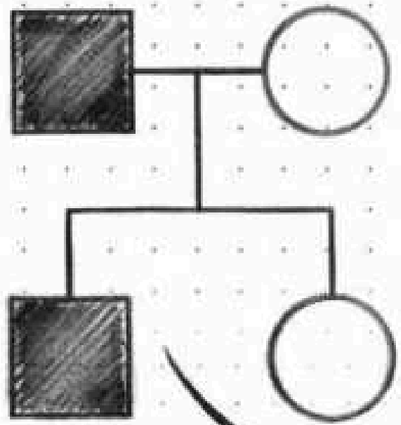
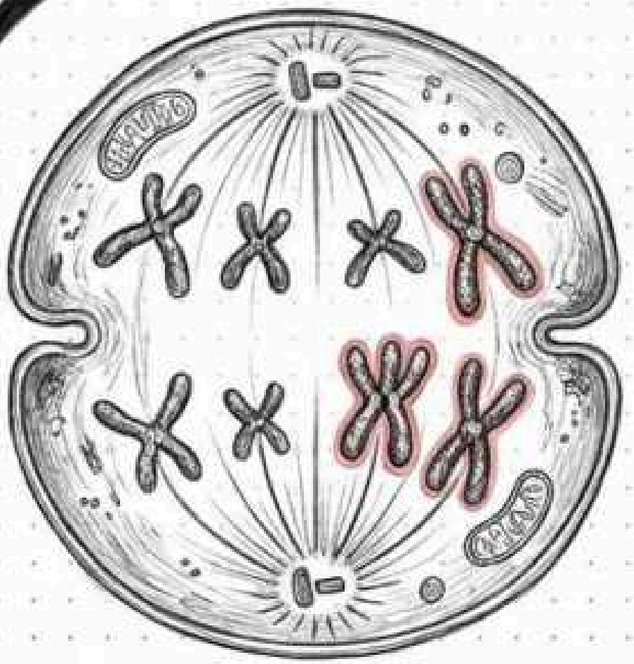
¶4.1

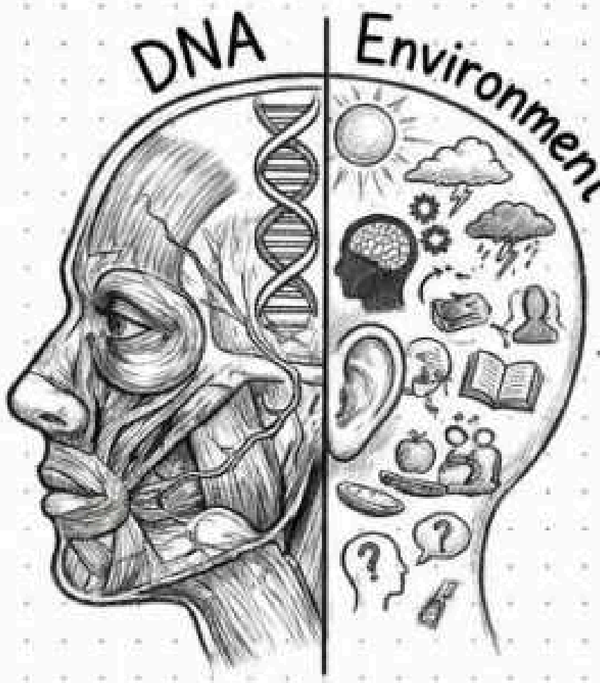


Inheritance Patterns

Three Core Questions of Human Heredity

Chromosome Errors





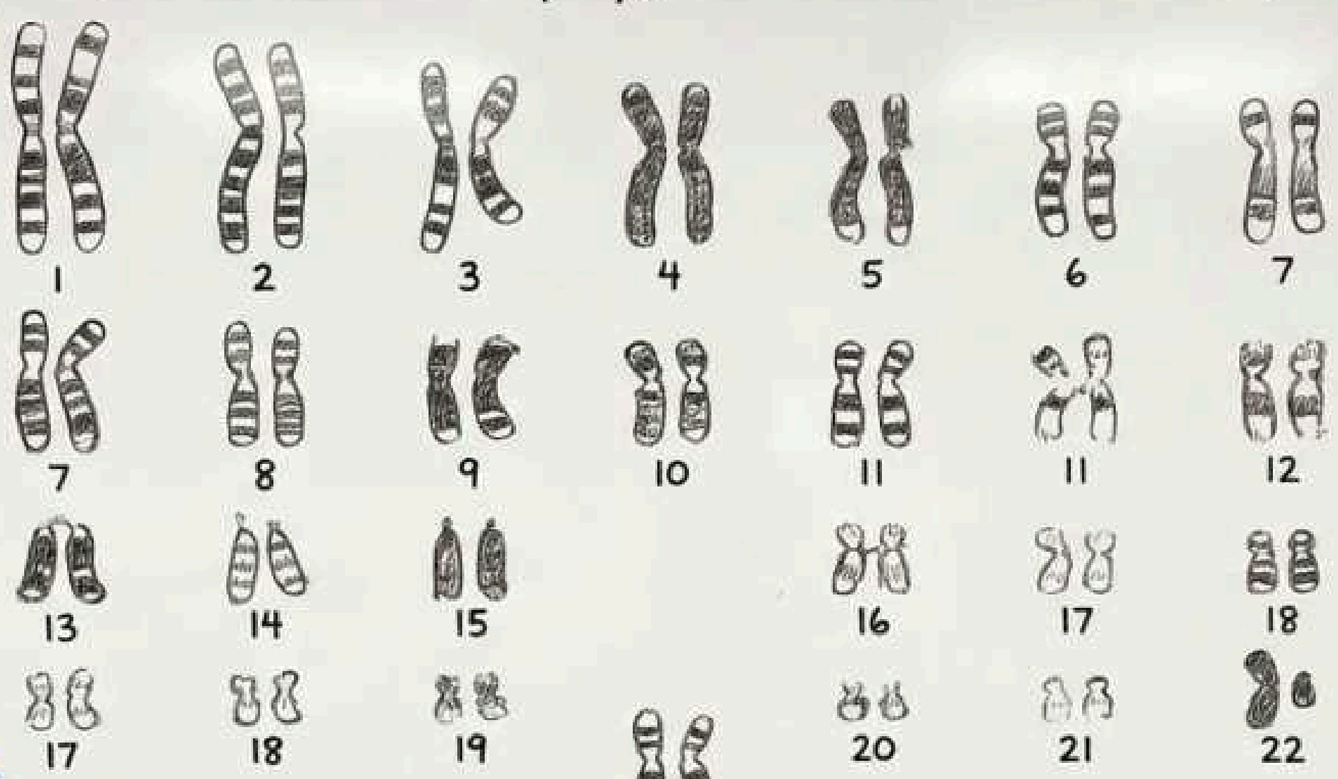
Nature vs. Nurture

Warning: Includes the history of eugenics—essential knowledge for genetics students.

The Human Karyotype and the SRY Switch

¶ 4.2

22 Pairs:
Autosomes
(Same in Males & Females)



XX
(Female)

XY
(Male)

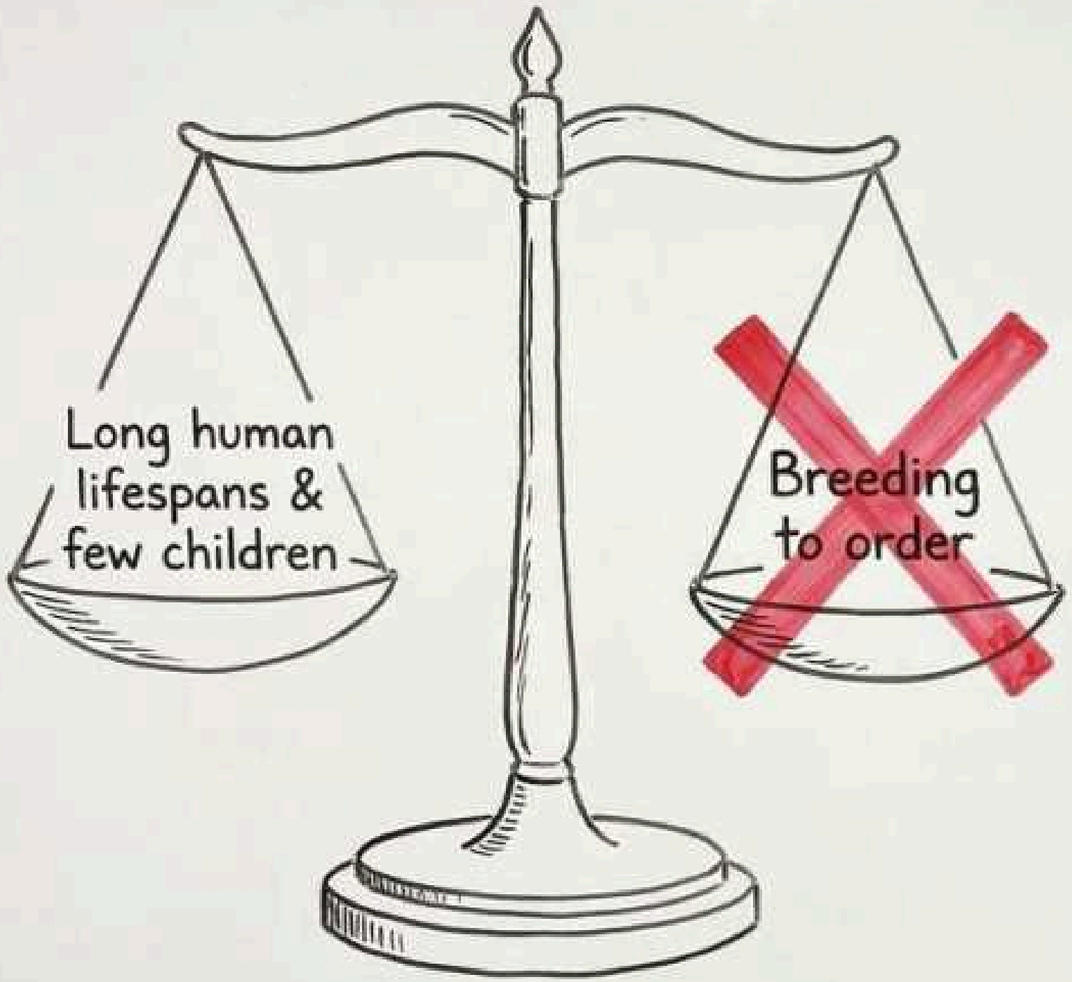
SRY
Gene

ON
ON

Normally switches on male development.

Reading the Family Tree

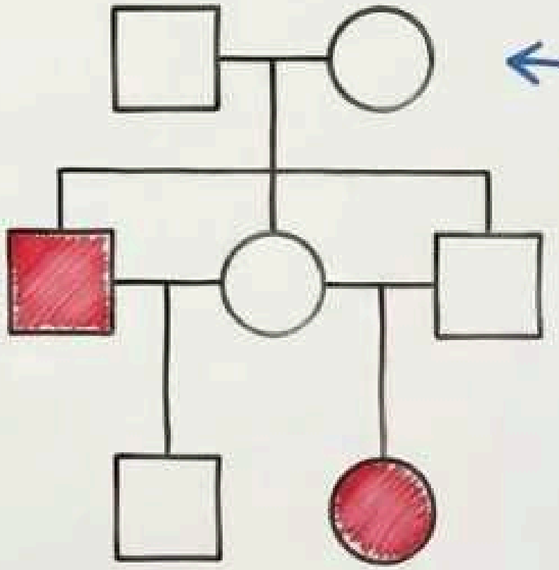
¶ 4.3



Male

Female

Affected Person



Pedigree shape reveals inheritance patterns.

Autosomal Dominant Inheritance

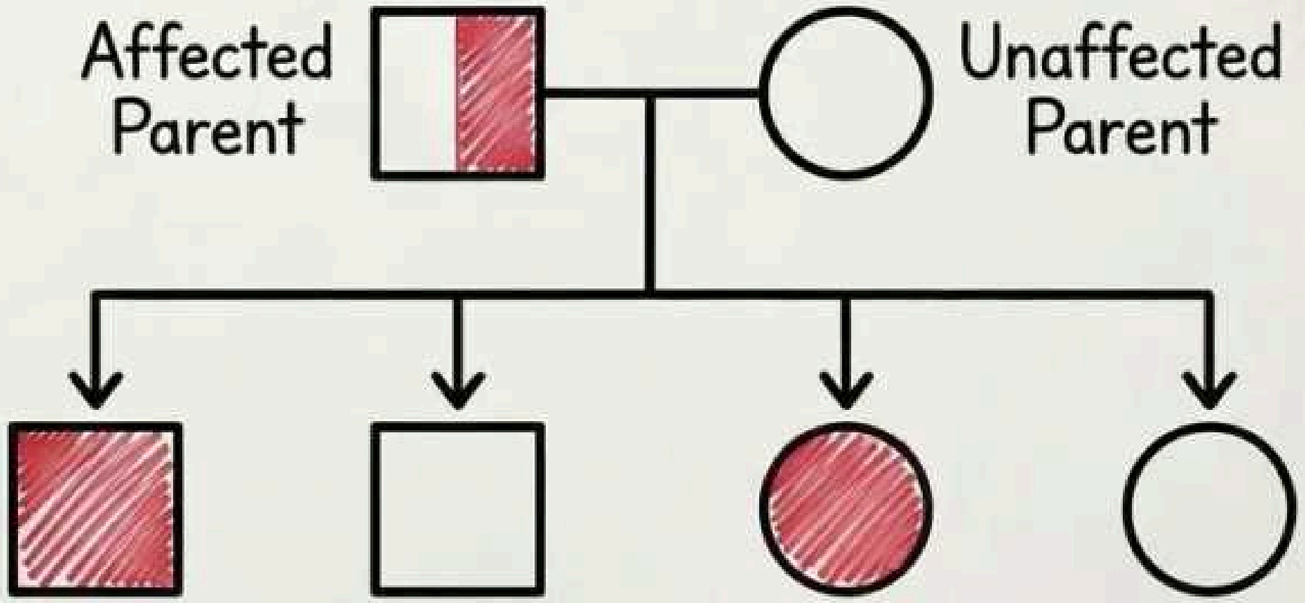
¶ 4.4

$$P(\text{Affected}) = \frac{1}{2}$$

1 Copy = Affected

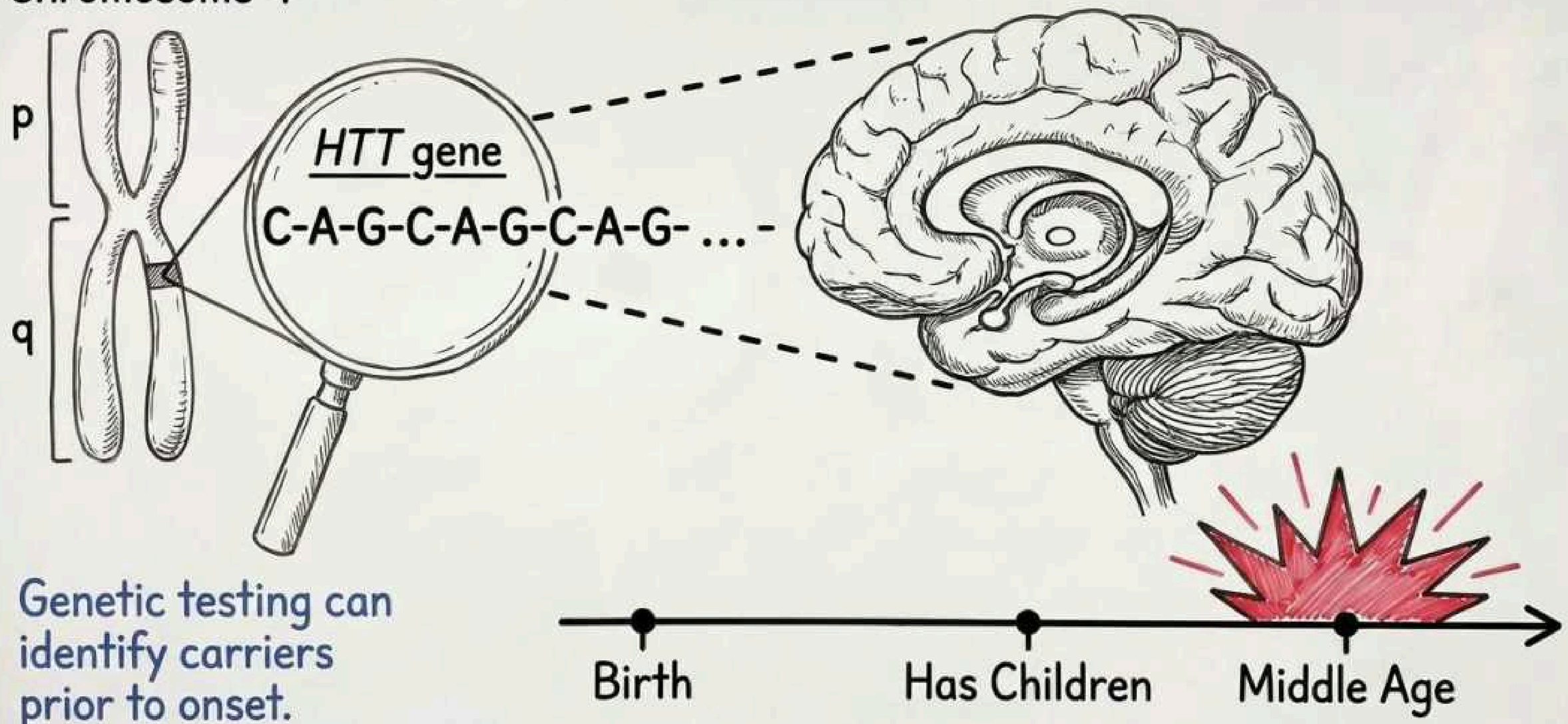
Appears in every generation

Males & Females equally affected



Huntington's Disease: A Dominant Mutation

¶ 4.5



Genetic testing can identify carriers prior to onset.

Autosomal Recessive Inheritance

¶ 4.6



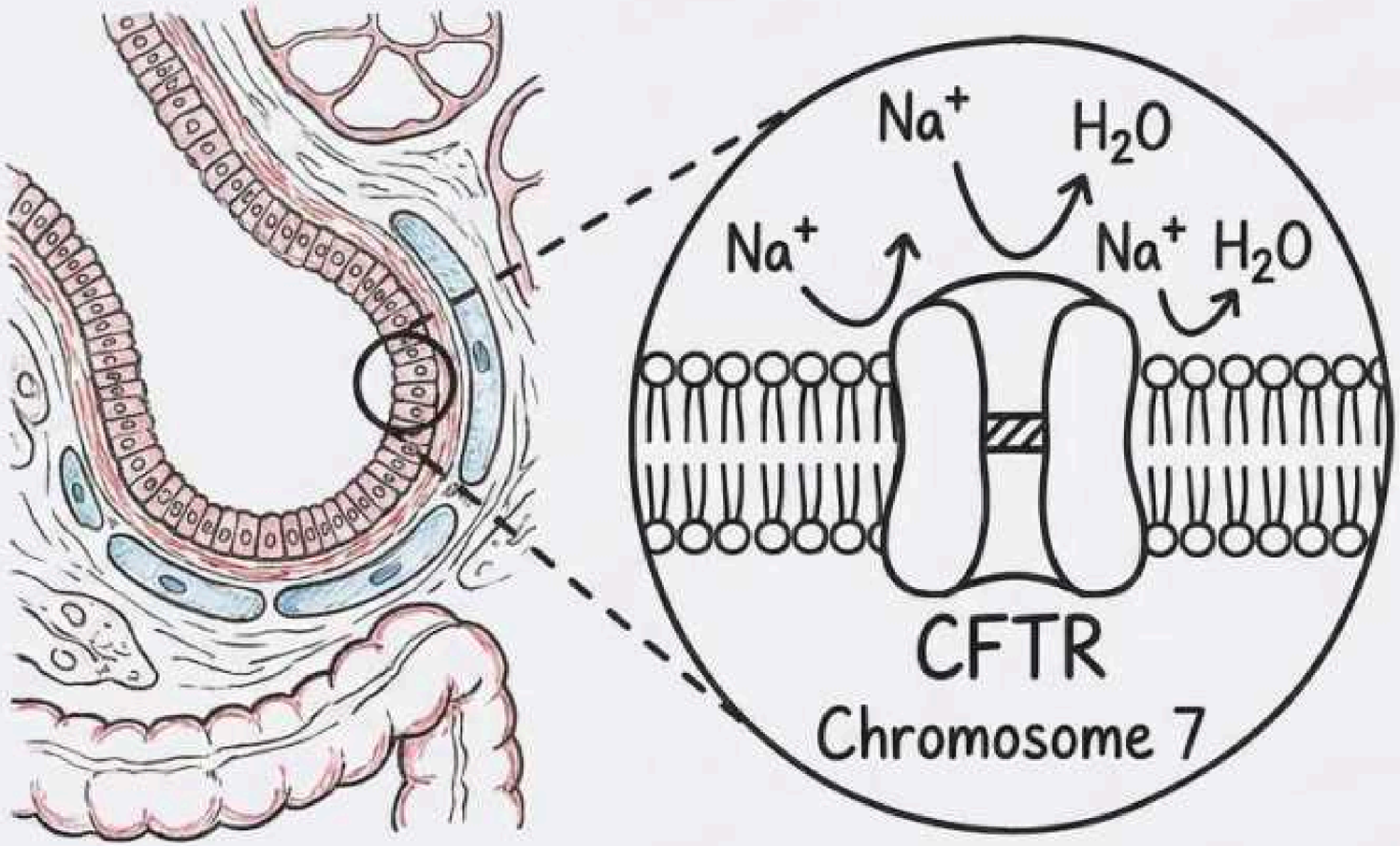
Unaffected Carriers

	Unaffected Carriers	
Unaffected Carriers	¼ Affected	½ Carrier
	½ Carrier	¼ Unaffected

Requires TWO altered copies. Parents are usually unaffected carriers.

Cystic Fibrosis: Cellular Transport Block

¶ 4.7



European Ancestry Populations
Carrier Rate = $\frac{1}{25}$
Affected Newborn Rate = $\frac{1}{3000}$

Treatments have greatly improved the outlook in recent years.

PKU: Where Genes and Environment Meet

¶ 4.8

Phenylalanine

Missing Enzyme → Buildup → Brain Damage

Special Diet → Bypass → Healthy Brain



Newborn screening (since the 1960s) prevents most harm through dietary intervention.

X-Linked Recessive Inheritance

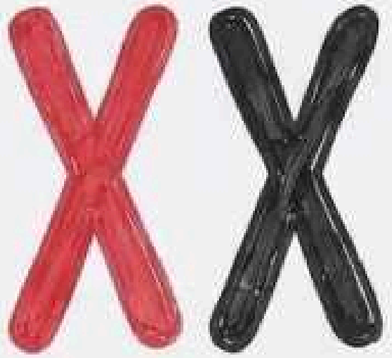
¶ 4.9

Male XY



Nothing to mask the altered allele = Affected.

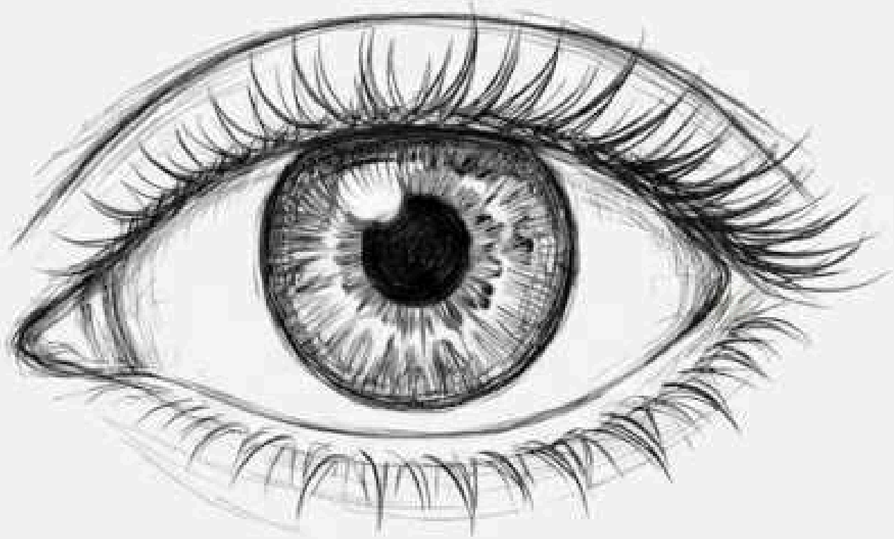
Female XX



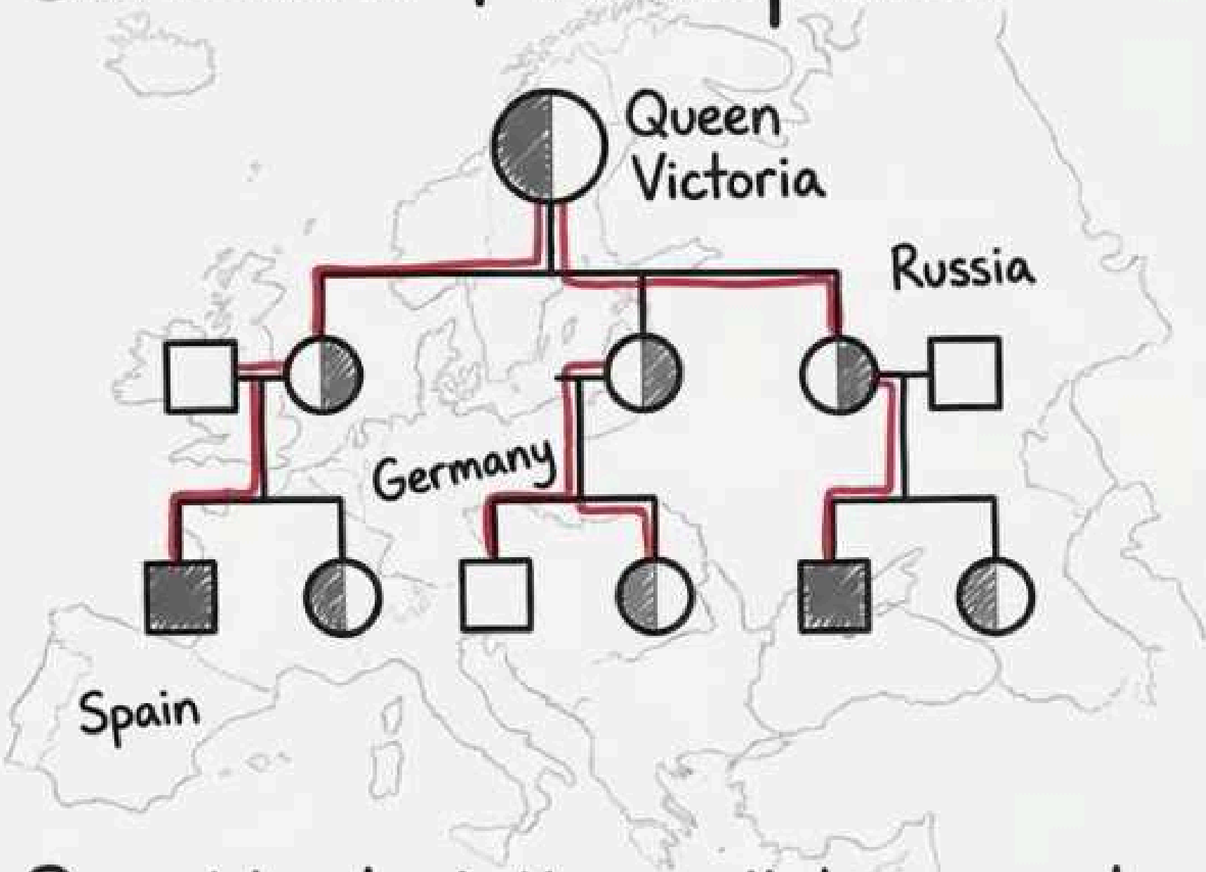
100% of Daughters → All become Carriers

100% of Sons → None are affected

X-Linked Examples: Color Blindness & Hemophilia



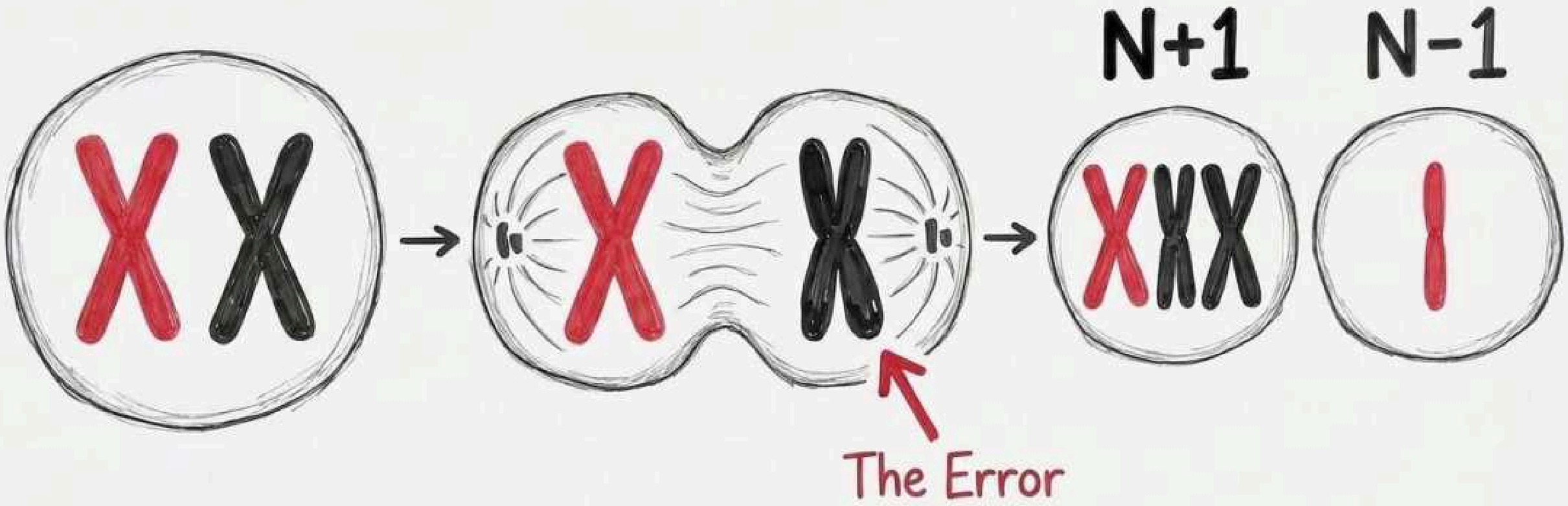
Northern European descent:
8% Men vs 0.5% Women



Poor blood clotting allele spread spread through daughters into European royal families.

Nondisjunction: Miscounted Chromosomes

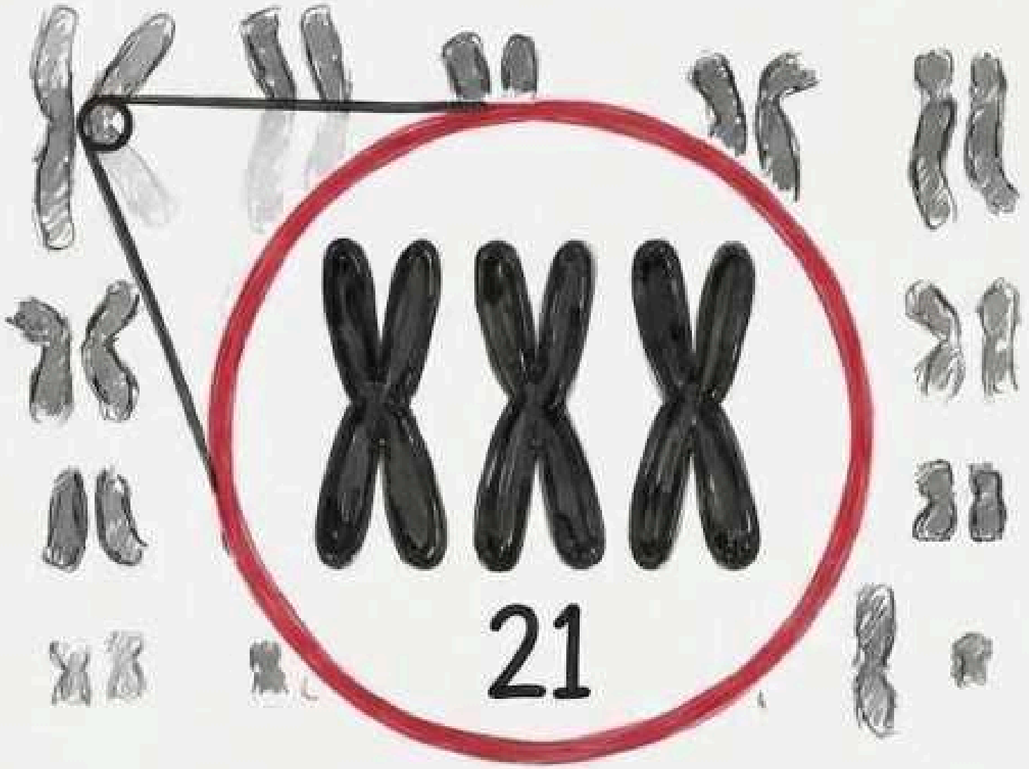
¶ 4.11



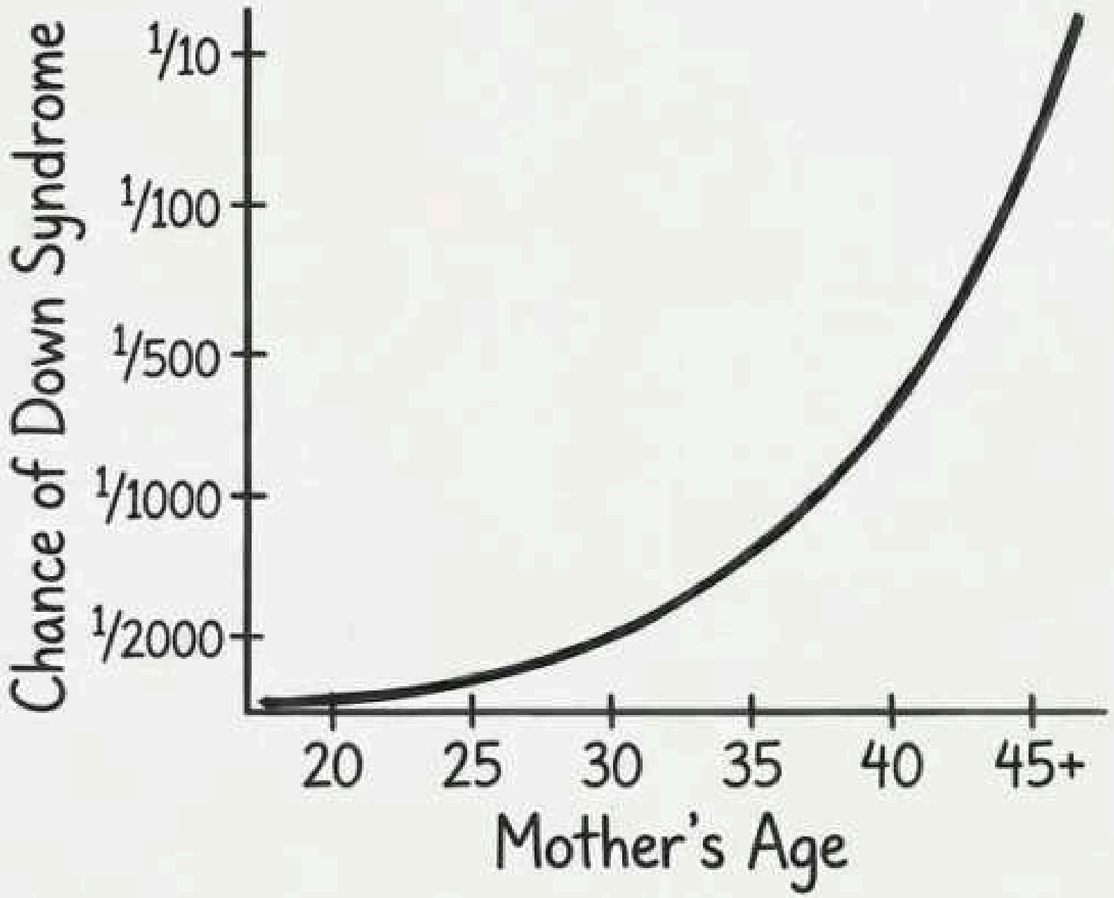
Most embryos with this error do not survive, but some exceptions exist.

Down Syndrome (Trisomy 21)

¶ 4.12



Extra chromosome identified by Jérôme Lejeune in 1959.

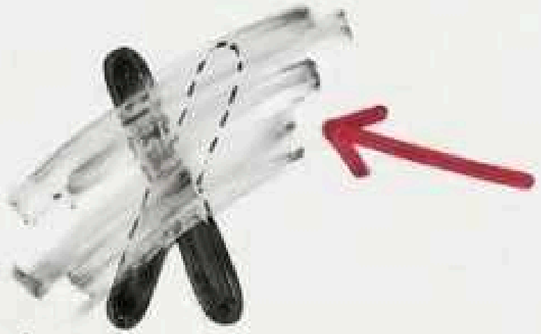


Modern life expectancy commonly extends into the sixties and beyond.

Sex Chromosome Changes

¶ 4.13

Turner Syndrome	Klinefelter Syndrome
XO	XXY
X	XXY

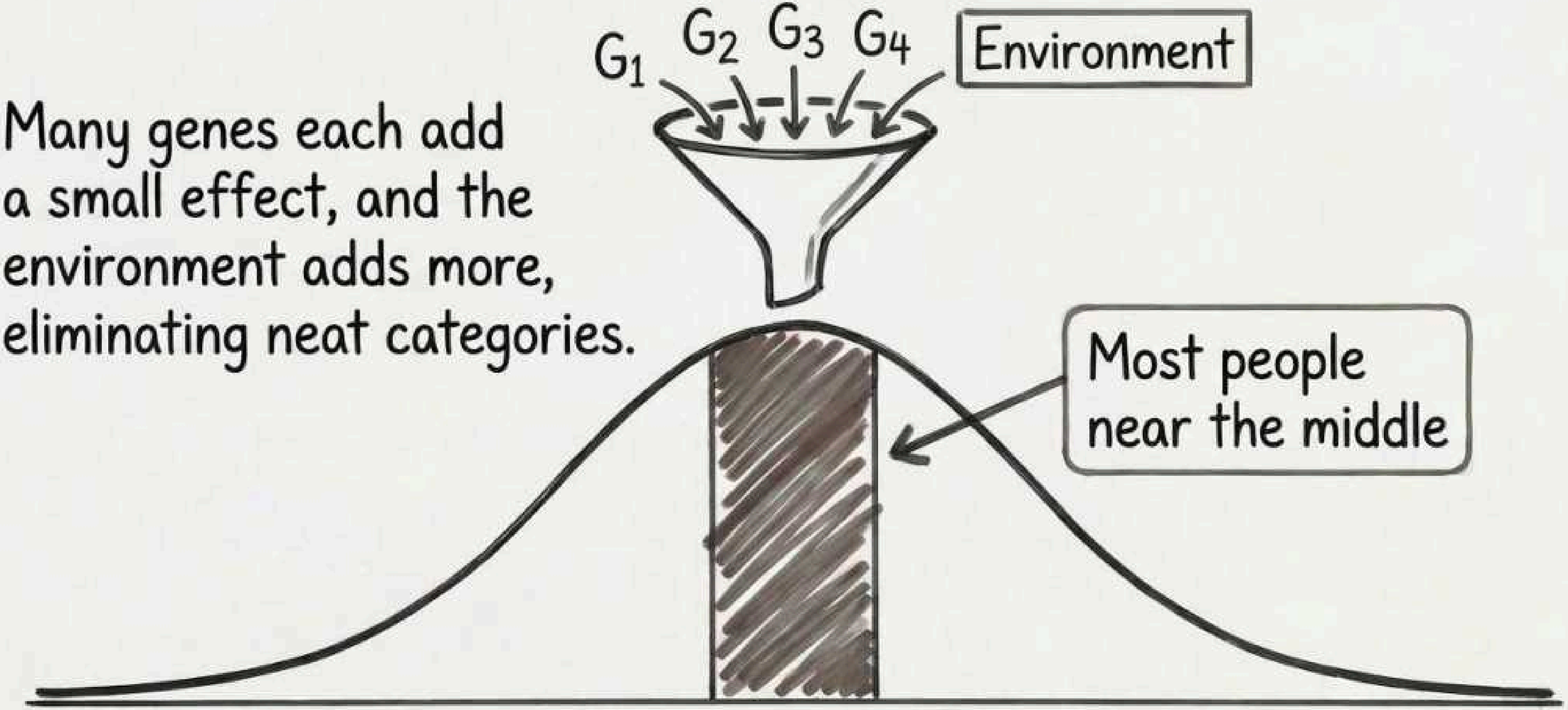


Relatively mild compared to autosomal changes because extra X chromosomes are largely switched off.

Polygenic Traits: Beyond Simple Rules

¶ 4.14

Many genes each add a small effect, and the environment adds more, eliminating neat categories.

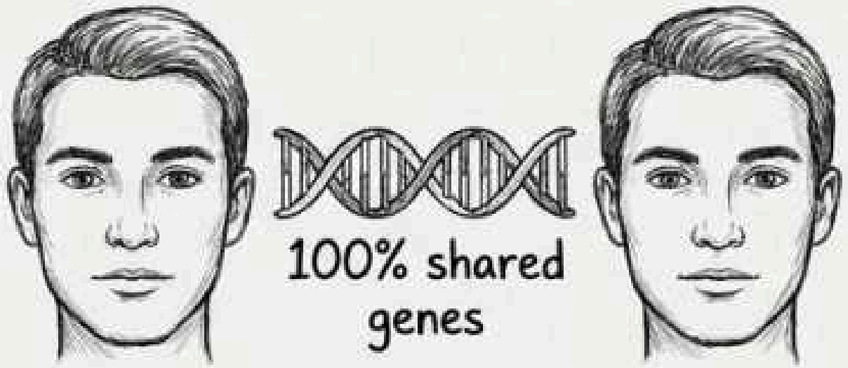


Height, Skin Color, Risk of Heart Disease

Heritability and Twin Studies

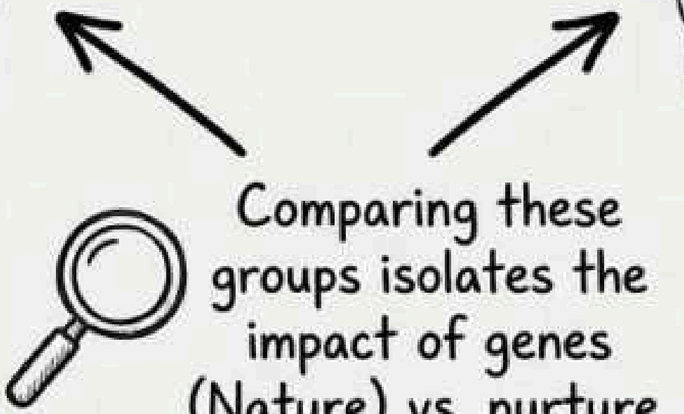
$$0 \leq H^2 \leq 1$$

(Estimates variation due to genetic differences within a population)

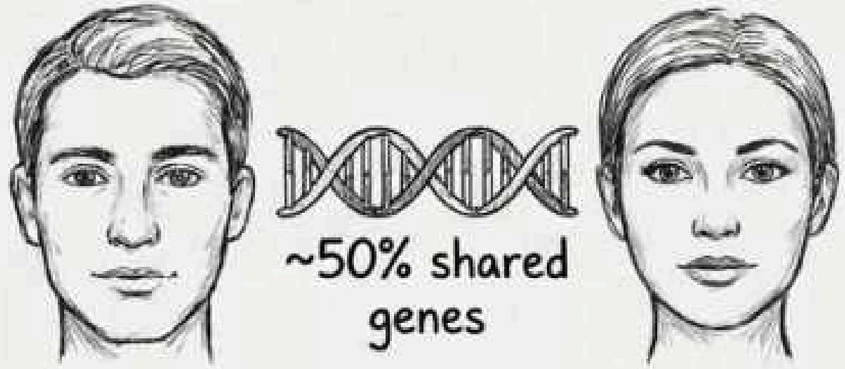


100% shared genes

Identical Twins



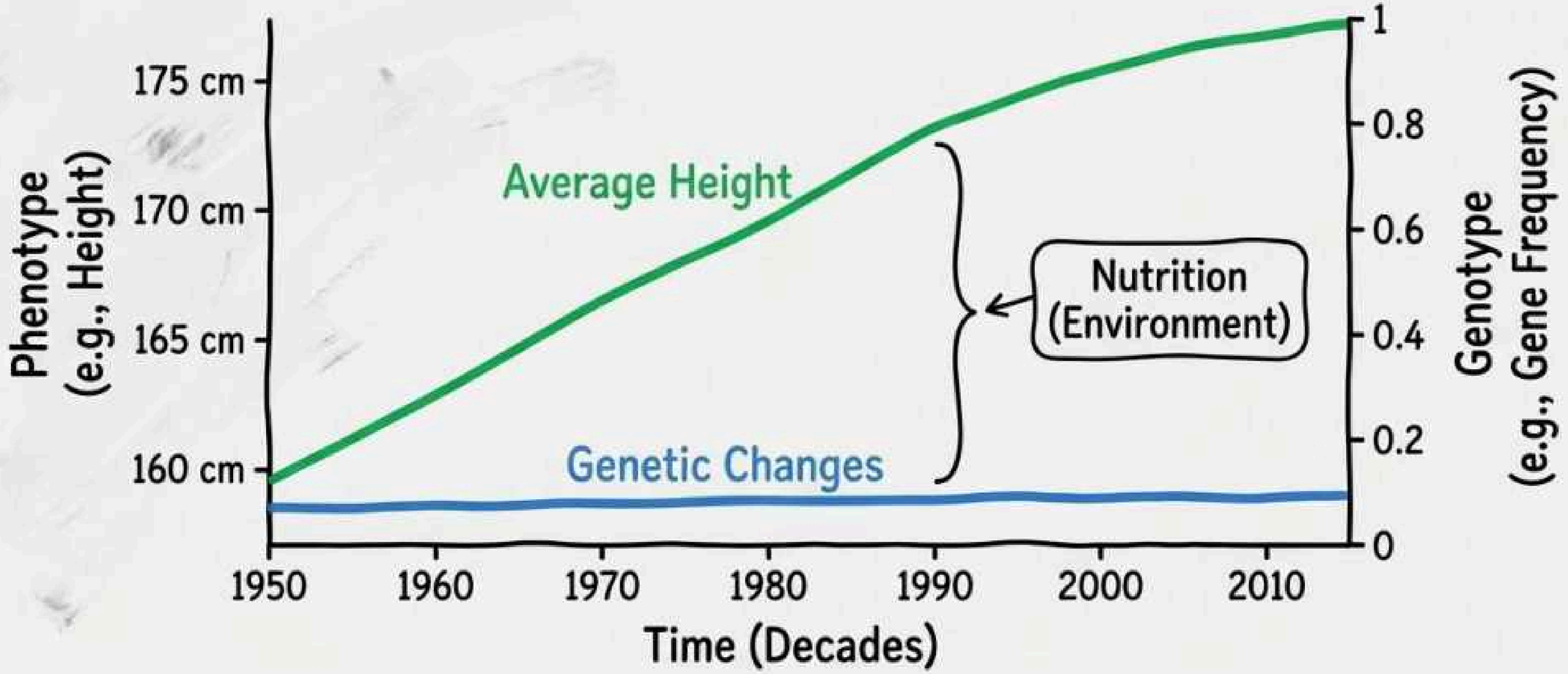
Comparing these groups isolates the impact of genes (Nature) vs. nurture.



~50% shared genes

Fraternal Twins

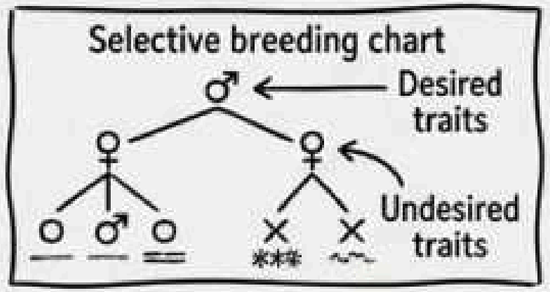
The Limits of Heritability



1. High heritability (~80% for well-fed adult height) describes differences within a group at one time.
2. It does not mean a single person's trait is 80% destined by genes. Genes and environment are partners.

The History of Eugenics

1883
Francis Galton (Darwin's cousin) coins "Eugenics"



Early 1900s
Spread to America; multiple states pass sterilization laws.

1927
Supreme Court upholds sterilization in Buck v. Bell



1930s–1940s
Nazi Germany takes eugenics to murderous extremes

1930s–1940s
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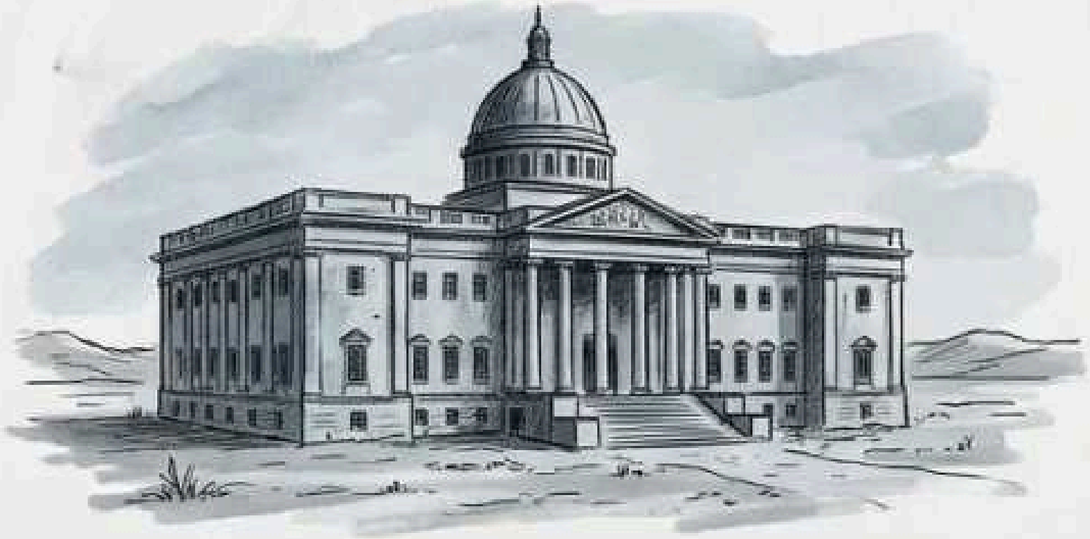


Ethics & Modern Genetic Counseling

¶ 4.18

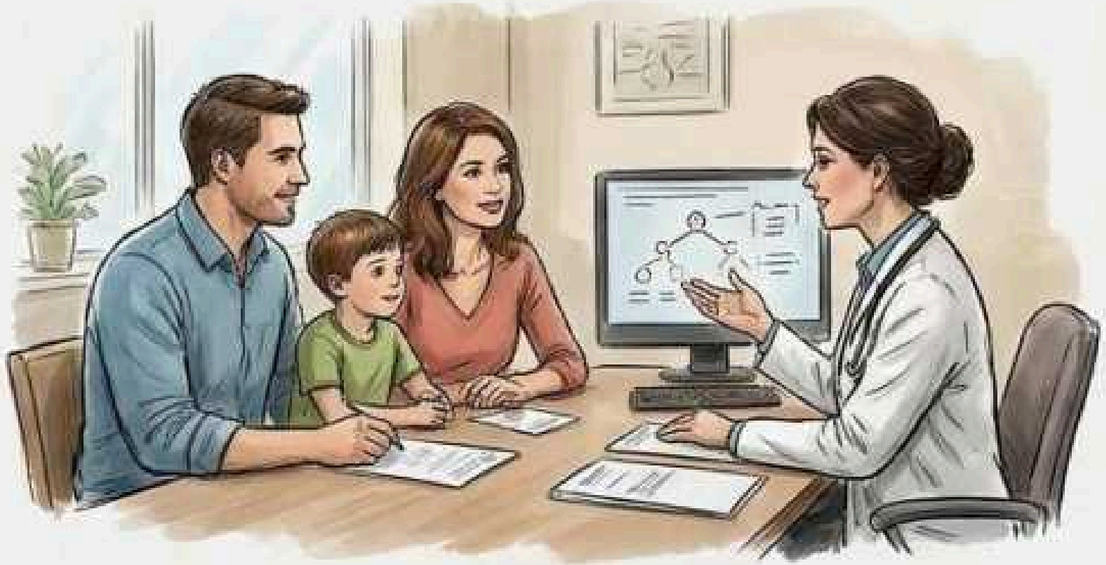
Then (Eugenics)

- Bad science + prejudice
- Targets poorly defined (e.g., “feeblemindedness”)
- Ignored environment
- State control over reproduction



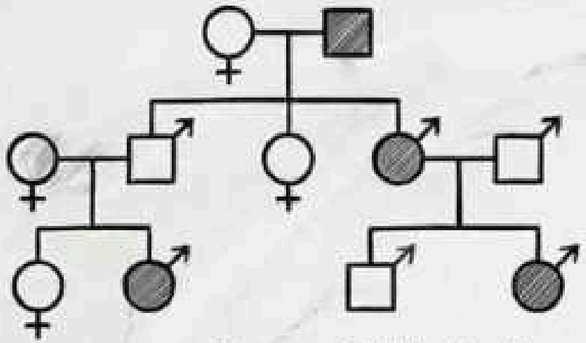
Now (Genetic Counseling)

- Accurate science + environment awareness

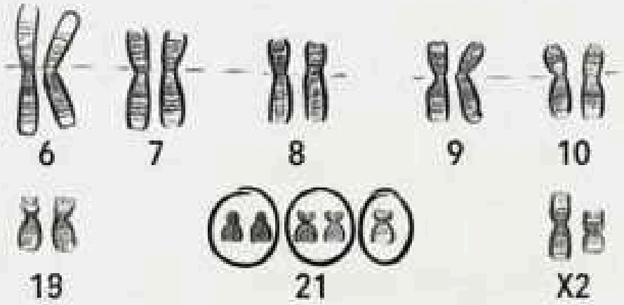


Counselor Explains Risks & Options → Decisions Made By Families (Not the State)

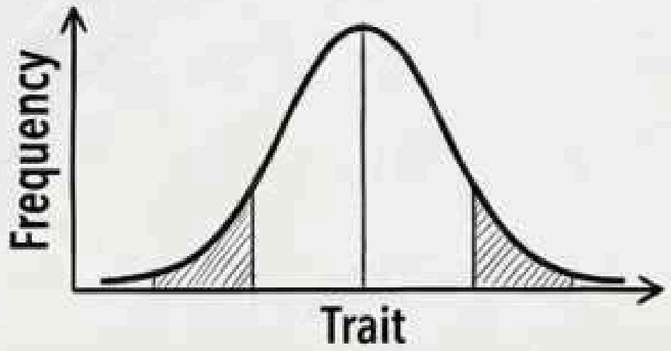
Lecture Synthesis



Dominant, recessive, & X-linked patterns (Huntington's, CF, Hemophilia).



Miscounted chromosomes (Down syndrome).



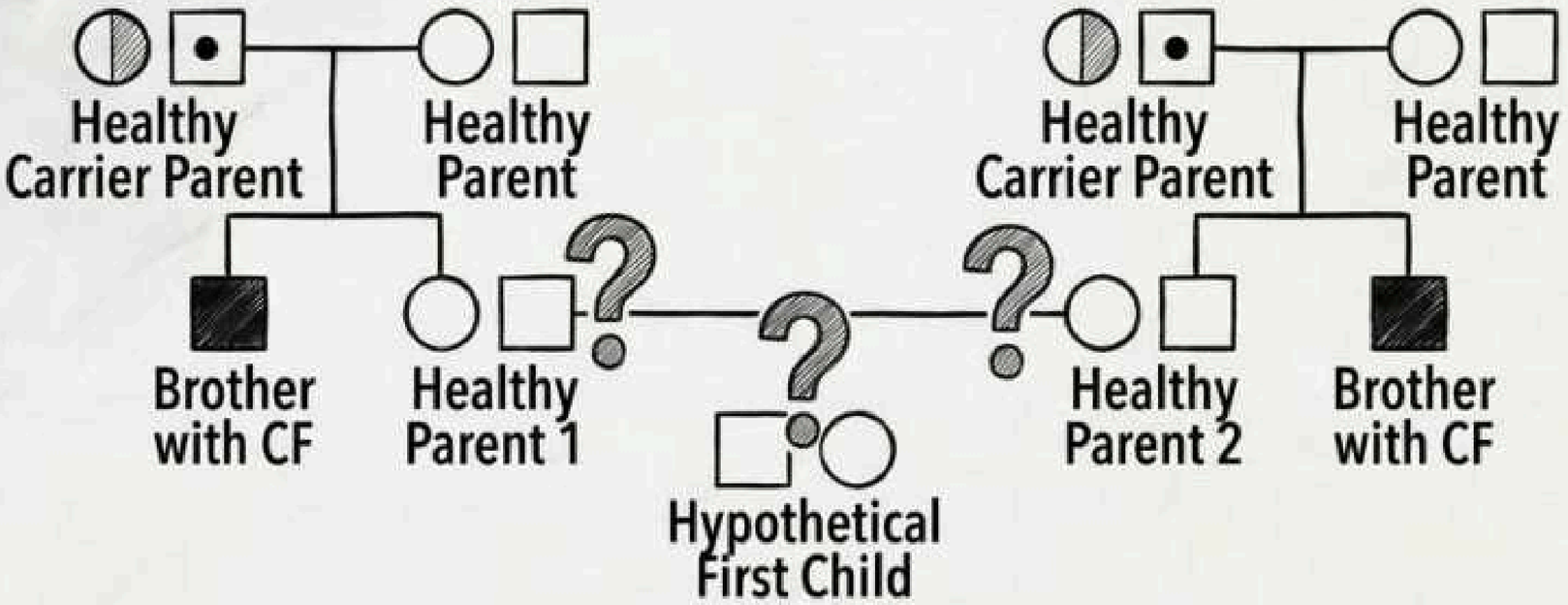
Most traits are polygenic; Heritability measures variation, not destiny.



Eugenics history warns us to use genetic knowledge carefully.

The Capstone Challenge

Two healthy parents each have a brother with cystic fibrosis, and each has a healthy parent who is a carrier.



- ☐ What are the odds the first child is affected?
- ☐ Why does a pedigree help work this out?
- ☐ What would a good genetic counselor do?