

Inbreeding

Differences driven by patterns of gene flow/ interbreeding…

- True when looking at different populations
  - BUT doesn't *have to* be "between populations"
- Related concept: **inbreeding**
  - Breeding between closely related individuals
  - Capacity for dispersal often limited
  - *Changes distribution of genotypes*

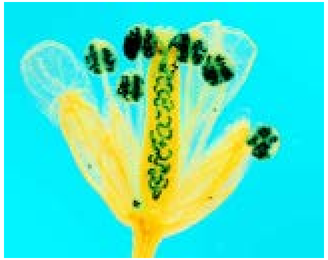
An extreme form of inbreeding: self-fertilization

Gen0 25% AA 50% Aa 25% aa

A thale cress plant is caught in the act of self-reproduction in this photograph by a team at Tallinn University of Technology in Estonia.

Greenish stain reveals the plant's pollen—both inside the plant's pistil (center), which leads down to the ovary, and on six anthers, which produce the pollen and are shown growing toward the pistil's sticky opening.

An extreme form of inbreeding: self-fertilization



Gen0	25% AA	50% Aa	25% aa
	25% AA	¼ (50%) AA, ½ (50%) Aa, ¼ (50%)aa	25%aa

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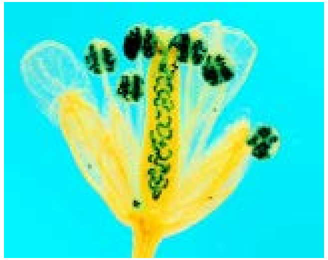

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Gen1	37.5% AA	25% Aa	37.5% aa

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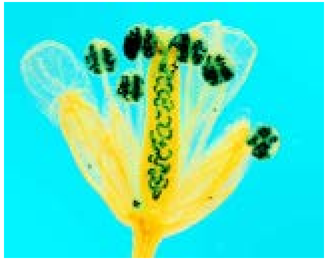

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Gen1	37.5% AA	25% Aa	37.5% aa
Gen2	43.75% AA	12.5% Aa	43.75% aa

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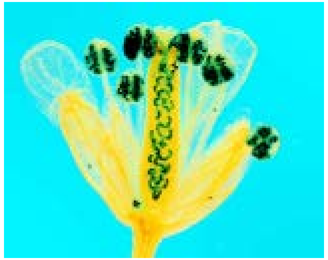

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Gen2	43.75% AA	12.5% Aa	43.75% aa
GenX	50% AA	0% Aa	50% aa

Every generation, heterozygote fraction goes down, and feeds alleles into homozygotes... creates "pure breeding" lines

Quantifying inbreeding...

- Inbreeding (even if not “selfing”) reduces % heterozygotes
  - Feeds alleles into homozygotes
- Reduction in % heterozygotes from HW expected quantifies inbreeding

Measure we'll use: Wright's inbreeding coefficient, F

- F ranges from 0 to 1

  0 : at HW expectation for % heterozygotes

  0 < F < 1 : somewhat fewer heterozygotes than predicted

  1 : no heterozygotes

- $$F = \frac{\text{HW predicted } 2pq - \%\text{ observed hetz}}{\text{HW predicted } 2pq}$$

F calculation example

- Studying a population with the following genotypes:

AA	553
Aa	294
aa	153

F calculation example

- Studying a population with the following genotypes:

AA	553	= 0.553	p(A) = 0.7
Aa	294	= 0.294	q(a) = 0.3
aa	153	= 0.153	

N=1000

F calculation example

- Studying a population with the following genotypes:

AA	553	= 0.553	p(A) = 0.7
Aa	294	= 0.294	q(a) = 0.3
aa	153	= 0.153	

N=1000

HW expectation

AA:	0.49
Aa:	0.42
aa:	0.09

F calculation example

- Studying a population with the following genotypes:

AA	553	= 0.553	p(A) = 0.7
Aa	294	= 0.294	q(a) = 0.3
aa	153	= 0.153	

N=1000

F = (0.42-0.294)/0.42
= 0.30

<u>HW expectation</u>

AA:	0.49
Aa:	0.42
aa:	0.09

F calculation practice

- Genotypes on Croatian islands:

AA	0.8136
Aa	0.1728
aa	0.0136

- Calculate **F**!

F calculation practice

- Genotypes on Croatian islands:

AA	0.8136
Aa	0.1728
aa	0.0136

F calculation practice

- Genotypes on Croatian islands:

AA	0.8136	p=0.9
Aa	0.1728	q=0.1
aa	0.0136	

- **F** = (0.18-0.1728)/0.18 = 0.04

Inbreeding F vs F_{ST}

- Both based on same principle
  - Seeing fewer heterozygotes than HW prediction
  - Indicates *"non-random mating"*
  - Symptom of "Wahlund effect"

- Apply **inbreeding F**
  - Looking at individuals *within one population*
- Apply $\mathbf{F_{ST}}$
  - Quantifying difference *between populations*

How is this calculation used?

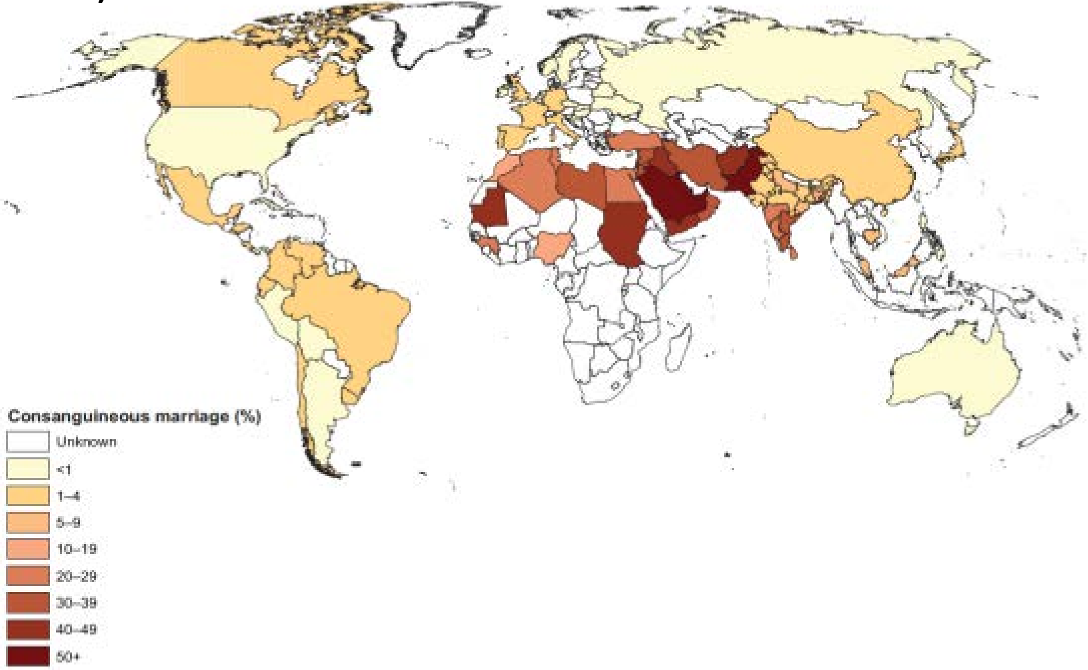
- Association between inbreeding and health/ disease
  - *Inbreeding effects on fetal growth in Beirut, Lebanon*

- Patterns of gene flow between “social classes”
  - *Consanguineous marriage within social/occupational class boundaries in Pakistan*

- Other cultural effects on patterns of breeding
  - *Consanguinity in Spain: Socioeconomic, demographic, and geographic influences*

Inbreeding around the world

Global distribution of marriages between couples related as second cousins or closer ($F \geq 0.0156$).



… but isn't inbreeding "bad"…?

- BY ITSELF, inbreeding only changes the distribution of alleles among genotypes
  - DOES NOT make any alleles "go away"

- Nonetheless, many know of "inbreeding depression"... This requires NATURAL SELECTION as well as inbreeding...

Populations often harbor many *individually* rare recessive mutations

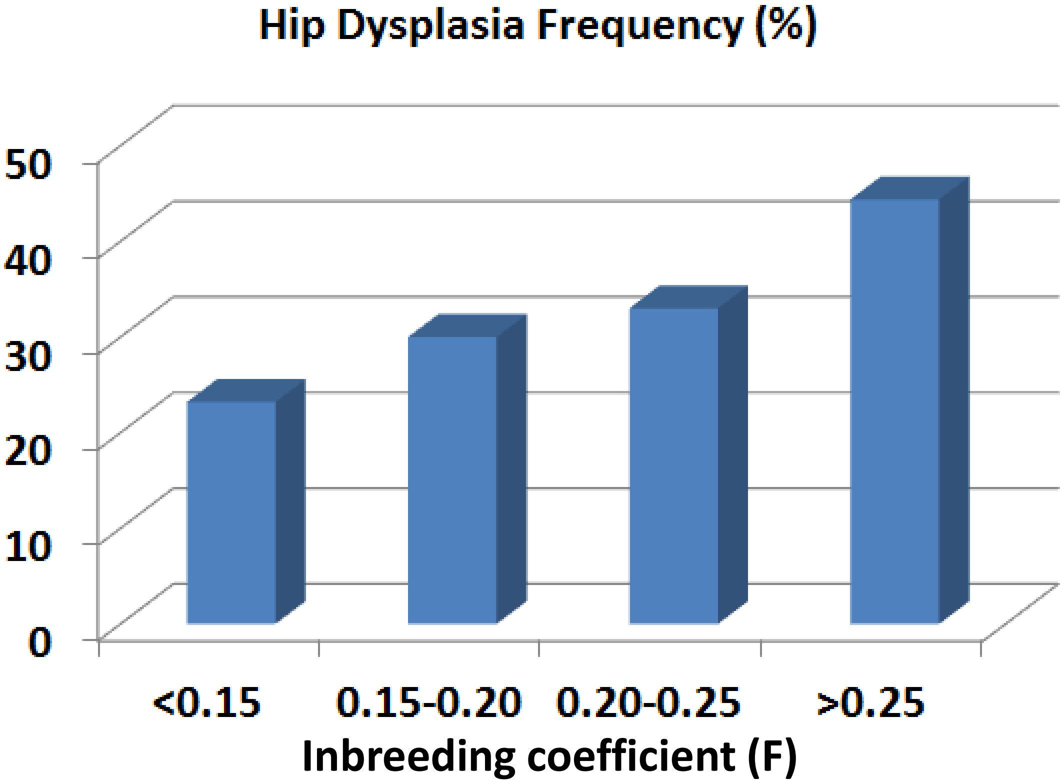
- A few new detrimental mutations each generation
- Effects often not seen since both rare and recessive
  - Very few homozygotes
- BUT if mate two relatives:
  - Likely to have same recessive mutation
  - More likely to produce homozygous offspring

AABBCCDDEeFF	AABBCCDDEEFF	AABBCCDDEEFF	AABBCCDDEEFF
AABbCCDDEEFF	AABBCCDDEEFF	AABBCCDDEEFF	AABBCCDDEEFF
AABBCCDDEEFF	AABBCCDDEEFF	AABBCCDDEEFF	AABBCCDdEEFF
AABBCCDDEEFF	AABBCCDdEEFF	AABBCCDDEEFF	AABBCCDDEEFF
AABBCCDDEEFF	AABBCCDDEEFF	AABBCCDDEEFF	AaBBCCDDEEFF

Dog breeds & inbreeding

- Breeding of relatives has made many dog "breeds"
  - Maintained "desirable" qualities
- Inbreeding coefficient (F) in some dogs > 0.5! (unusual)
  - Some poodle varieties have F=0.7
  - UPenn study showed 75% puppies with F>0.67 die within 10 days
  - King Charles spaniels get syringomyelia
    - Dog's skull is too small for its brain
  - Boxers- high epilepsy; Pugs- breathing problems; Bulldogs- unable to mate or give birth unassisted; Mastiffs, Saint Bernards, Great Dane- hip dysplasia

Hip dysplasia with inbreeding…



Icelandic sheepdogs have more hip dysplasia with inbreeding

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