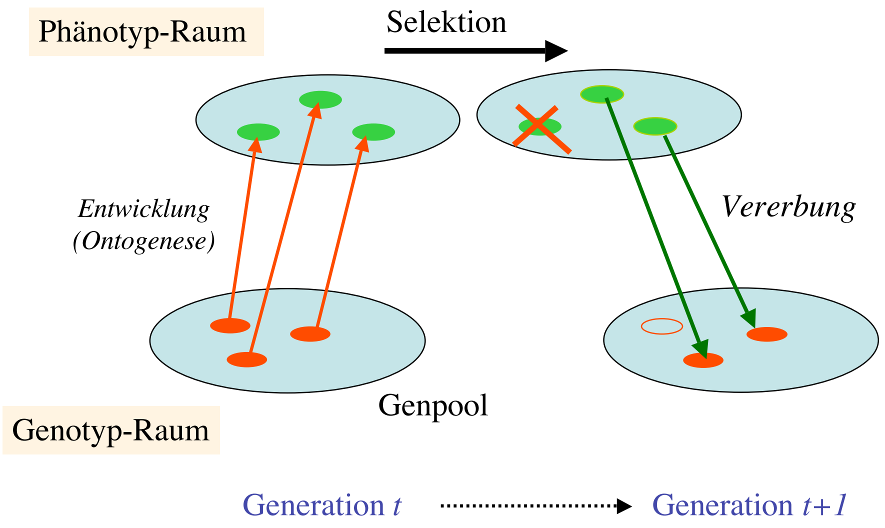
Ursprung und Erhaltung genetischer Variation

Thema 5

Genotyp und Phänotyp



Phänotyp-Raum

Selektion

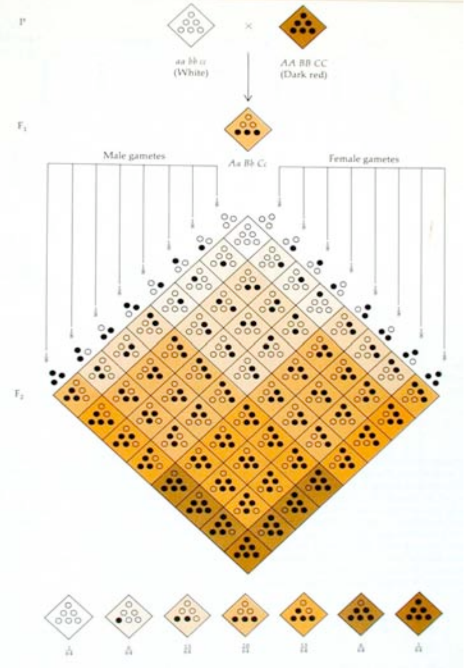
Entwicklung (Ontogenese)

Vererbung

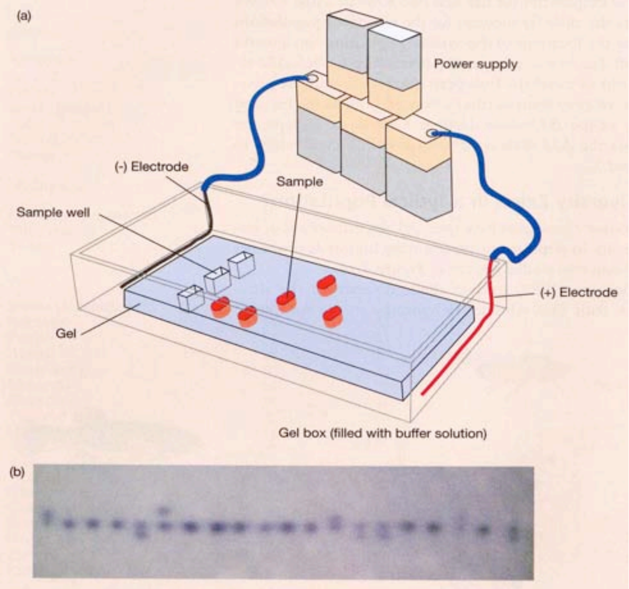
Genpool

Genotyp-Raum

Generation *t* → Generation *t+1*

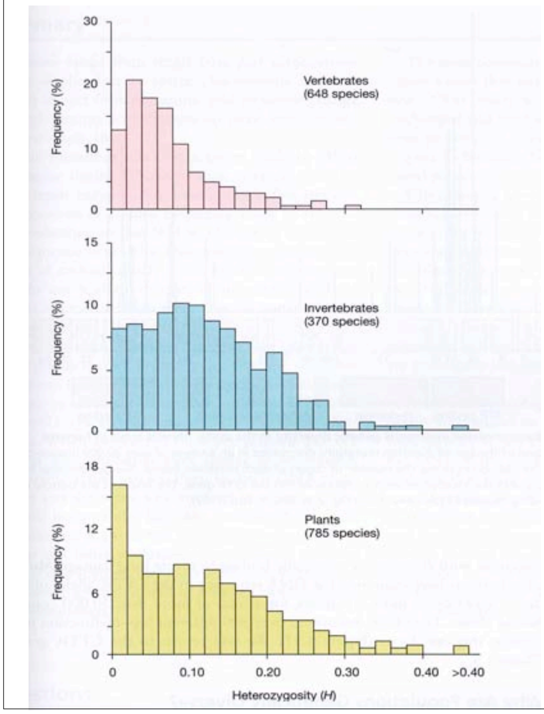

P
aa bb cc
(White)
×
AA BB CC
(Dark red)
F1
Aa Bb Cc
Male gametes
Female gametes
F2

Ayala

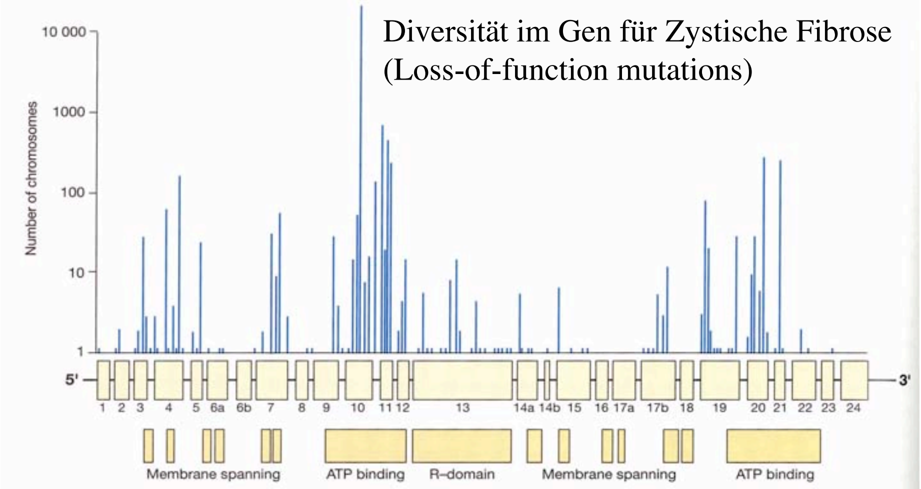

(a)
Power supply
(-) Electrode
Sample
Sample well
(+) Electrode
Gel
Gel box (filled with buffer solution)
(b)

FH3, Box 4.1

Ursprung u. Erhaltung genetischer Variation



FH3, Fig 4.16



Diversität im Gen für Zystische Fibrose
(Loss-of-function mutations)

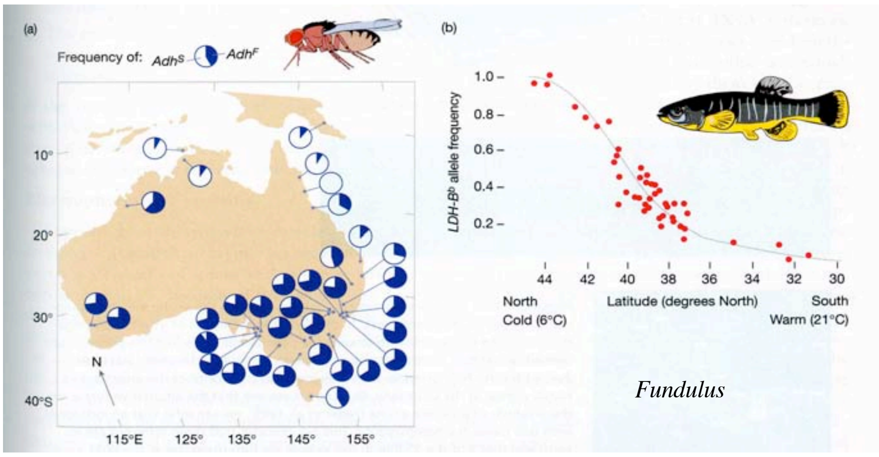
FH3, Fig 4.17

H = % der heterozygoten Loci unter allen Individuen

Locus	Enzym	*H*
Acph	Acid phosphotase	0.52
Pgm-1	Phosphoglucomutase-1	0,36
Pgm-2	Phosphoglucomutase-2	0.38
Adk	Adenylate kinase	0,09
Pept-A	Peptidase-A	0,37
Pept-C	Peptidase-C	0,02
Pept-D	Peptidase-D	0.02
Adn	Adenosine deaminase	0.11
6Pgdh	6-Phosphogluconate dehydrogenase	0.05
Aph	Alkaline phosphatase (placental)	0.53
Amy	Amylase (pancreatic)	0.09
Gpt	Glutamate-pyruvate transaminase	0.50

(gemessen: 71 Loci bei Europäern)

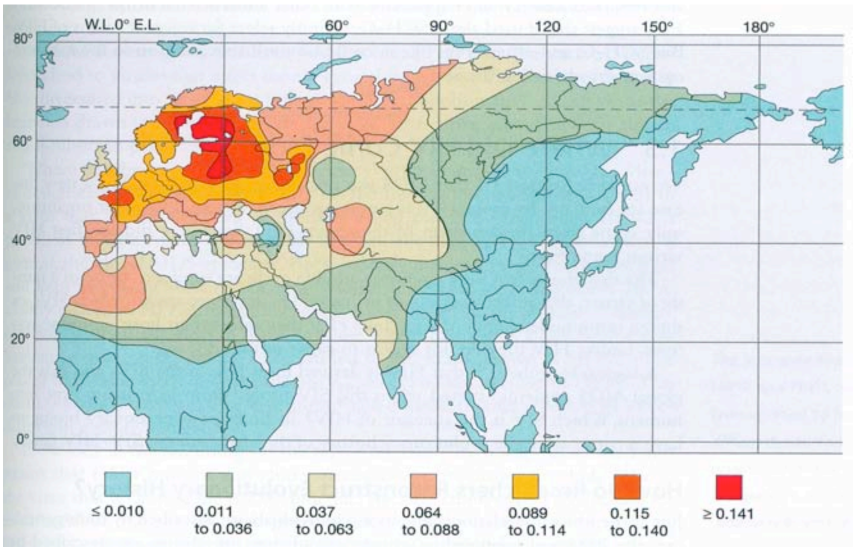
after Harris & Hopkinson (1972) J. Human Genet 36:9
(Ayala 1982, Tab.2.9)



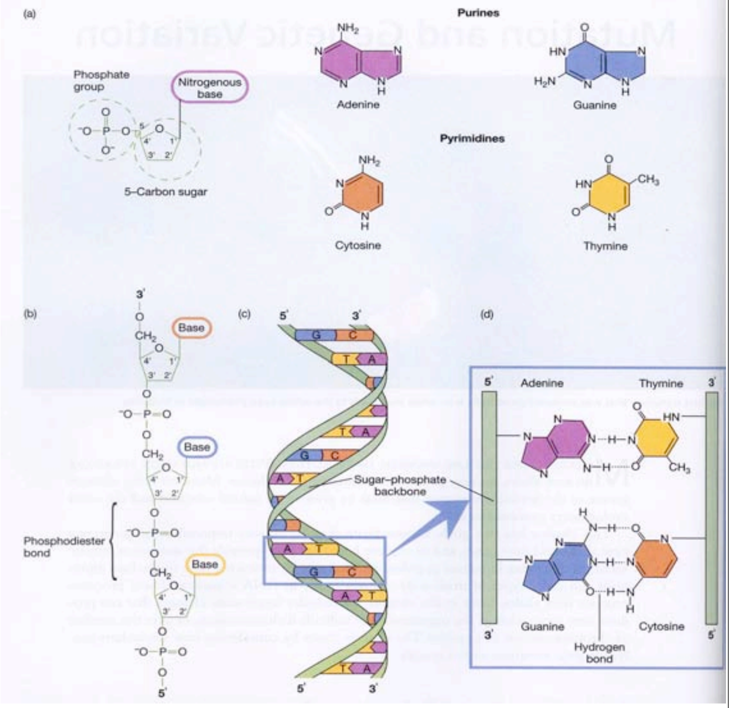
Fundulus

Drosophila

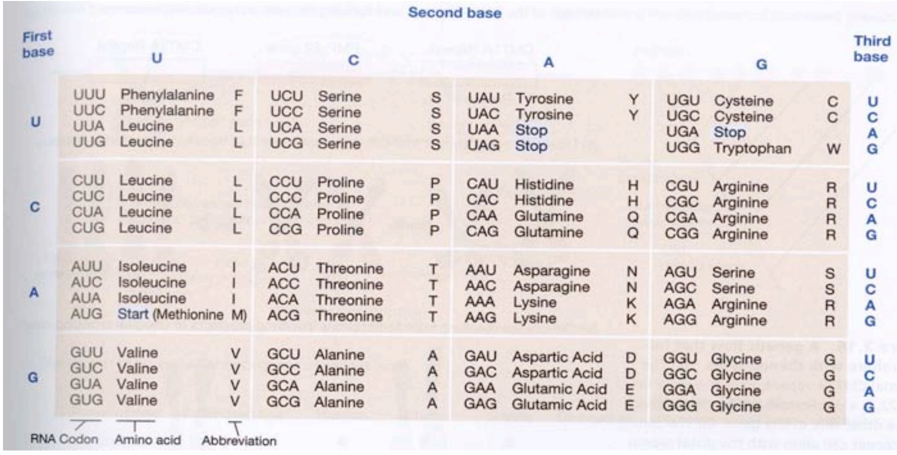
FH3, Fig 4.14


W.L.0° E.L.
60°
90°
120
150°
180°
80°
60°
40°
20°
0°
≤ 0.010
0.011 to 0.036
0.037 to 0.063
0.064 to 0.088
0.089 to 0.114
0.115 to 0.140
≥ 0.141

FH3, Fig 1.11


(a)
Purines
Phosphate group
Nitrogenous base
5-Carbon sugar
Adenine
Guanine
Pyrimidines
Cytosine
Thymine
(b)
(c)
(d)
Base
Phosphodiester bond
Sugar-phosphate backbone
5'
3'
Adenine
Thymine
Guanine
Cytosine
Hydrogen bond

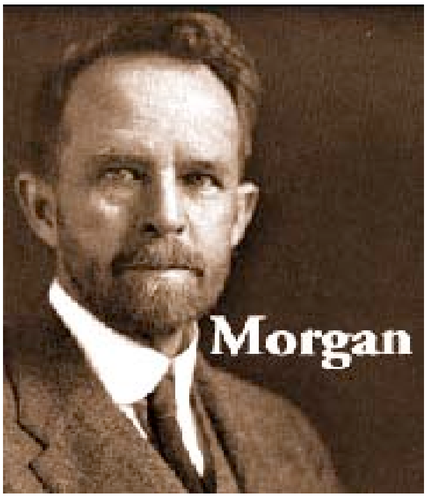
FH3, Fig 4.1



First base	Second base: U			C			A			G			Third base
U	UUU	Phenylalanine	F	UCU	Serine	S	UAU	Tyrosine	Y	UGU	Cysteine	C	U
	UUC	Phenylalanine	F	UCC	Serine	S	UAC	Tyrosine	Y	UGC	Cysteine	C	C
	UUA	Leucine	L	UCA	Serine	S	UAA	Stop		UGA	Stop		A
	UUG	Leucine	L	UCG	Serine	S	UAG	Stop		UGG	Tryptophan	W	G
C	CUU	Leucine	L	CCU	Proline	P	CAU	Histidine	H	CGU	Arginine	R	U
	CUC	Leucine	L	CCC	Proline	P	CAC	Histidine	H	CGC	Arginine	R	C
	CUA	Leucine	L	CCA	Proline	P	CAA	Glutamine	Q	CGA	Arginine	R	A
	CUG	Leucine	L	CCG	Proline	P	CAG	Glutamine	Q	CGG	Arginine	R	G
A	AUU	Isoleucine	I	ACU	Threonine	T	AAU	Asparagine	N	AGU	Serine	S	U
	AUC	Isoleucine	I	ACC	Threonine	T	AAC	Asparagine	N	AGC	Serine	S	C
	AUA	Isoleucine	I	ACA	Threonine	T	AAA	Lysine	K	AGA	Arginine	R	A
	AUG	Start (Methionine	M)	ACG	Threonine	T	AAG	Lysine	K	AGG	Arginine	R	G
G	GUU	Valine	V	GCU	Alanine	A	GAU	Aspartic Acid	D	GGU	Glycine	G	U
	GUC	Valine	V	GCC	Alanine	A	GAC	Aspartic Acid	D	GGC	Glycine	G	C
	GUA	Valine	V	GCA	Alanine	A	GAA	Glutamic Acid	E	GGA	Glycine	G	A
	GUG	Valine	V	GCG	Alanine	A	GAG	Glutamic Acid	E	GGG	Glycine	G	G

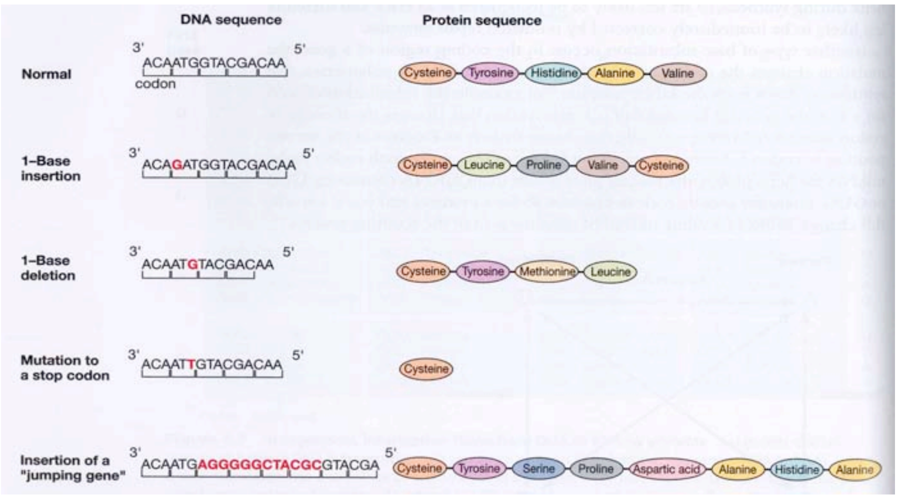
RNA Codon Amino acid Abbreviation

FH3, Fig 2.15

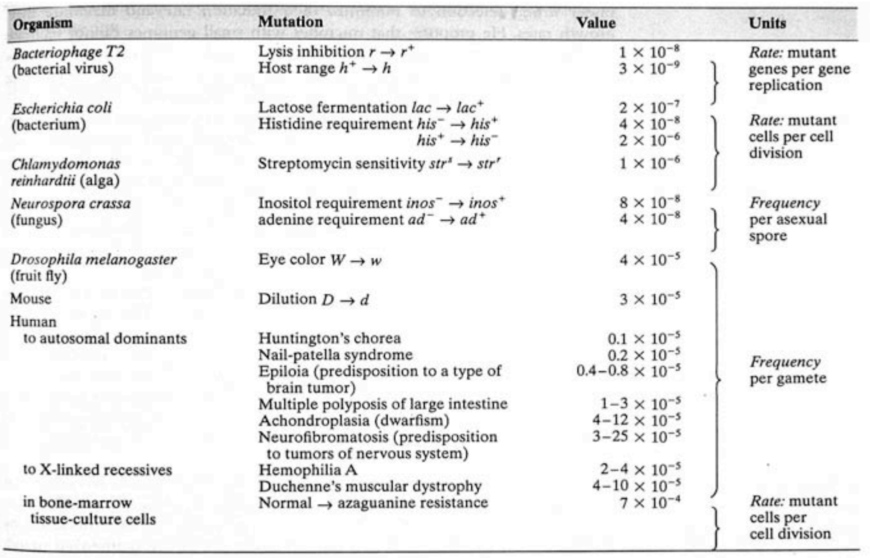


Sewall Wright

Ursprung u. Erhaltung genetischer Variation

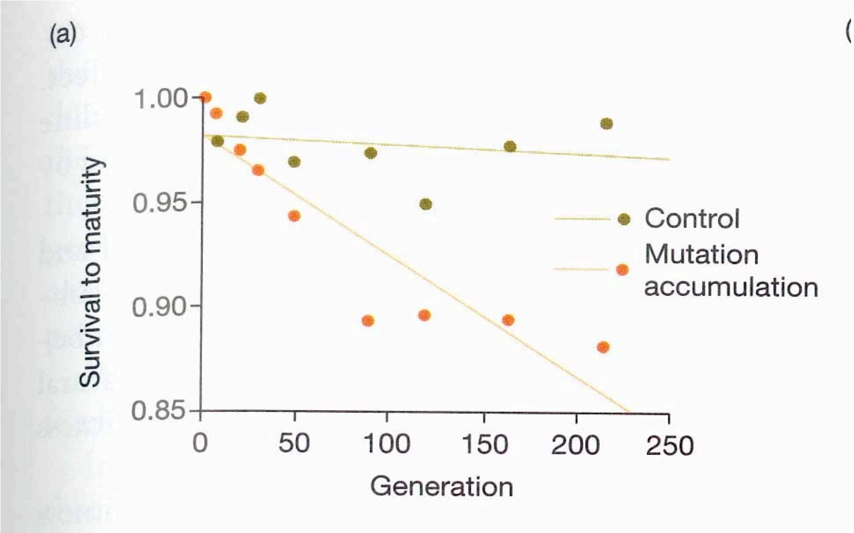

DNA sequence
Protein sequence
Normal
3'
ACAATGGTACGACAA
5'
codon
Cysteine
Tyrosine
Histidine
Alanine
Valine
1–Base insertion
3'
ACAGATGGTACGACAA
5'
Cysteine
Leucine
Proline
Valine
Cysteine
1–Base deletion
3'
ACAATGTACGACAA
5'
Cysteine
Tyrosine
Methionine
Leucine
Mutation to a stop codon
3'
ACAATTGTACGACAA
5'
Cysteine
Insertion of a "jumping gene"
3'
ACAATGAGGGGGCTACGCGTACGA
5'
Cysteine
Tyrosine
Serine
Proline
Aspartic acid
Alanine
Histidine
Alanine

FH3, Fig 4..5

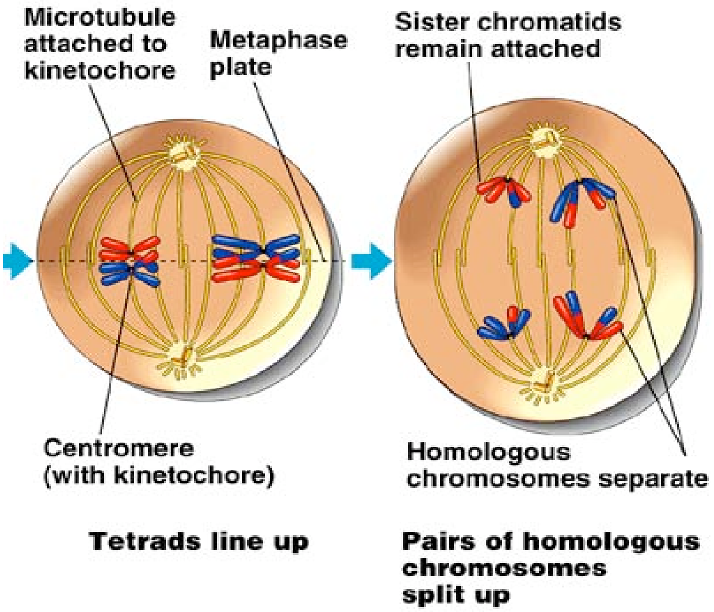


Organism	Mutation	Value	Units
Bacteriophage T2 (bacterial virus)	Lysis inhibition $r \to r^+$	1×10^{-8}	*Rate:* mutant genes per gene replication
	Host range $h^+ \to h$	3×10^{-9}	
Escherichia coli (bacterium)	Lactose fermentation $lac \to lac^+$	2×10^{-7}	*Rate:* mutant cells per cell division
	Histidine requirement $his^- \to his^+$	4×10^{-8}	
	$his^+ \to his^-$	2×10^{-6}	
Chlamydomonas reinhardtii (alga)	Streptomycin sensitivity $str^s \to str^r$	1×10^{-6}	
Neurospora crassa (fungus)	Inositol requirement $inos^- \to inos^+$	8×10^{-8}	*Frequency* per asexual spore
	adenine requirement $ad^- \to ad^+$	4×10^{-8}	
Drosophila melanogaster (fruit fly)	Eye color $W \to w$	4×10^{-5}	*Frequency* per gamete
Mouse	Dilution $D \to d$	3×10^{-5}	
Human			
to autosomal dominants	Huntington's chorea	0.1×10^{-5}	
	Nail-patella syndrome	0.2×10^{-5}	
	Epiloia (predisposition to a type of brain tumor)	$0.4–0.8 \times 10^{-5}$	
	Multiple polyposis of large intestine	$1–3 \times 10^{-5}$	
	Achondroplasia (dwarfism)	$4–12 \times 10^{-5}$	
	Neurofibromatosis (predisposition to tumors of nervous system)	$3–25 \times 10^{-5}$	
to X-linked recessives	Hemophilia A	$2–4 \times 10^{-5}$	
	Duchenne's muscular dystrophy	$4–10 \times 10^{-5}$	
in bone-marrow tissue-culture cells	Normal → azaguanine resistance	7×10^{-4}	*Rate:* mutant cells per cell division

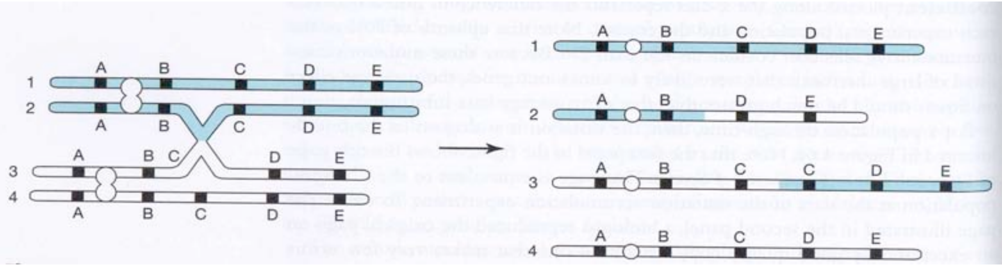
FH, Tab.4.2, 4.1



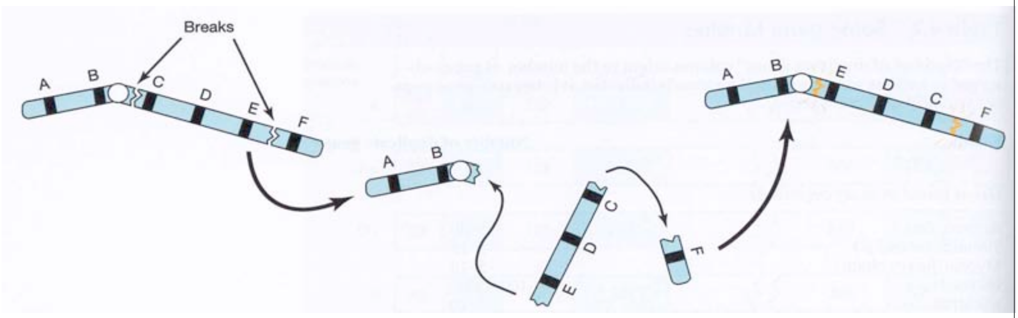
FH3, Fig 4.6


Microtubule attached to kinetochore
Metaphase plate
Sister chromatids remain attached
Centromere (with kinetochore)
Homologous chromosomes separate
Tetrads line up
Pairs of homologous chromosomes split up

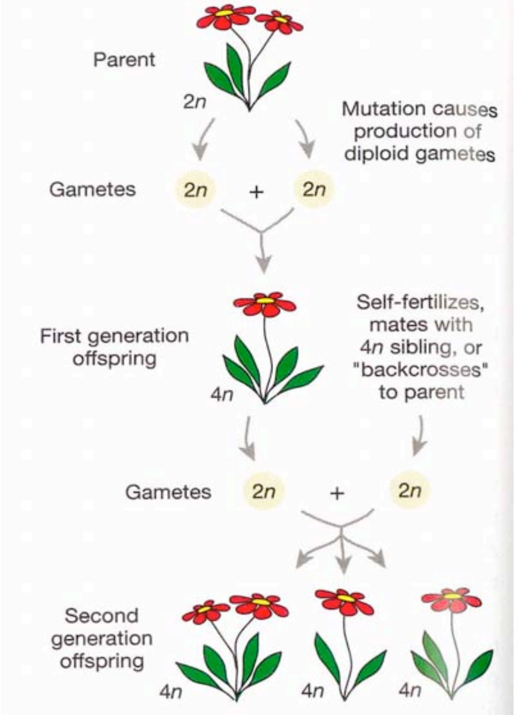
Campbell 6, Fig.13.9



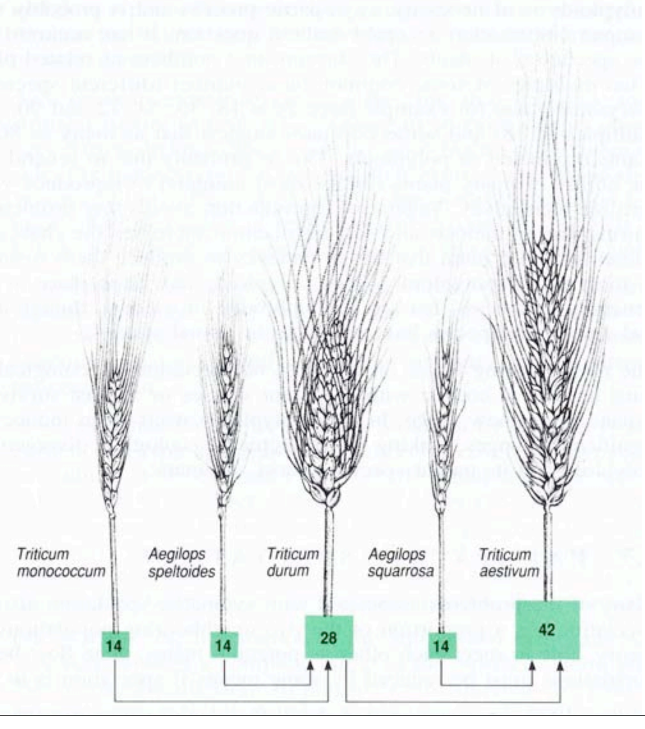
FH3, Fig 4.7


Breaks
A
B
C
D
E
F

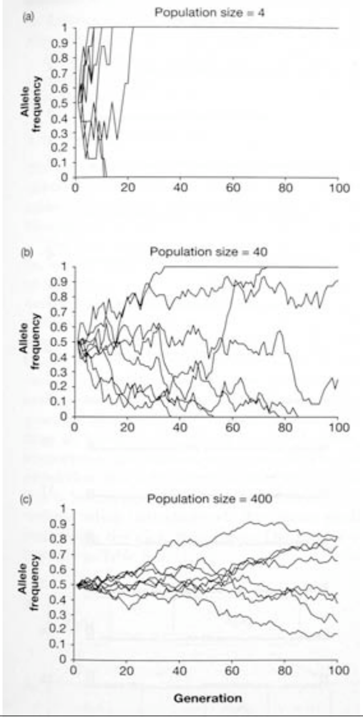
FH3, Fig 4.10

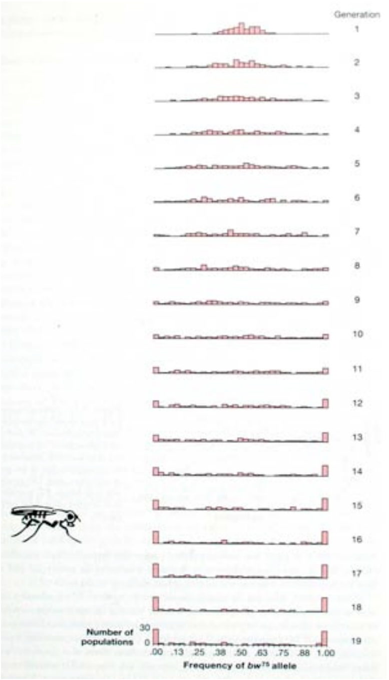

Parent
2n
Mutation causes production of diploid gametes
Gametes
2n
+
2n
First generation offspring
4n
Self-fertilizes, mates with 4n sibling, or "backcrosses" to parent
Gametes
2n
+
2n
Second generation offspring
4n
4n
4n

FH3, Fig 4.12


Triticum monococcum
Aegilops speltoides
Triticum durum
Aegilops squarrosa
Triticum aestivum
14
14
28
14
42

Skelton, Fig 9.25


(a)
Population size = 4
Allele frequency
0
0.1
0.2
0.3
0.4
0.5
0.6
0.7
0.8
0.9
1
0
20
40
60
80
100
(b)
Population size = 40
(c)
Population size = 400
Generation


Generation
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
Number of populations
30
0
.00
.13
.25
.38
.50
.63
.75
.88
1.00
Frequency of bw^{75} allele

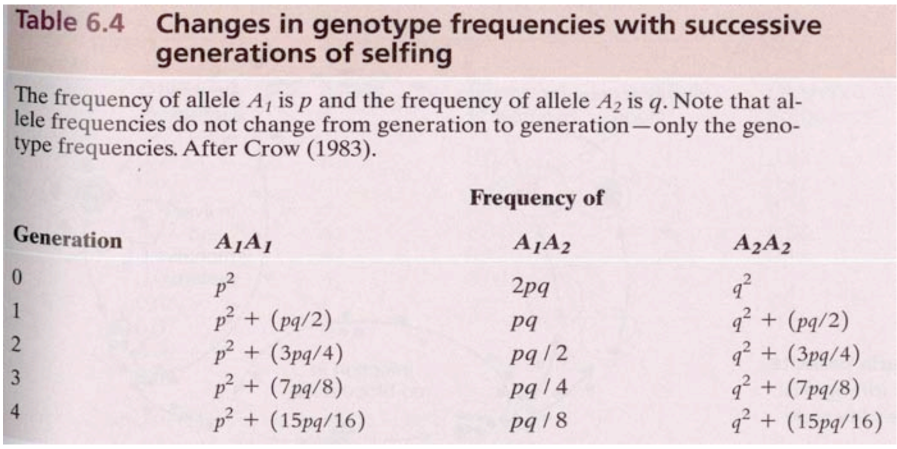
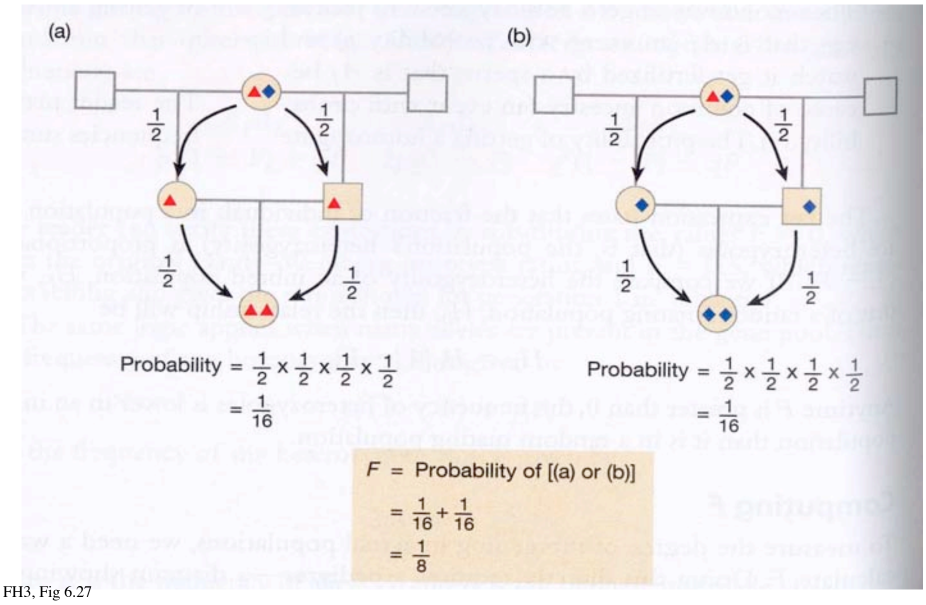


Table 6.4 Changes in genotype frequencies with successive generations of selfing

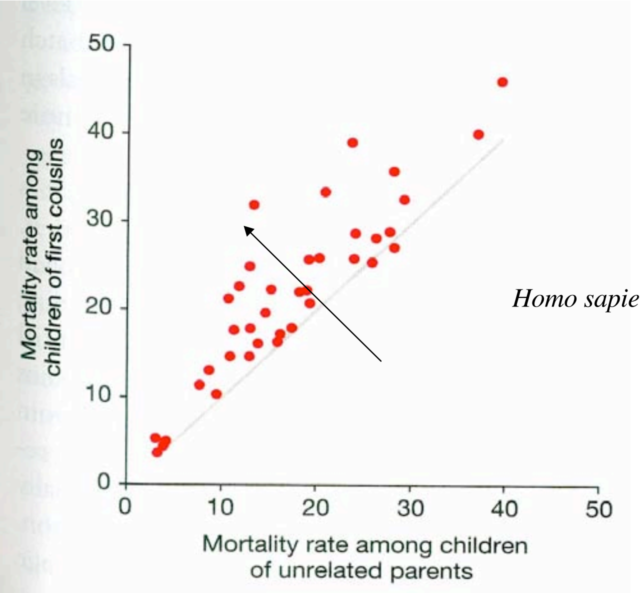
The frequency of allele A_1 is p and the frequency of allele A_2 is q. Note that allele frequencies do not change from generation to generation—only the genotype frequencies. After Crow (1983).

	Frequency of		
Generation	A_1A_1	A_1A_2	A_2A_2
0	p^2	$2pq$	q^2
1	$p^2 + (pq/2)$	pq	$q^2 + (pq/2)$
2	$p^2 + (3pq/4)$	$pq/2$	$q^2 + (3pq/4)$
3	$p^2 + (7pq/8)$	$pq/4$	$q^2 + (7pq/8)$
4	$p^2 + (15pq/16)$	$pq/8$	$q^2 + (15pq/16)$

FH3, Tab. 6.4

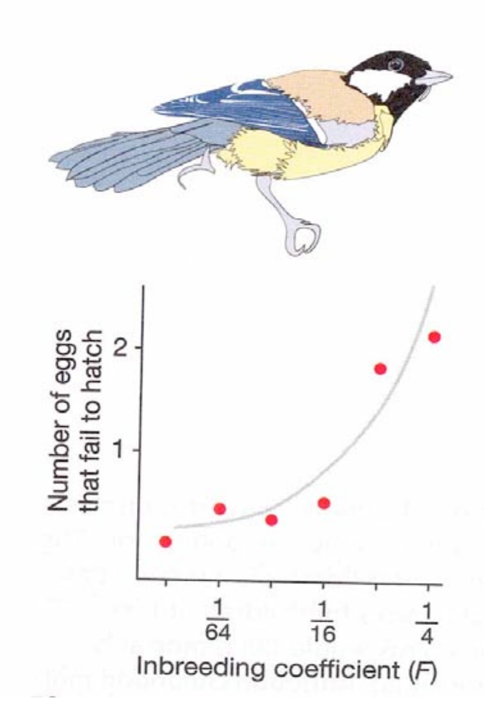

(a)
(b)
1/2
1/2
1/2
1/2
Probability = 1/2 x 1/2 x 1/2 x 1/2
= 1/16
Probability = 1/2 x 1/2 x 1/2 x 1/2
= 1/16
F = Probability of [(a) or (b)]
= 1/16 + 1/16
= 1/8

FH3, Fig 6.27



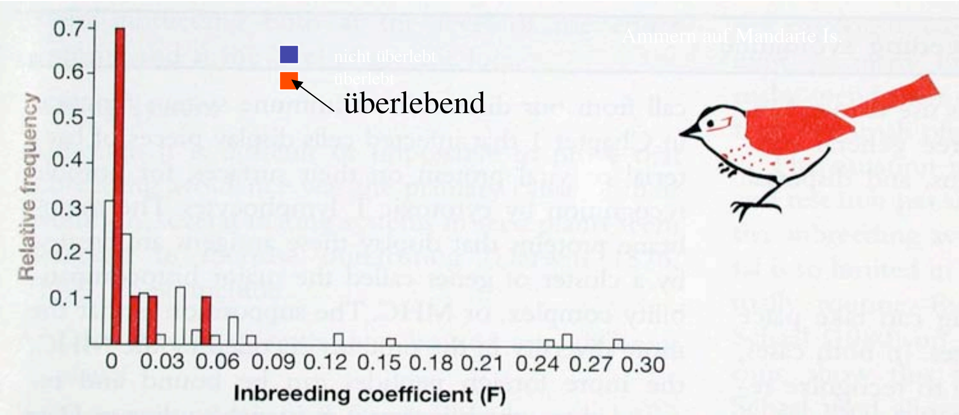
Homo sapiens

FH3, Fig 6.28



FH3, Fig 6.30

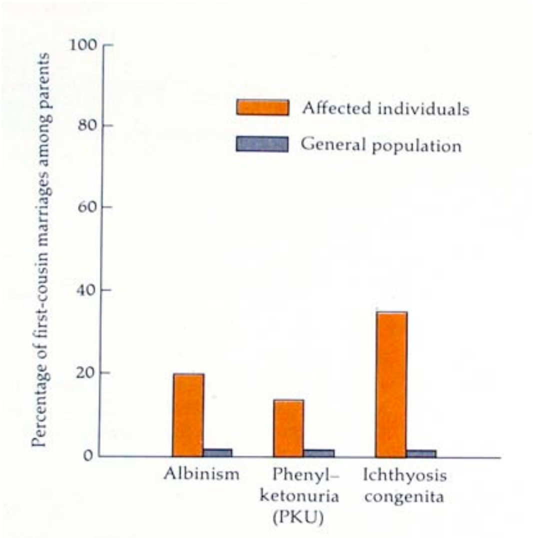
Ursprung u. Erhaltung genetischer Variation



überlebend

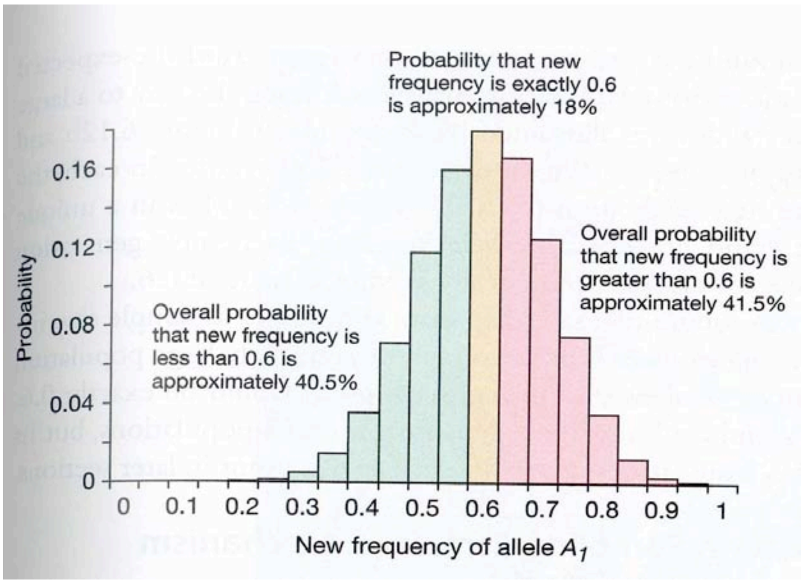
FH1, Fig .5.29

Anteil der Betroffenen (affected) aus Heiraten zwischen Cousins im Vergleich zur Gesamtbevölkerung

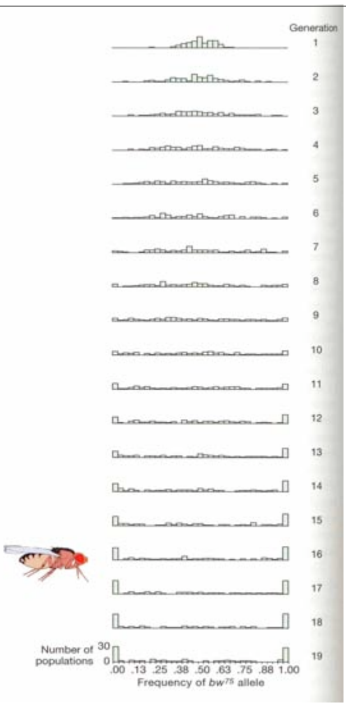


Ayala, F.J. (1982), Fig.5.6

Ursprung u. Erhaltung genetischer Variation

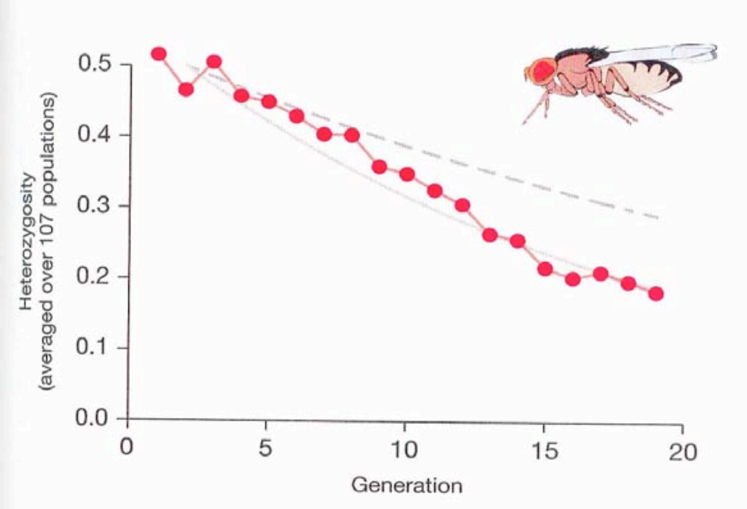


FH3, Fig 6.16

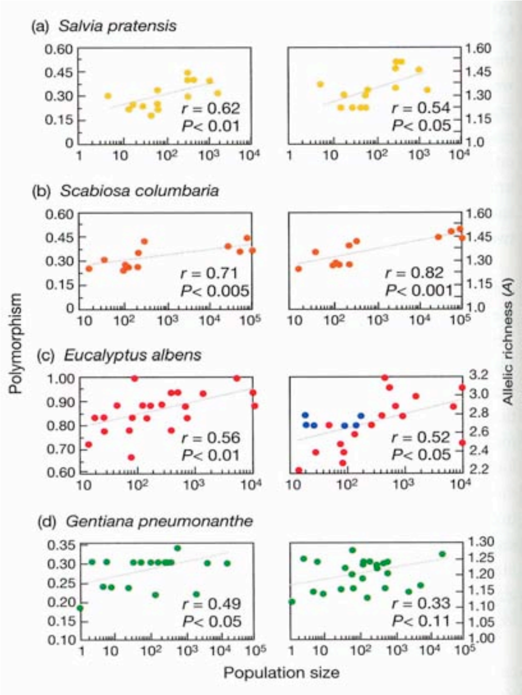


FH3, Fig 6.16

Ursprung u. Erhaltung genetischer Variation

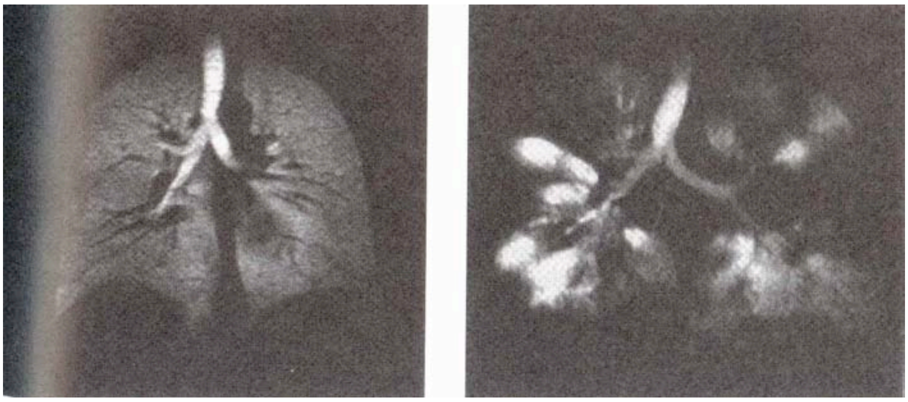


FH3, Fig 6.17



FH3, Fig 6.19

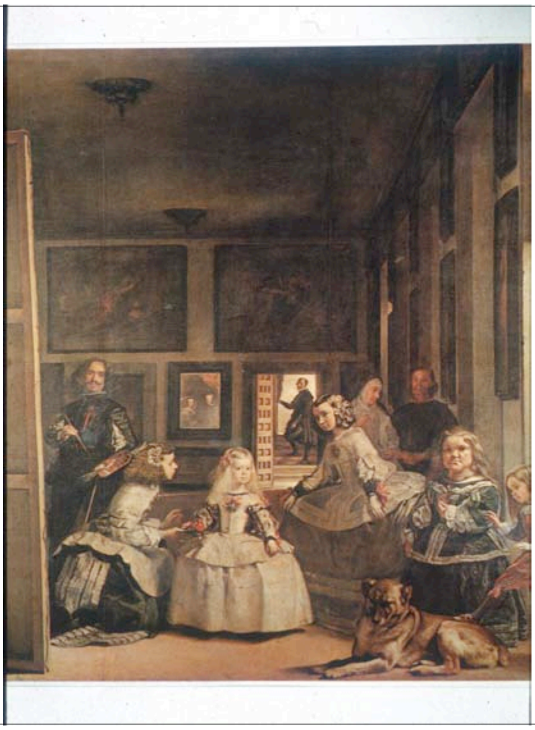
Zystische Fibrose



normal

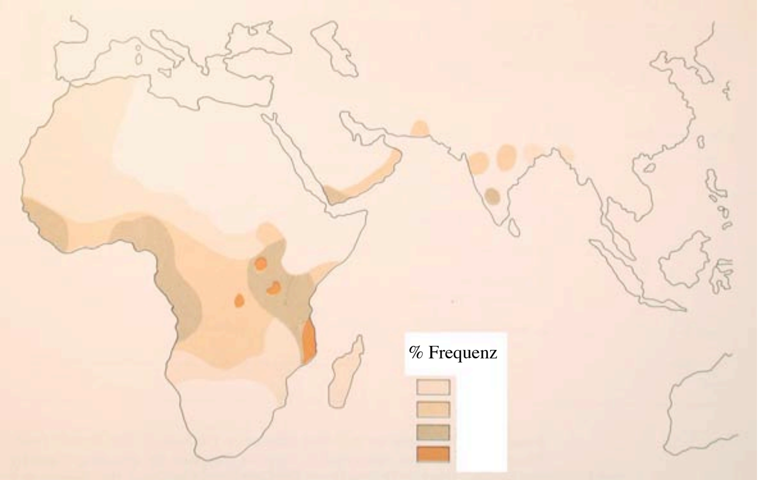
betroffen

FH3, Fig 5.26

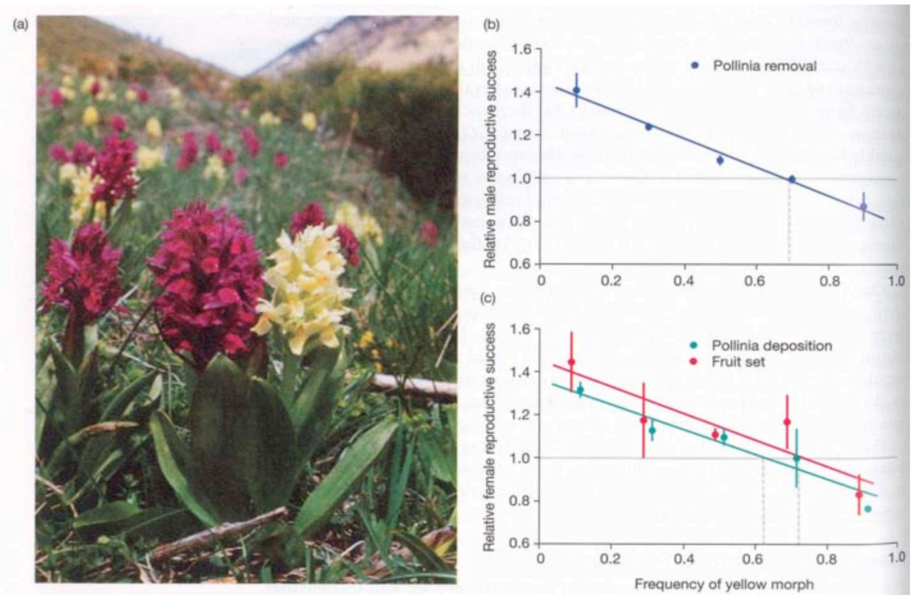


Diego Velazquez: Las Meniñas (1656)

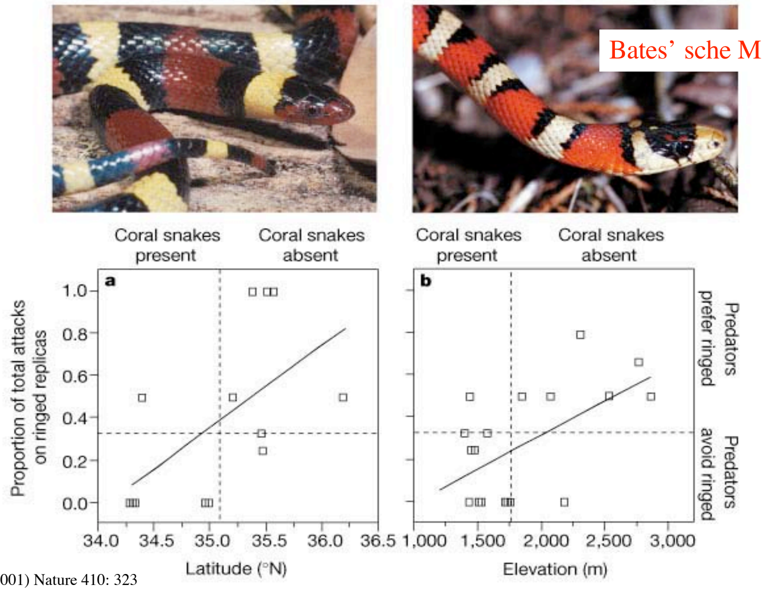
Malaria und Hämozyten



Ursprung u. Erhaltung genetischer Variation

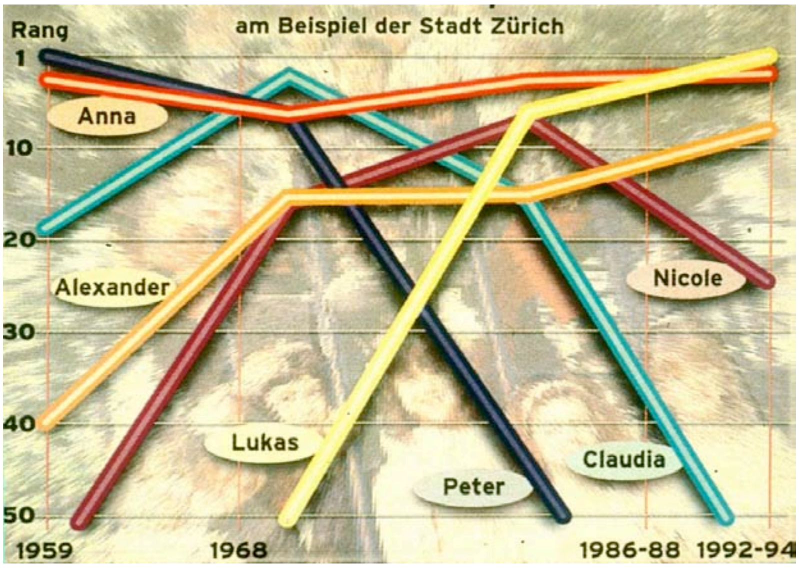


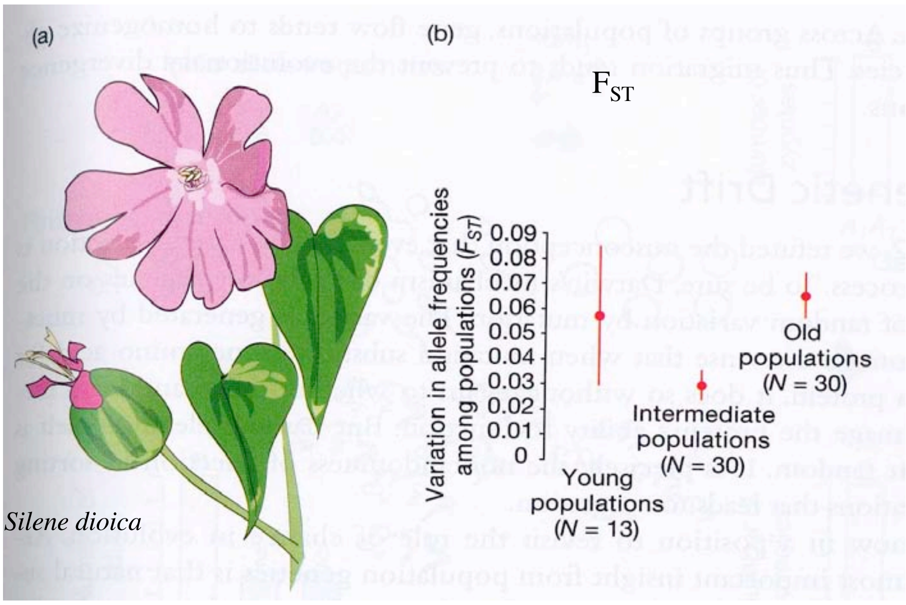
FH3, Fig 5.21



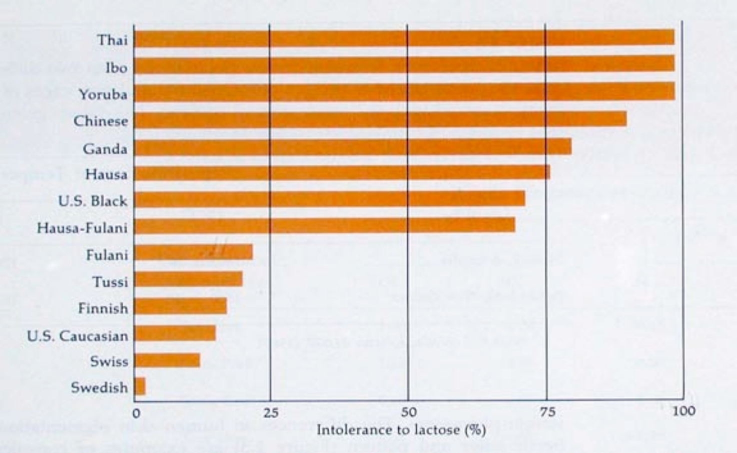
Bates' sche Mimikry

Pfennig et al. (2001) Nature 410: 323


am Beispiel der Stadt Zürich
Rang
1
10
20
30
40
50
1959
1968
1986-88
1992-94
Anna
Alexander
Nicole
Lukas
Peter
Claudia


(a)
(b)
F_{ST}
Variation in allele frequencies among populations (F_{ST})
0.09
0.08
0.07
0.06
0.05
0.04
0.03
0.02
0.01
0
Young populations (N = 13)
Intermediate populations (N = 30)
Old populations (N = 30)
Silene dioica

FH3, Fig 6.9

Ursprung u. Erhaltung genetischer Variation