

Human (kompleks) Genetikk og epigenetikk

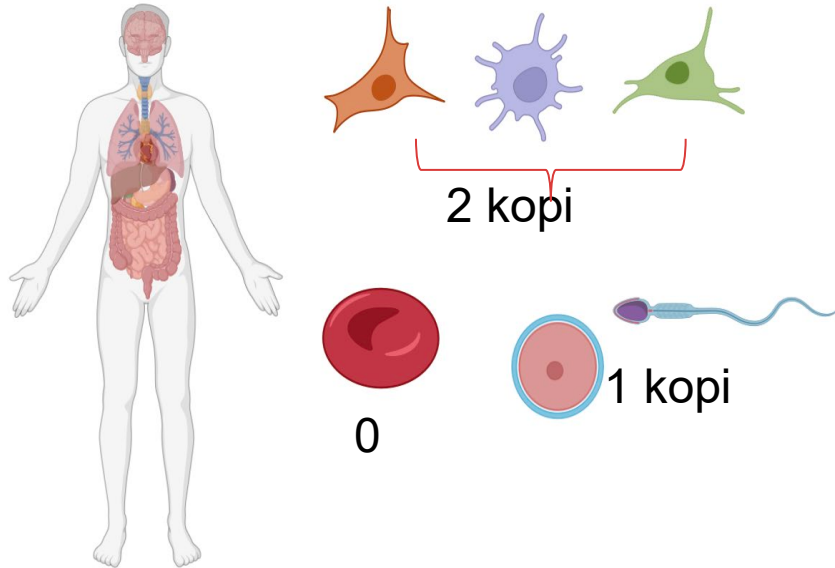
Stéphanie Le Hellard, Professor in Human Genetics
Institutt Klinisk Medisin 2
Medisin Fakultet, UiB



Lærernes
dag 2026



The Human Genome

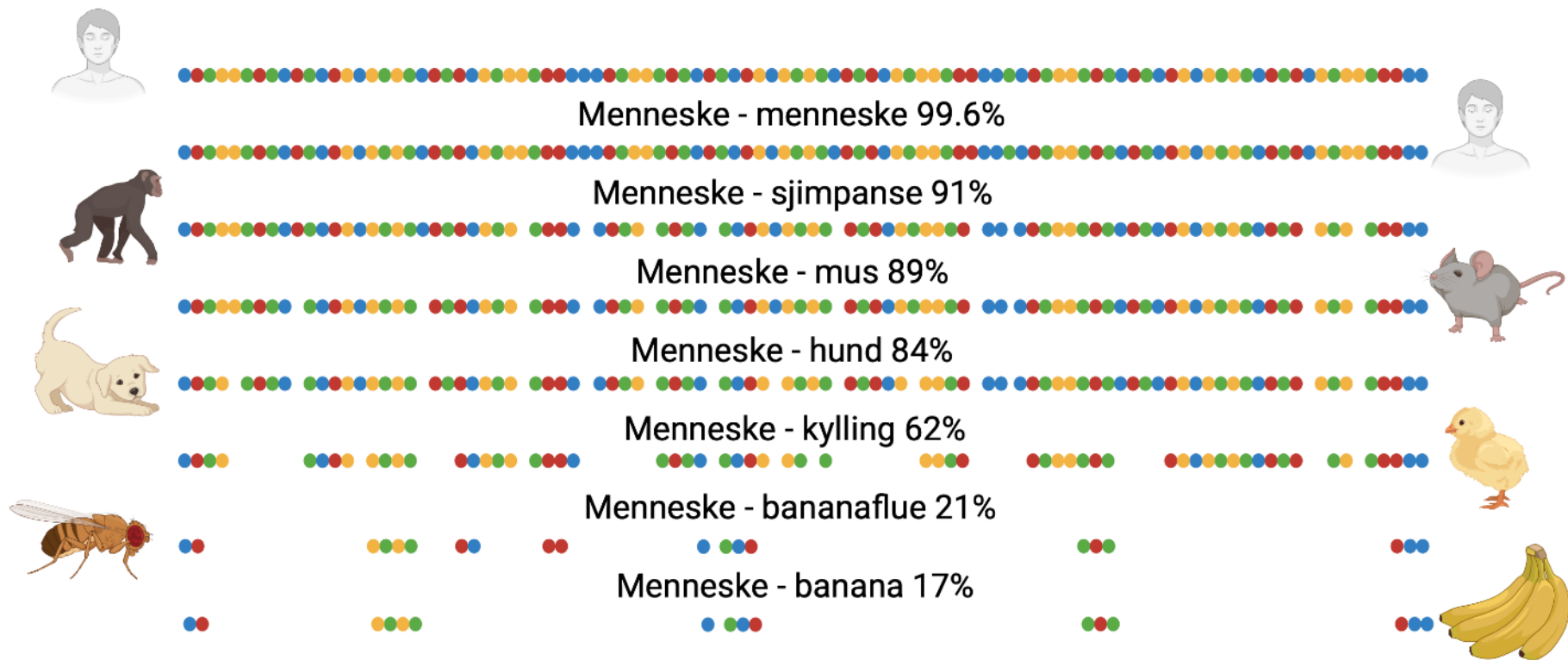


1 kopi av genomet er 23 DNA
molekyler, dvs 23 kromosomer
Ca. 3,000,000,000 nukleotider total.

75-90% brukes
1-2 % gir proteiner
Resten regulerer genuttrykk + “noise”

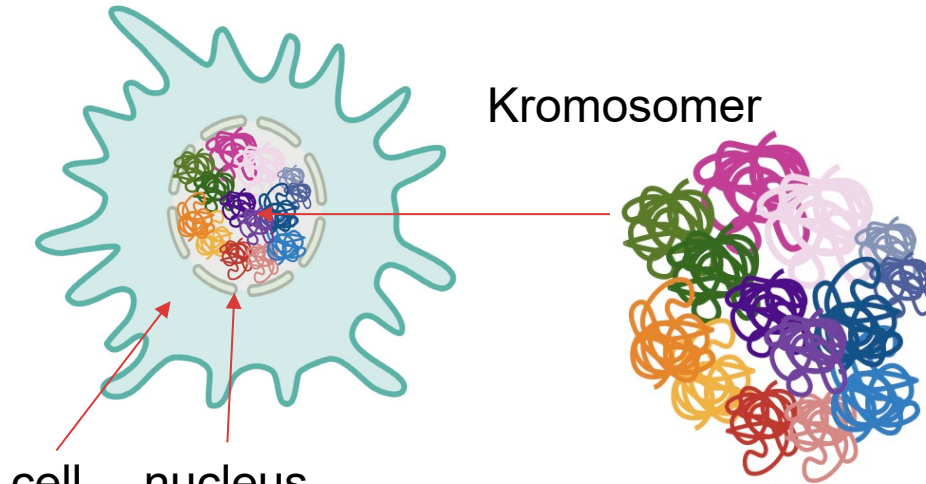
19,000 – 20,000 gener

Komparativ genomikk: hvor like er to (eller flere) genomer? Det forteller oss hvor langt fra hverandre to arter står i evolusjonen.



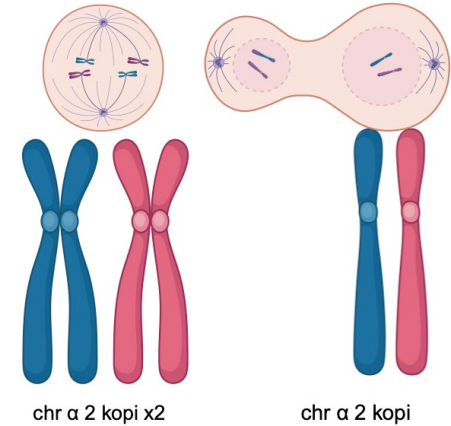
!!! Dette er for de 1-2% protein kodende !!!

Kromosomer



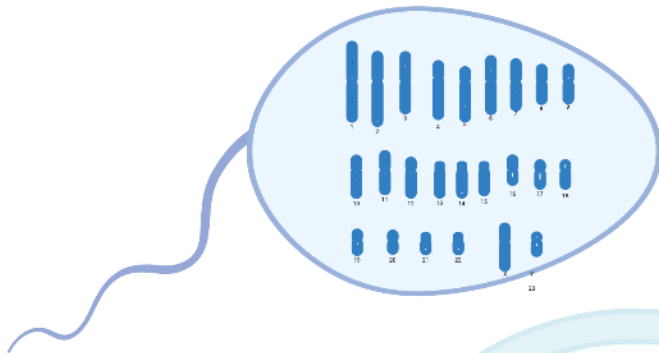
Kromosomer

Kromosomer i aktive celler er organisert for genuttrykk og befinner seg i såkalte "chromosome territories"

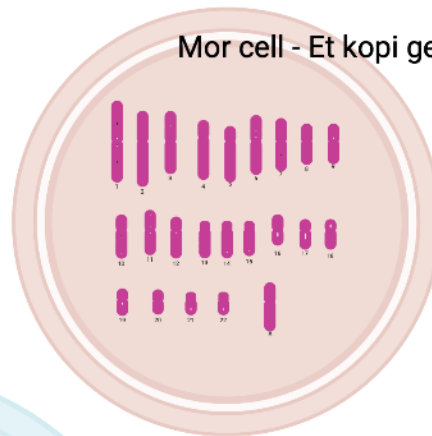


Når cellene deler seg, blir kromosomene pakket sammen, og hvert kromosom blir kopiert slik at dattercellene får én kopi hver.
Disse kromosomer er veldig kompakt og derfor inactive.

Far cell - Et kopi genom



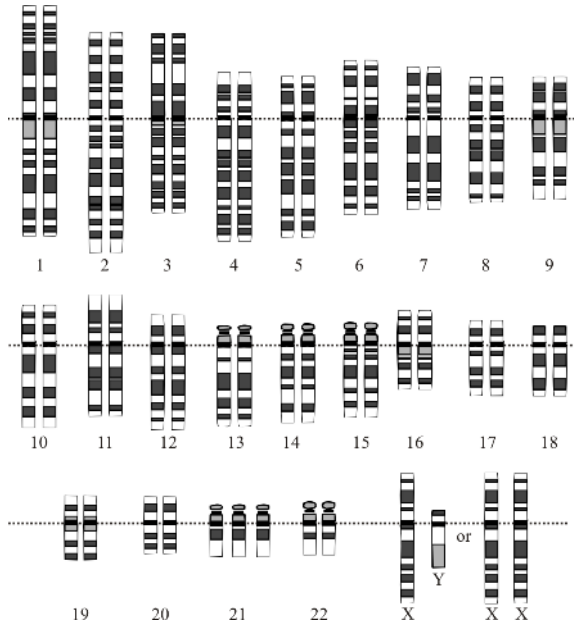
Mor cell - Et kopi genom



Barn cell - To kopi genom

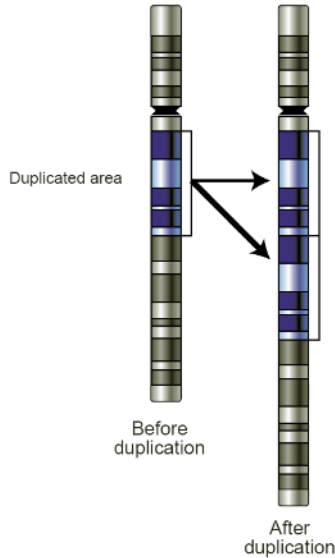


Genetiske variasjoner -størrelse



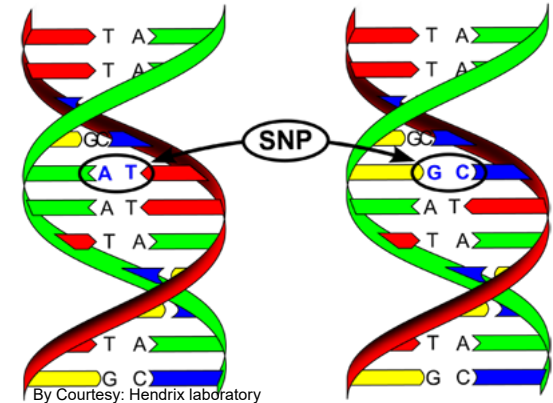
By Courtesy: National Human Genome Research Institute - Human Genome Project, Public Domain,
<https://commons.wikimedia.org/w/index.php?curid=889304>

Kromosom tall
Sjelden veldig få “viable”



By Courtesy: National Human Genome Research Institute Public Domain,
<https://commons.wikimedia.org/w/index.php?curid=552474>

Kopi Tall Variasjon
Fra 1 nukleotide til
10,000s
Alle har flere



Single nucleotide
polymorphism
>30 millioner i genom.
2 variasjoner kalles
alleler



Menneske - menneske 99.6%



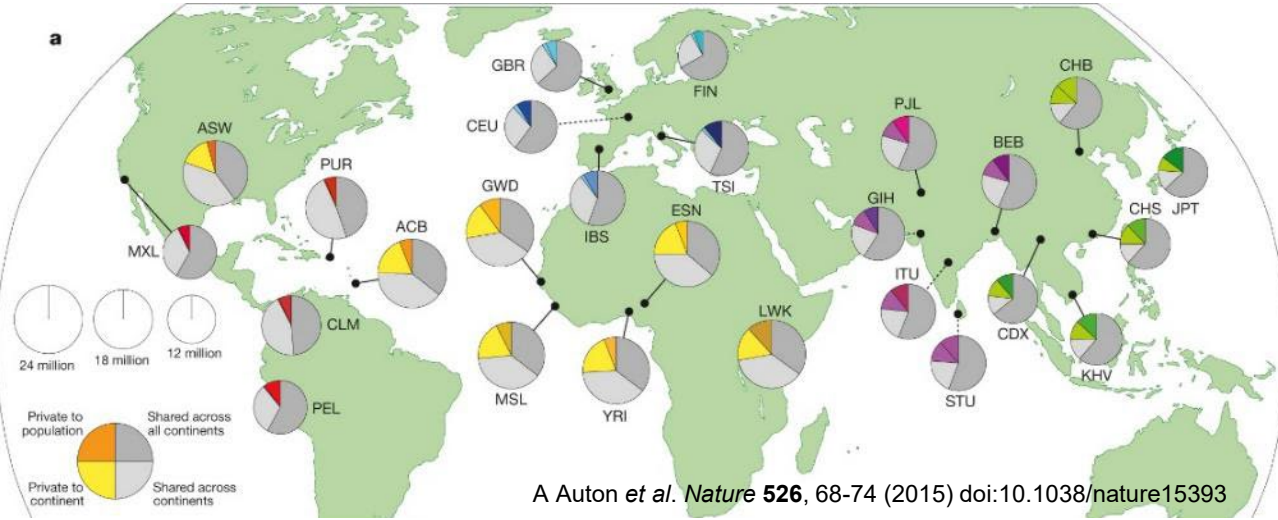
Genetiske variasjoner mellom to mennesker utgjør omtrent 0,4 %.

Rundt 0,4 % av genomet varierer, noe som tilsvarer > 12 millioner genetiske variasjoner ($0,4 \times 3\,000\,000\,000 = 12$ millioner).

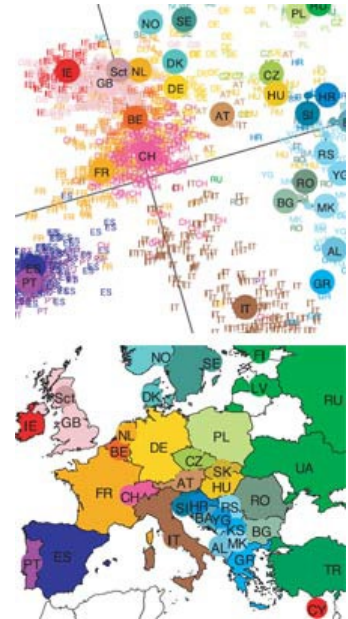
Genetiske variasjoner kjennetegnes av:

- Størrelse
- Frekvens
- Effekt

> $\frac{3}{4}$ variasjoner er felles mellom alle populasjoner



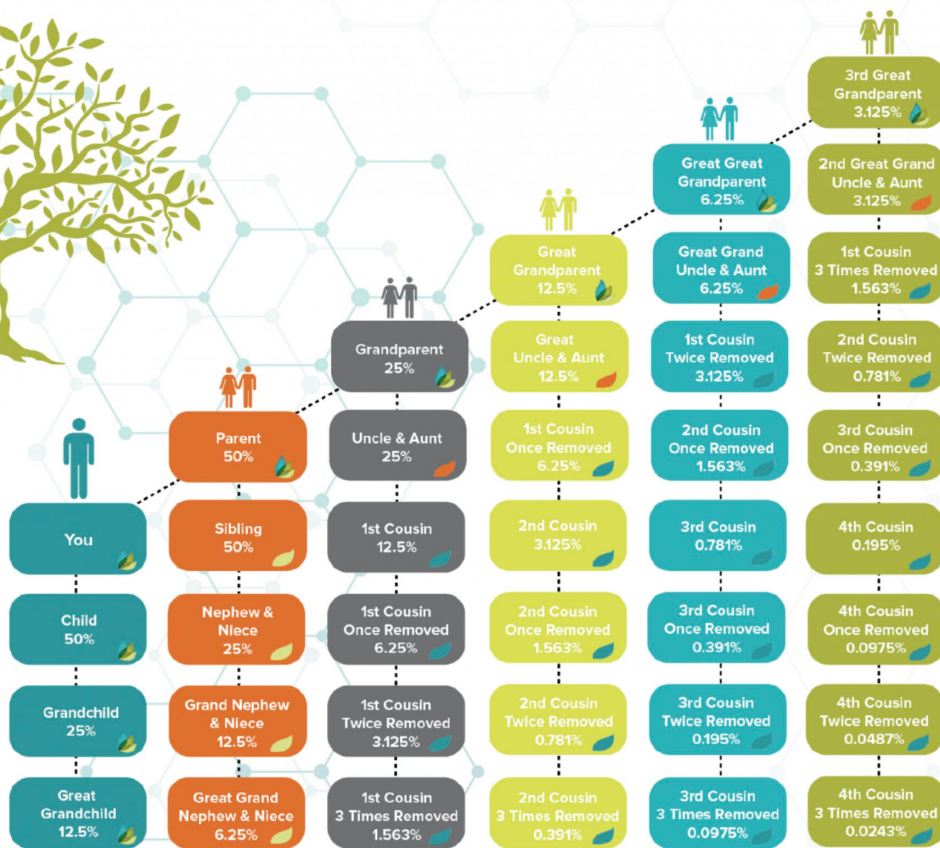
Andre variasjoner kan informere veldig presis om hvor folk er fra



DNA & Relationship Quick Reference Chart



- Direct Ancestors or Descendants
- Siblings of Direct Ancestors
- Cousins
- Siblings & Their Descendants
- Percentage of Shared DNA



- ✓ Full-service genealogical research firm
- ✓ DNA analysis and genetic genealogy
- ✓ Family narratives and books
- ✓ Highest client ratings in the world
- ✓ Global network of archives and researchers

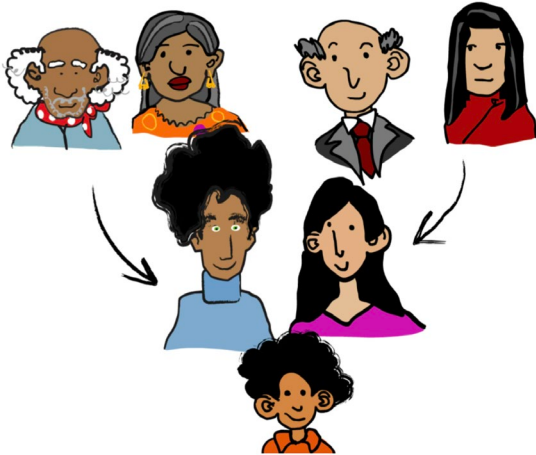
Let us help you discover your story!

*Percentages provided are approximate, and may vary. Some levels of relationship may not share detectable amounts of DNA.

We Do the Research. You Enjoy the Discoveries. | www.legacytree.com

Genetiske variasjoner –effekt på trekk

Mendelsk trekk



Svært få trekk skyldes én enkelt genetisk variasjon.

1 genetisk variant kan føre til 1 trekk/sykdom.

Dette gjelder sjeldne sykdommer, eks. cystisk fibrose og Huntingtons sykdom.

Kompleks trekk

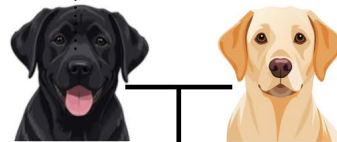
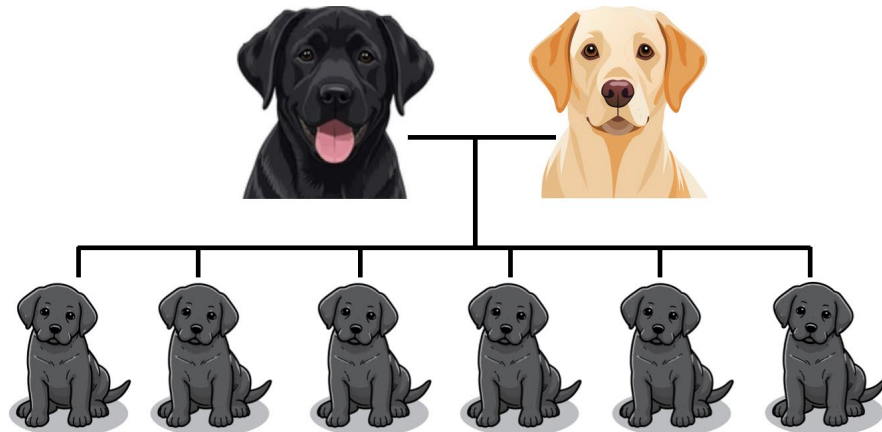


The Trapp family is pictured in a provided image. Photo Credit: Kevin Scott Ramos/Guinness World Records

Nesten alle trekk der barn ligner foreldrene sine, er komplekse trekk.

De skyldes mange genetiske variasjoner, hver med liten effekt, som virker additivt.

De fleste vanlig genetiske sykdommer: psykiske lidelser, metabolske sykdom,...

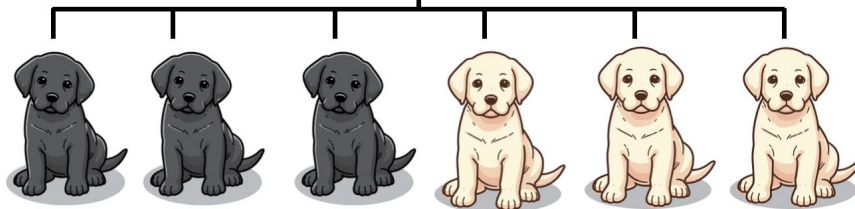


Which colour is dominant?

Black or beige?

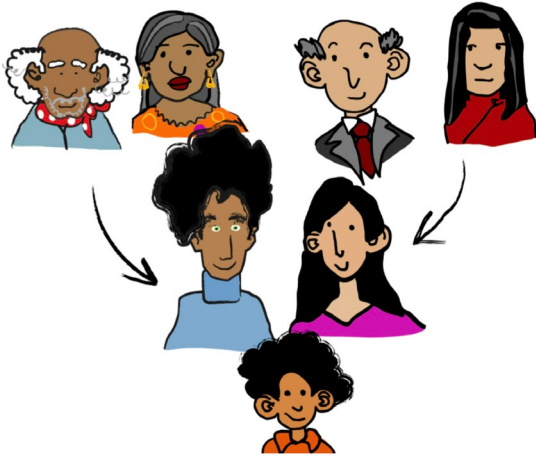
Black -> B

Beige -> b



Genetiske variasjoner –effekt på trekk

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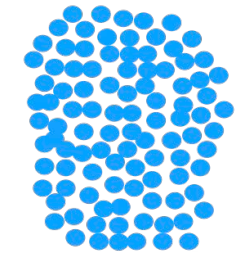
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Kompleks trekk : mange varianter, lite effekt hver, additiv

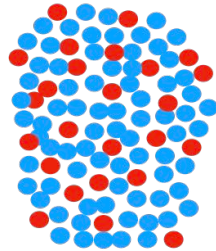
Polygenetisk trekk: Mange genetiske variasjoner påvirker et trekk, og hver variasjon har to alleler som enten øker ● eller minsker ● egenskapen.

Vi tar høyde som eksempel, og 100 variasjoner som påvirker høyde

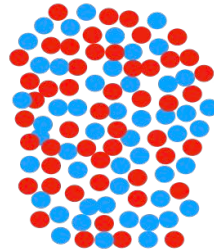
● gir 0.3 cm høyere enn ●



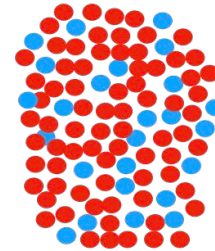
0 % ●
100 % ●
 $1.75 - 50 \cdot 0.3 =$
1.60m



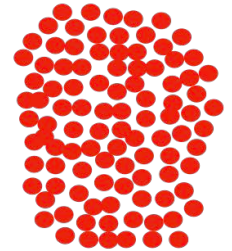
25 % ●
75 % ●
 $1.75 - 25 \cdot 0.3 =$
1.675 m



50 % ●
50 % ●
Gjennomsnitt høyde
1.75

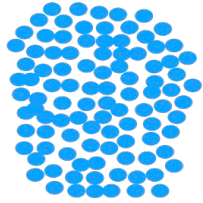


75 % ●
25 % ●
 $1.75 + 25 \cdot 0.3 =$
1.825

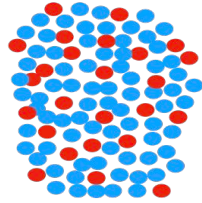


100 % ●
0 % ●
 $1.75 + 50 \cdot 0.2 =$
1.90

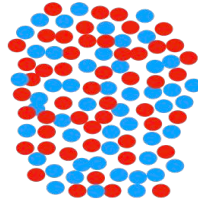
Kompleks trekk : mange varianter, lite effect hver, additiv



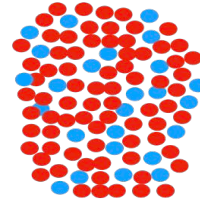
0 % ●
100 % ●
 $1.75 - 50 \cdot 0.3 =$
1.60m



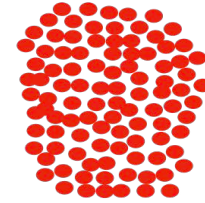
25 % ●
75 % ●
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1.675 m



50 % ●
50 % ●
Gjennomsnitt høyde
1.75



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25 % ●
 $1.75 + 25 \cdot 0.3 =$
1.825



100 % ●
0 % ●
 $1.75 + 50 \cdot 0.2 =$
1.90

Sannsynlighet å ha disse kombinasjoner

<1%

~25 %

~50 %

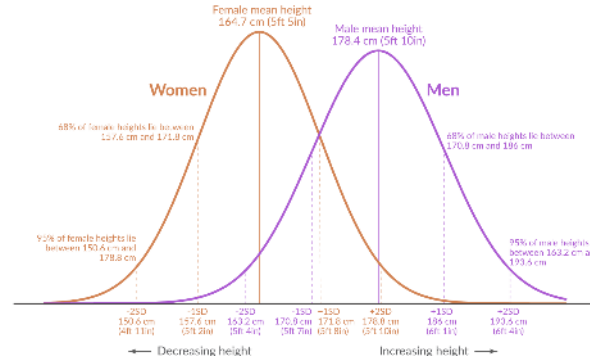
~25 %

<1%

Normalfordeling i befolkning

The distribution of male and female heights

The distribution of adult heights based on large cohort studies across 20 countries in North America, Europe, East Asia, and Australia. Shown is the sample-weighted distribution across all cohorts born between 1960 and 1994 (so reaching the age of 18 between 1998 and 2012).

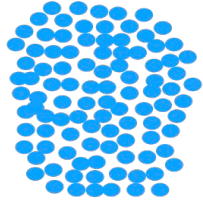


Data source: Adamić et al. (2016). Genetic and environmental influences on adult human height across birth cohorts from 1886 to 1994.

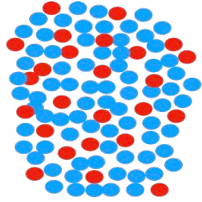
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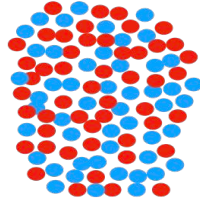
Kompleks trekk : hvorfor barna ligner foreldrene



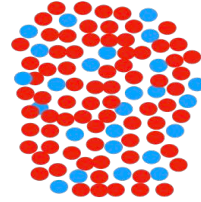
0 % ●
100 % ●
 $1.75 - 50 \cdot 0.3 =$
1.60m



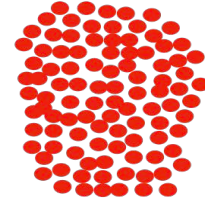
25 % ●
75 % ●
 $1.75 - 25 \cdot 0.3 =$
1.675 m



50 % ●
50 % ●
Gjennomsnitt høyde
1.75



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 $1.75 + 25 \cdot 0.3 =$
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100 % ●
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Foreldre 1

Foreldre 2

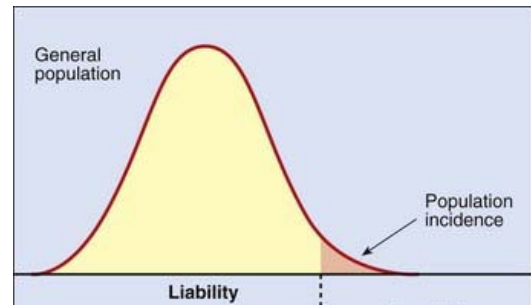
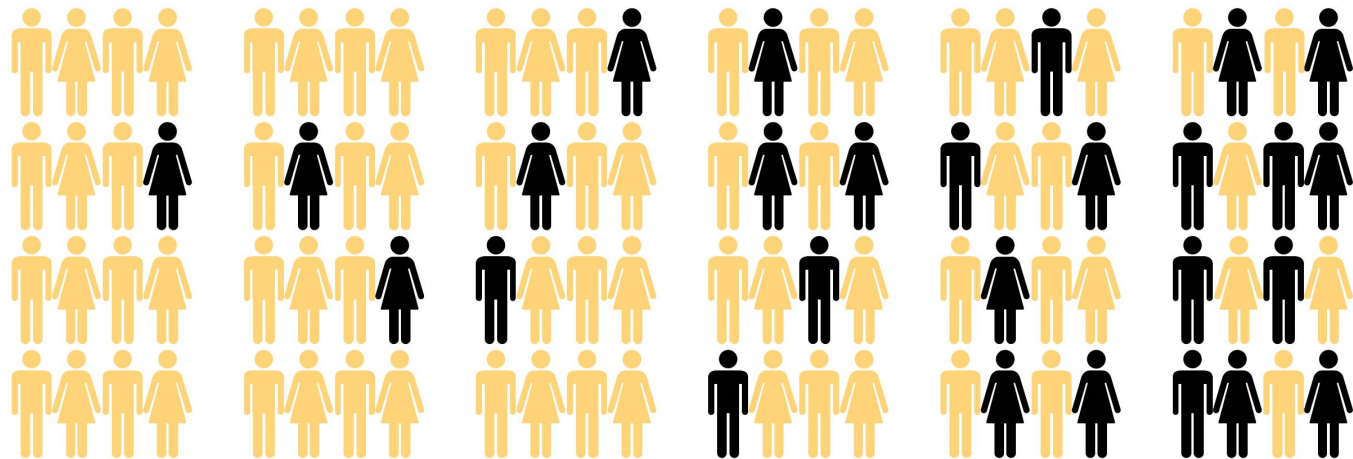
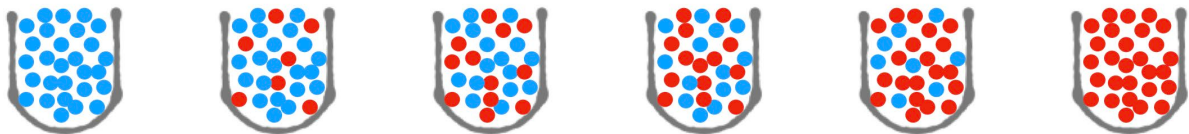
Barn har genetiske variasjoner felles med foreldrene sine.

-> Mer like sine foreldre enn tilfeldig person

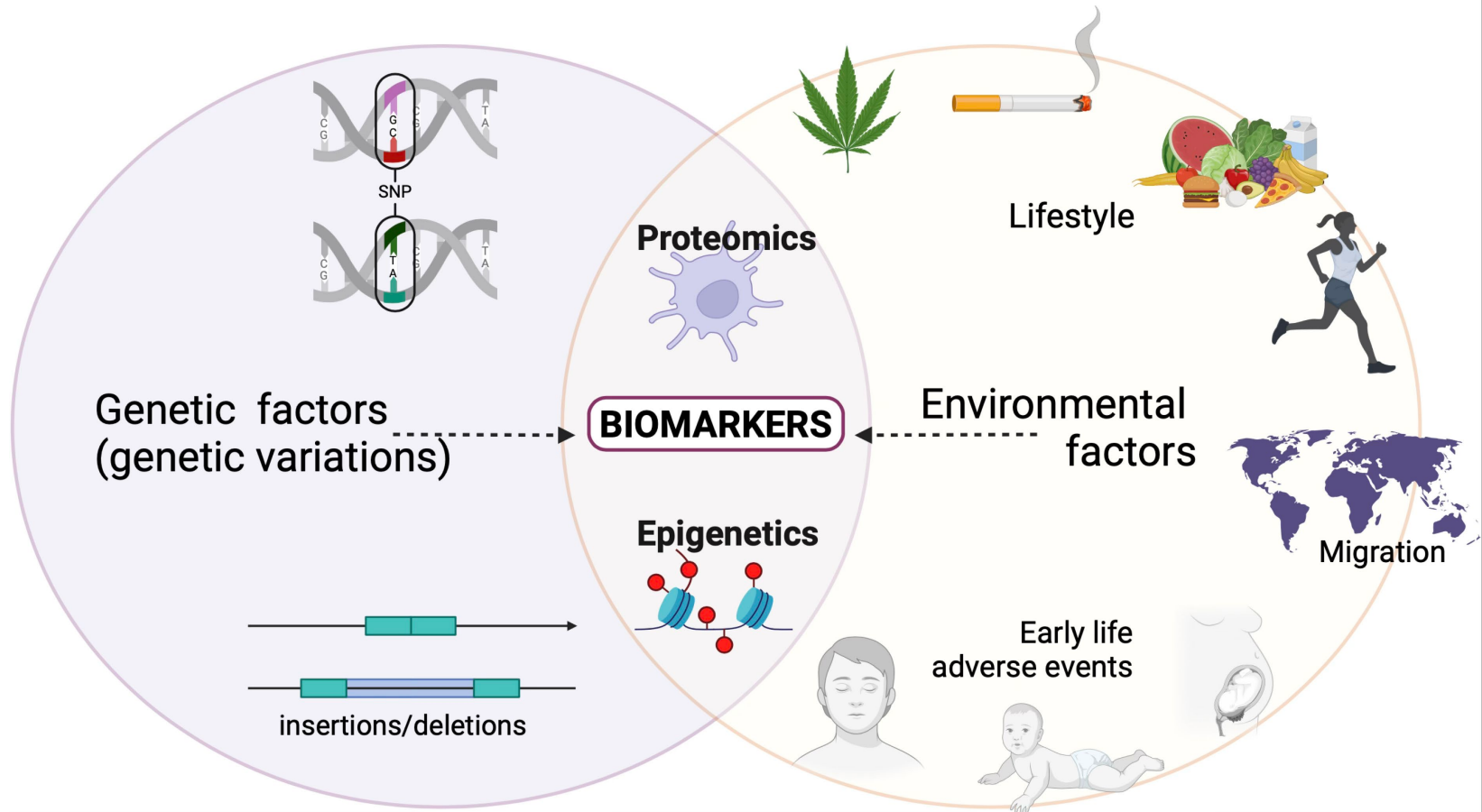
Seleksjon av høyre folk
Bedre mat / miljø
-> Økning i befolkning høyde

Kompleks genetikk sykdom (polygen arv)

Genetic Beskyttende Risiko



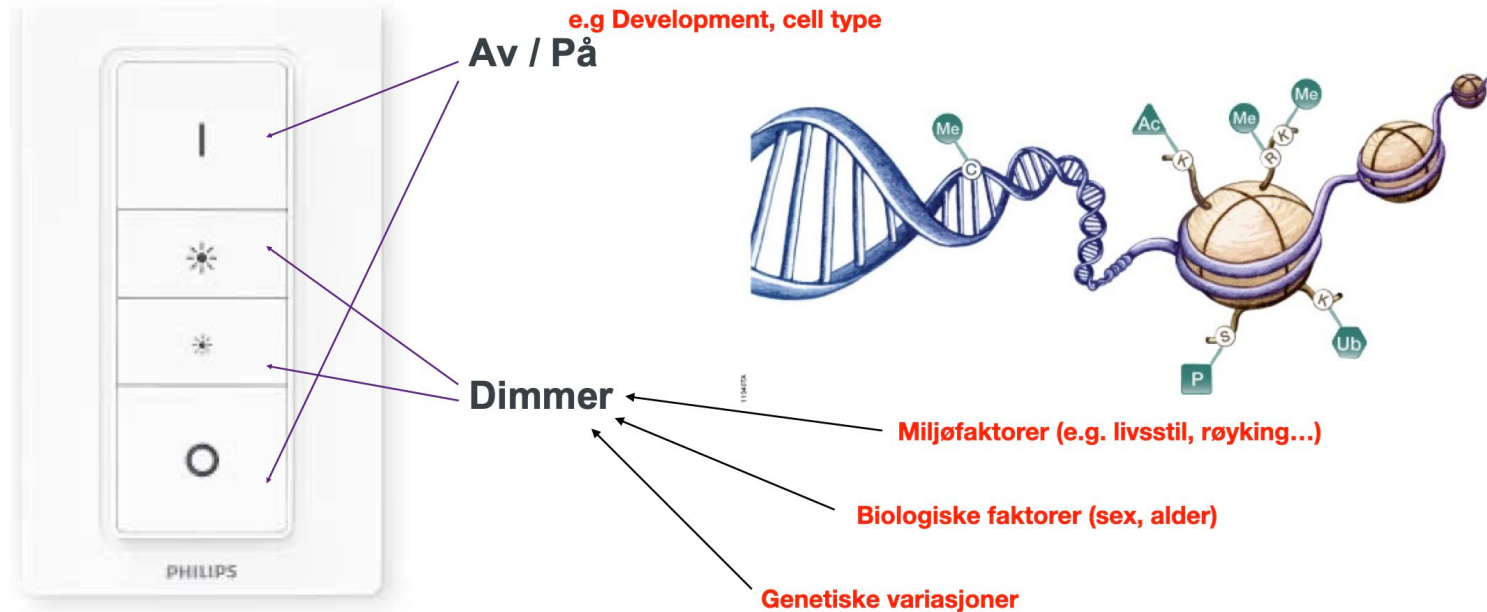
Kompleks trekk : genetisk + miljø

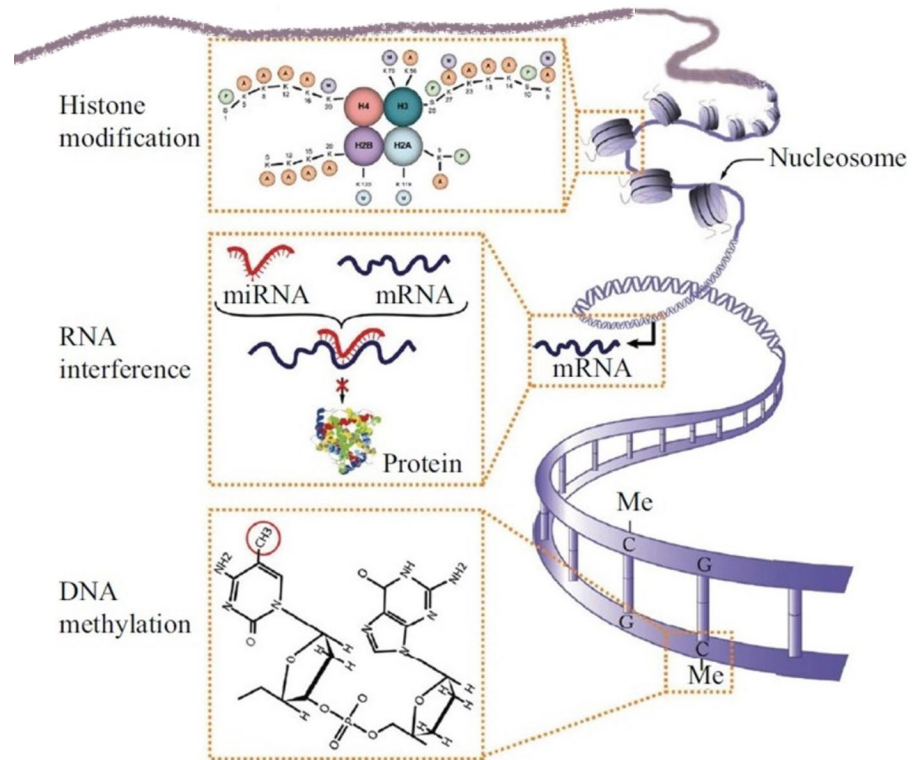
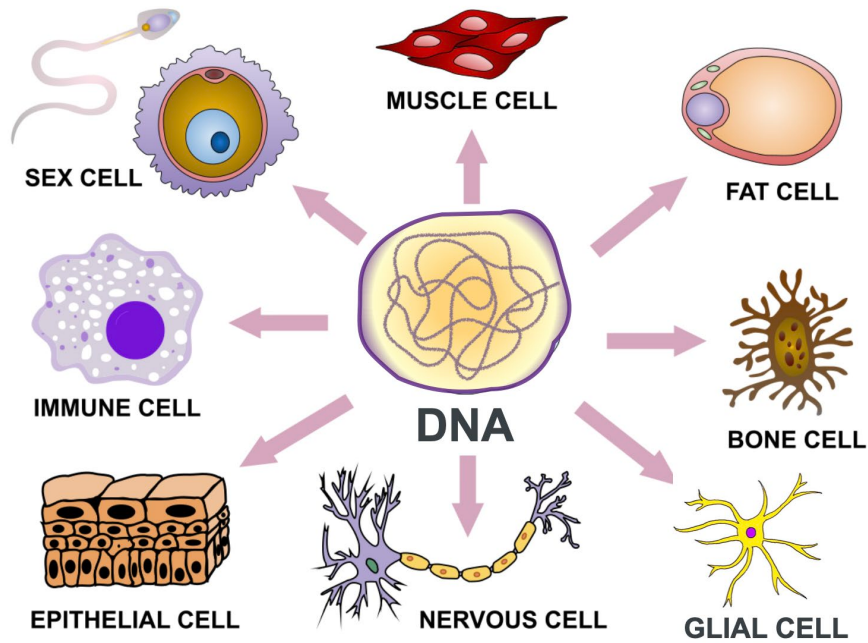


Kompleks trekk : genetisk + miljø

Hvordan miljø påvirker gener?

Ved å lage endringer på gener (uten å endre sekvens), epigenetikk modifikasjoner





Epigenetics

[View Teach.Genetics for Classroom Materials](#)

WHAT IS EPIGENETICS?

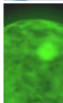
As an organism grows and develops, carefully orchestrated chemical reactions activate and deactivate parts of the genome at strategic times and in specific locations. Epigenetics is the study of these chemical reactions and the factors that influence them.



THE EPIGENOME AT A GLANCE

Meet the epigenome and learn how it influences DNA.

[video](#)



GENE CONTROL

Change the level of gene expression in a cell with the turn of a dial!

[interactive explore](#)



THE EPIGENOME LEARNS FROM ITS EXPERIENCES

Epigenetic tags record the gene-regulating signals the cell receives.

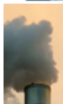
[learn more](#)



EPIGENETICS & INHERITANCE

Parents have a role in shaping the epigenomes of their offspring.

[learn more](#)



GENOMIC IMPRINTING

Certain genes are silenced during egg and sperm formation.

[learn more](#)

EPIGENETICS & THE ENVIRONMENT

The epigenome dynamically responds to the environment. Stress, diet, behavior, toxins, and other factors regulate gene expression.



INSIGHTS FROM IDENTICAL TWINS

Why do identical twins become more different as they age? See how the environment affects the epigenome in a pair of twins over time.

[explore](#)



CLICK YOUR RATS

What kind of mother are you? Care for a rat pup and shape its epigenome.

[interactive explore](#)



NUTRITION & THE EPIGENOME

What you eat can change your gene expression.

[learn more](#)



EPIGENETICS & THE HUMAN BRAIN

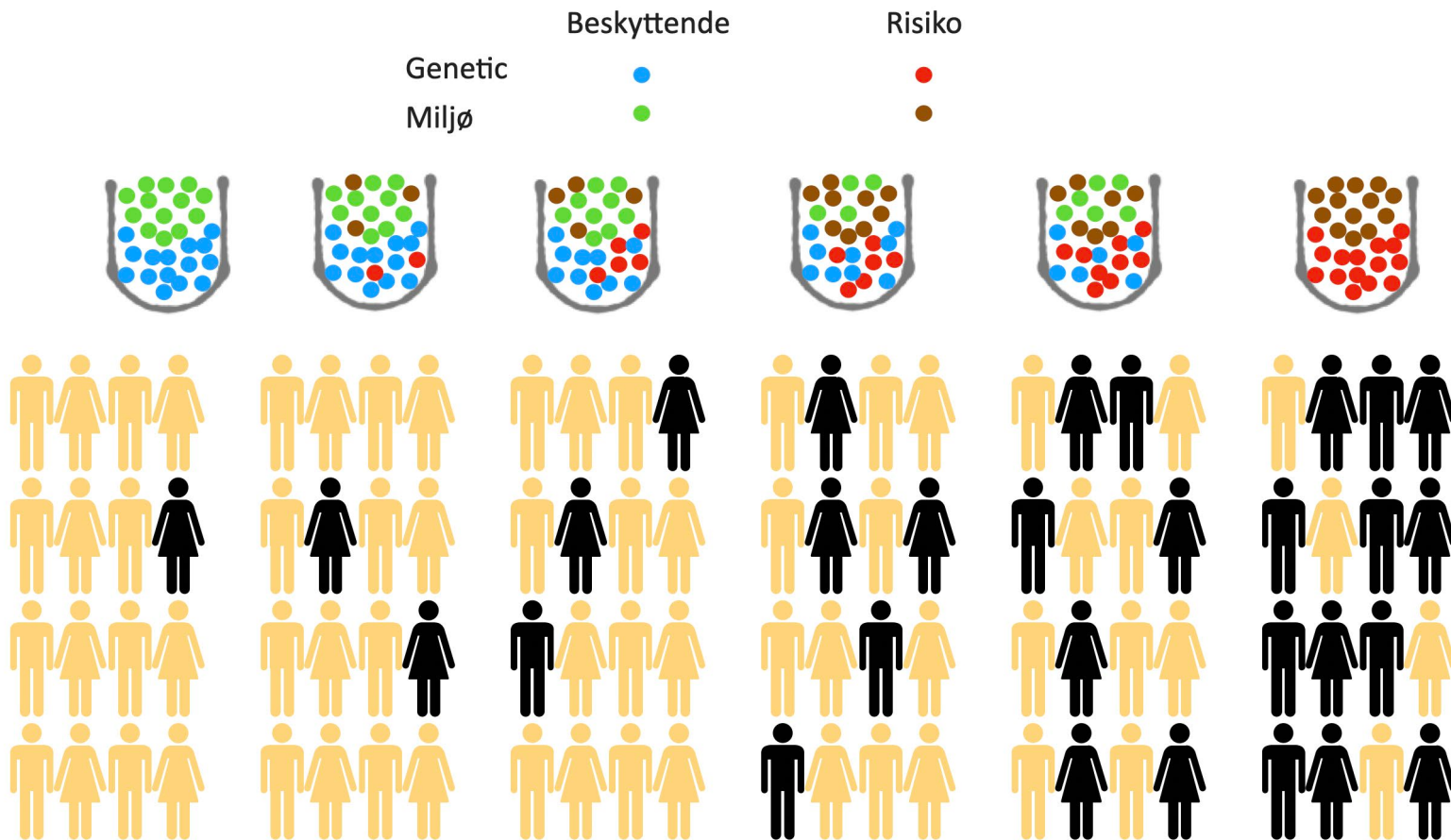
Epigenetic mechanisms play an important role both in normal brain function and in mental illness.

[video](#)



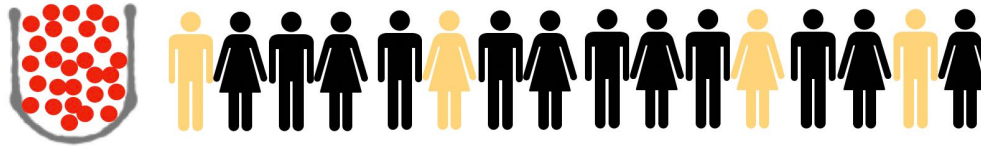
Men genetisk risiko samspiller med miljøet. **To modeller**

Modell . Additive effekter

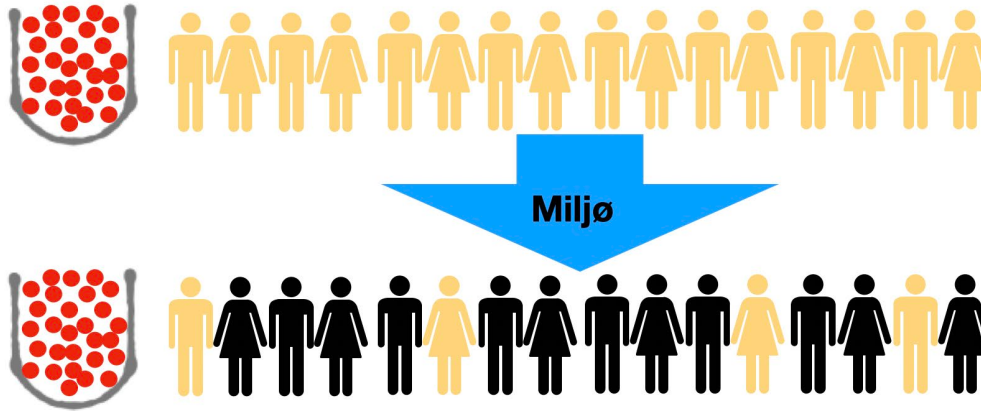


Men genetisk risiko samspiller med miljøet. **To modeller:**

Modell 2. Miljø som “trigger”
Eks. avhengighet



Alkoholavhengighet er påvirket i stor grad av genetiske faktorer.
Men uten tilgang til alkohol blir ingen avhengige:



UIB's enige etterutdanningsarena for lærere

Stephanie.lehellard@uib.no

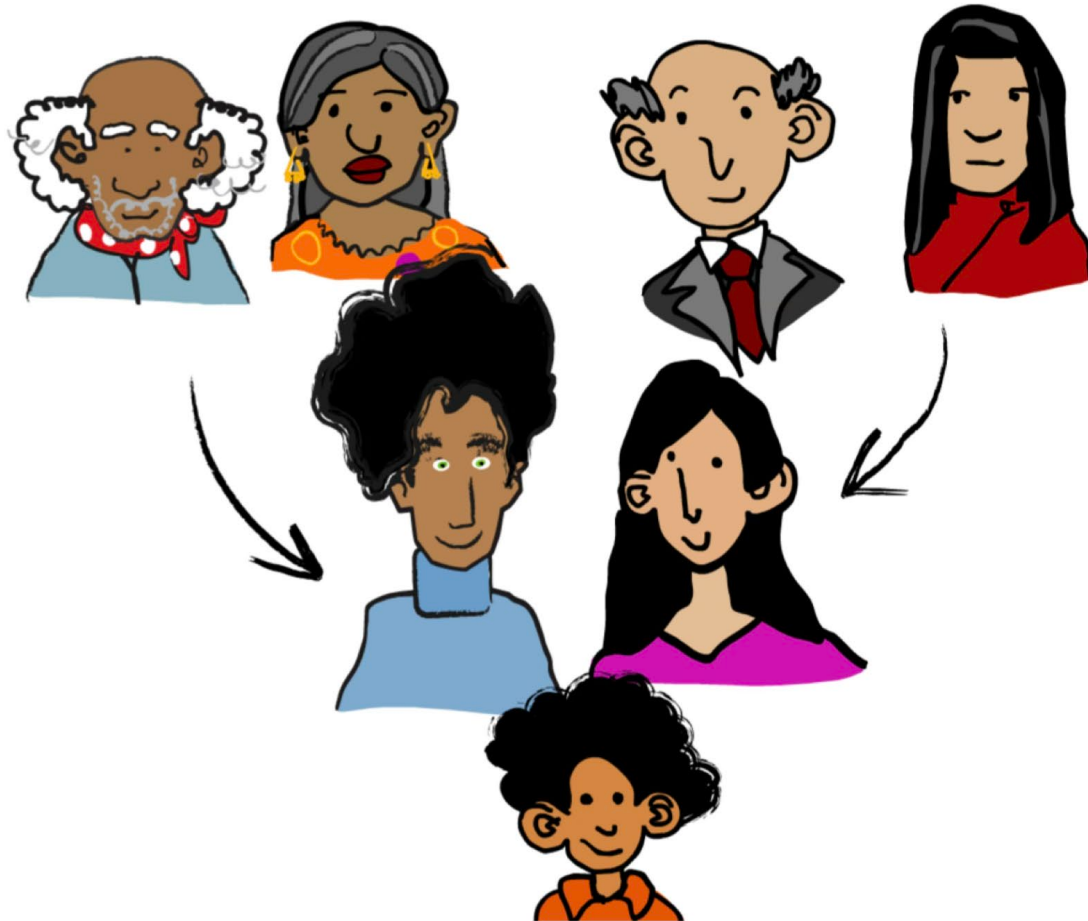
**Kan sende slides
Eller ta kontakt for
elegebesøk**



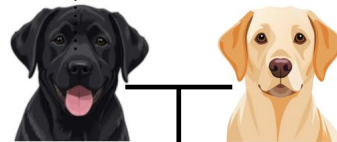
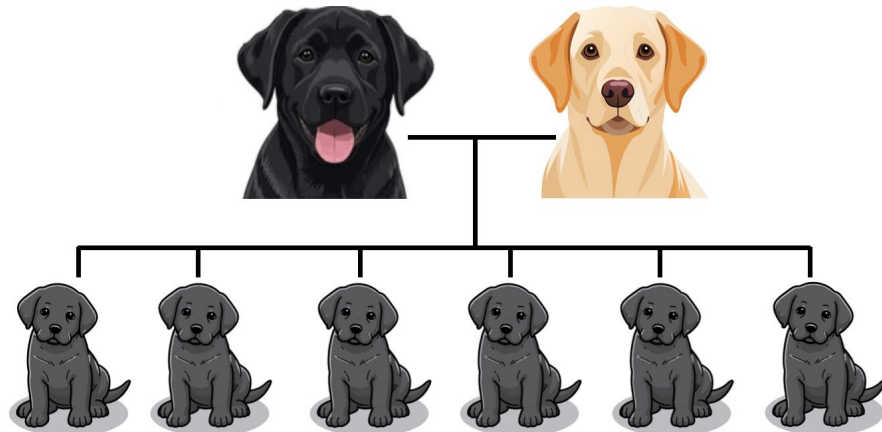
**Lærernes
dag 2026**



Lærernes
dag 2026



Dominant
Recessiv

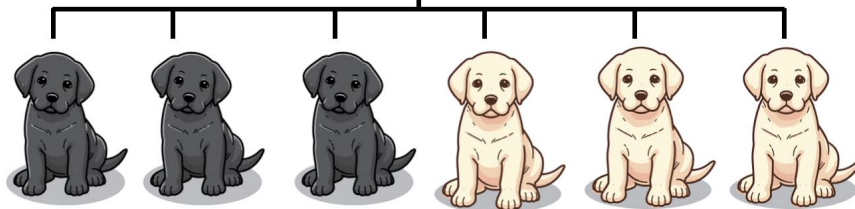


Which colour is dominant?

Black or beige?

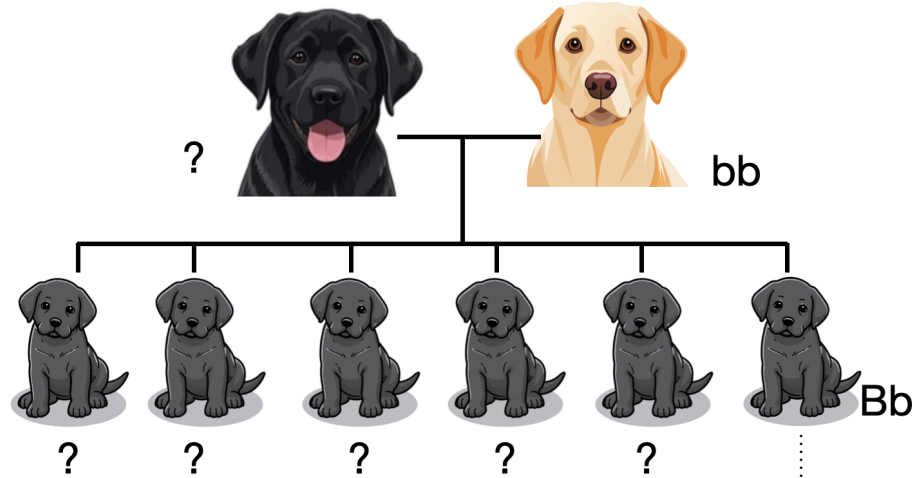
Black -> B

Beige -> b

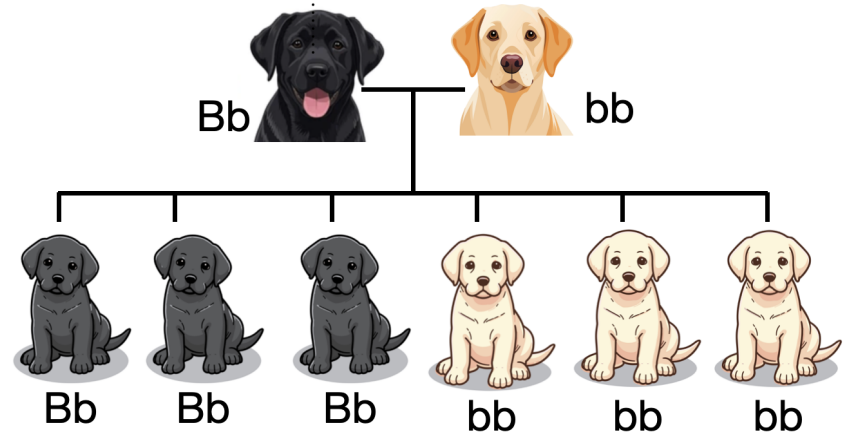


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Beige -> b

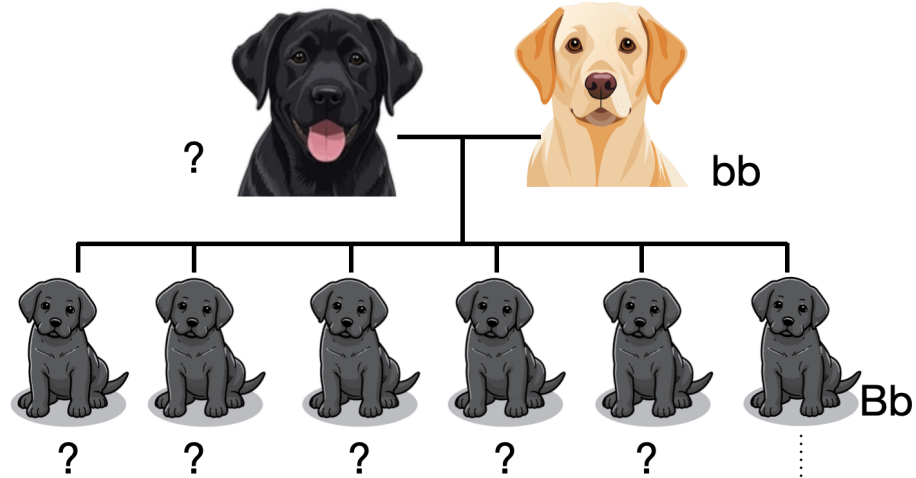


	B	b
b	Bb	bb
b	Bb	bb

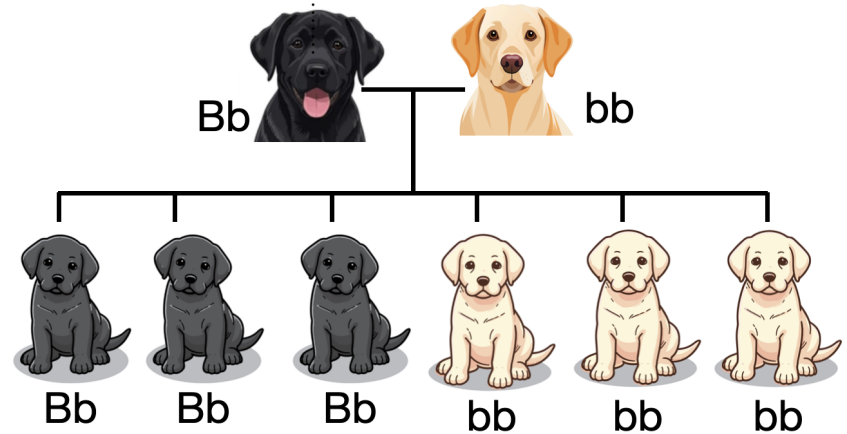


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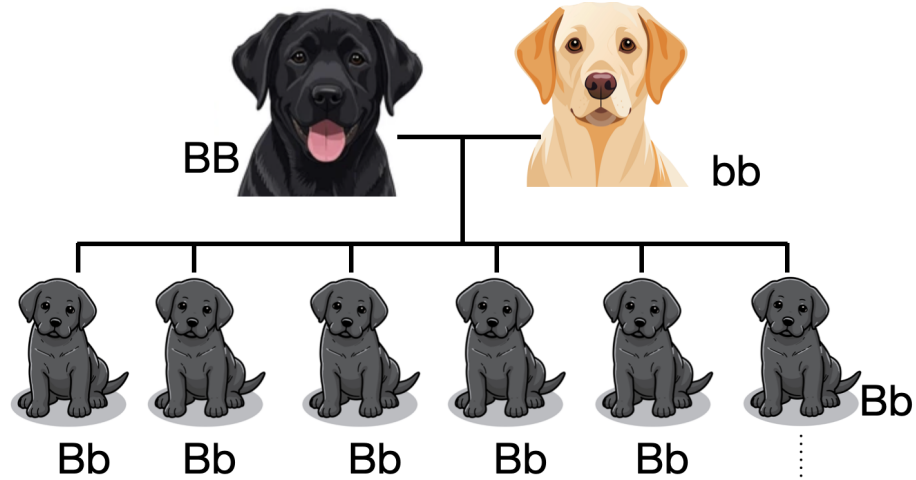


	B	b
b	Bb	bb
b	Bb	bb



Black -> B

Beige -> b



	B	b
b	Bb	bb
b	Bb	bb

	B	B
b	Bb	Bb
b	Bb	Bb

Likelihood - sannsynlighet

