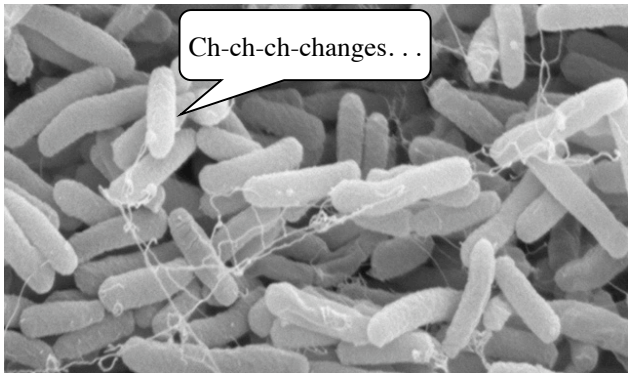
Modern Synthesis II

BIOL 4415: Evolution
Dr. Ben Waggoner

Can mutations ever be "good"?



Can mutations ever be "good"?

- Beginning in the 1970s, Barry Hall developed a strain of the bacterium *Escherichia coli* with its gene for the enzyme beta-galactosidase missing
  - These bacteria now could not use lactose as a food source
  - Hall then grew the bacteria on a lactose-containing medium, creating selection for bacteria that could use lactose as a food. . .
  - Not only did the bacteria recover the ability to break down lactose. . . they evolved two new control genes for it as well.

Can mutations ever be "good"?

- These *evolved beta-galactosidase* (ebg) genes didn't "just appear out of nowhere". . .
  - The *ebg* genes are mutated versions of genes elsewhere in the genome, used for other functions (exactly what is still uncertain)
  - The "wild" *ebg* enzyme has almost no ability to catalyze lactose—but a single point mutation (changing aspartic acid to asparagine at position 92) increases its affinity for lactose 47-fold
  - A second mutation was found (changing tryptophan to cysteine at position 977) that increased the *ebg* enzyme's activity 466-fold, and also enabled it to break down a new substrate, arabinose.

Richard Lenski has been running an evolutionary experiment on *E. coli* populations since 1988, encompassing 50,000 generations. (Check out the updates at http://myxo.css.msu.edu/ecoli/)

- A 2001 study of artificially induced mutations in *E. coli* showed that as many as 12% were beneficial when the bacteria were being grown on a new substrate, maltose. (*PNAS* vol. 98)
- In 2008, after over 33,000 generations, one population developed the ability to metabolize citrate aerobically, which wild-type *E. coli* can't do. (*PNAS* vol. 108)
  - This almost certainly results from multiple mutations, the first appearing around generation 27,000. Work is underway to characterize exactly what

Additional observations confirm this basic principle: mutations in existing genes can and do produce new and functional genes with new features

- A naturally occurring mutation in a Japanese population of *Flavobacterium* (K172), discovered in 1981, gave the bacteria the ability to digest byproducts of nylon—which didn't *exist* before 1937. . .
- In 1995, a different bacterium, *Pseudomonas aeruginosa*, was selected in the lab to break down the same nylon byproducts. Its enzymes are different from the *Flavobacterium* system.
- Antibiotic-resistant bacteria keep appearing in both lab experiments and "the wild"—including resistance to completely human-made antibiotics, such as fluoroquinulones.

Natural mutation rates

- The frequency of mutations varies between organisms, between genes in one organism, between different parts of the same gene, and even between different nucleotide positions. . .
  - Bacteria and viruses: typical observed mutation rates of 10^{-6} to 10^{-9} mutations per gene per generation
  - Lenski's *E. coli*: 8.9×10^{-11} per base pair per generation
    - Since *E. coli*'s genome is 4.6×10^{6} base pairs, this works out to one mutation every 2442 generations
  - Humans: Clinically significant mutations are observed in about 10^{-4} - 10^{-5} gametes per gene; similar numbers have been observed in corn

Natural mutation rates

- Cairns and colleagues (1988) showed, using *E. coli*, that stressful conditions (starvation by growth on a food source that the bacteria couldn't use) seemed to *cause* the "right" mutations to enable the bacteria to use the food source
- This is the *adaptive mutagenesis* hypothesis, and it sounds almost Lamarckian—mutations aren't "random", because a cell can produce those mutations that it needs
- New interpretation: Stress increases the overall mutation rate, but doesn't make "good" mutations more likely
  - Other sources of selection can also affect the mutation rate, partly by acting on the DNA repair genes themselves

Allele Frequency

- Calculated simply as the sum of all copies of a particular allele in a population, divided by the sum of all alleles in the population
  - When a gene has two alleles, the frequencies are usually indicated as *p* and *q*
- Modern Synthesis evolutionary theory starts by looking at whether and how allele frequencies change in a population

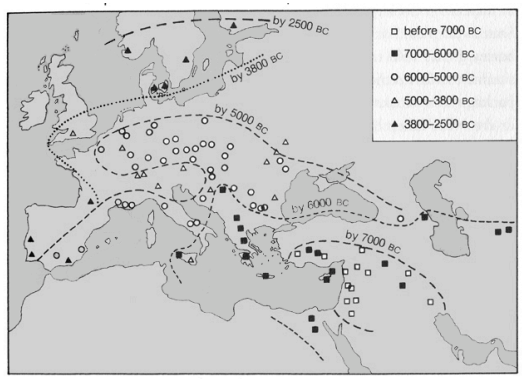
Allele Frequency

- Allele frequencies by themselves can sometimes be used as indicators of a population's past history
  - Human history can be studied by looking at a human population's allele frequencies
  - We can also look at the frequencies of alleles that only appear in one copy per person—such as mitochondrial genes or Y-chromosome genes. These are known as *haplotypes*.

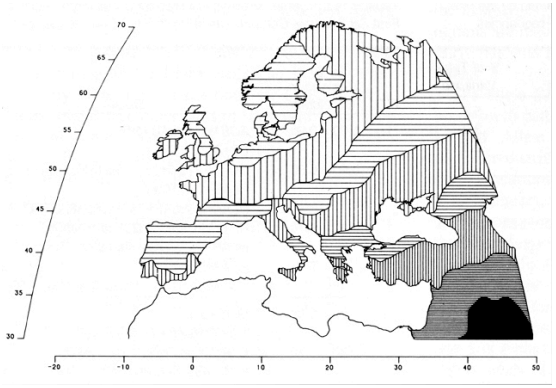
Allele Frequency

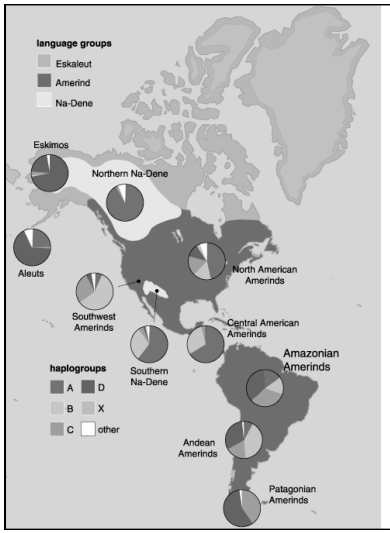
- EXAMPLE: A protein expressed on red blood cells called *Duffy* exists in two alleles, which we can call Fy+ and Fy-.
- In European populations, Fy+ is virtually the only allele of the Duffy protein; its frequency is nearly 100%
- In African populations, Fy- is virtually the only allele of the Duffy protein.
- Frequency of these alleles in African-American populations: about 30% and 70%

Archaeology shows that agriculture—and presumably farmers—appeared in the Middle East in about 8000 BC, and slowly spread into Europe.



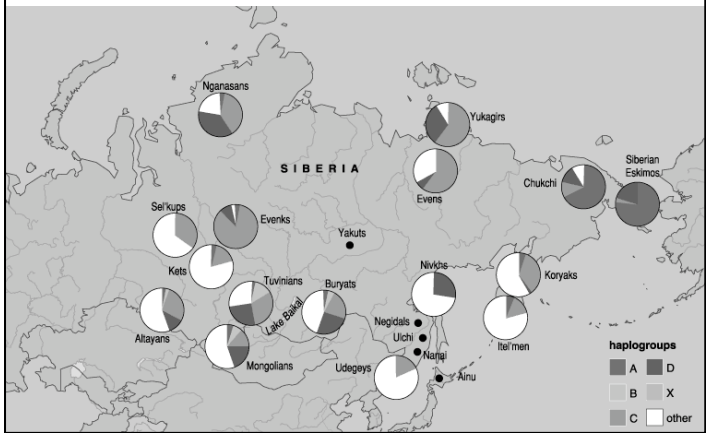
The combined frequencies of many alleles show a *cline* (a continuous trend) from the Middle East into Europe—which is what you'd expect from an ancient migration of people.

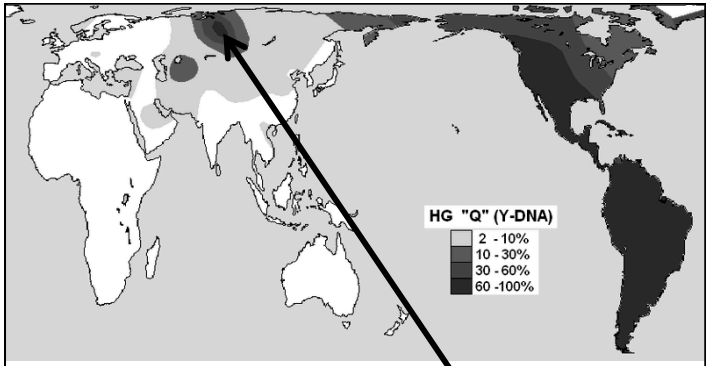




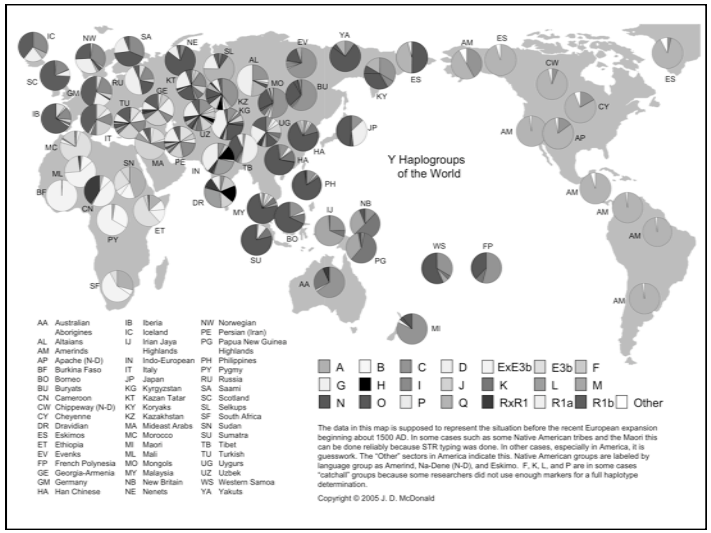
Different Native American groups show different frequencies of mitochondrial haplotypes, but the great majority are haplotypes known as A, B, C, and D.

The A, B, C, and D haplotypes also happen to be common among the native populations of northern Asia and Siberia.





A Y-chromosome haplotype called Q is by far the most common among Native Americans. It's also abundant among two native peoples of Siberia: the Kets and the Sel'kups. The next slide shows this in more detail. . .


Y Haplogroups
of the World
AA Australian Aboriginies
AL Altaians
AM Amerinds
AP Apache (N-D)
BF Burkina Faso
BO Borneo
BU Buryats
CN Cameroon
CW Chippeway (N-D)
CY Cheyenne
DR Dravidian
ES Eskimos
ET Ethiopia
EV Evenks
FP French Polynesia
GE Georgia-Armenia
GM Germany
HA Han Chinese
IB Iberia
IC Iceland
IJ Irian Jaya Highlands
IN Indo-European
IT Italy
JP Japan
KG Kyrgyzstan
KT Kazan Tatar
KY Koryaks
KZ Kazakhstan
MA Mideast Arabs
MC Morocco
MI Maori
ML Mali
MO Mongols
MY Malaysia
NB New Britain
NE Nenets
NW Norwegian
PE Persian (Iran)
PG Papua New Guinea Highlands
PH Philippines
PY Pygmy
RU Russia
SA Saami
SC Scotland
SL Selkups
SF South Africa
SN Sudan
SU Sumatra
TB Tibet
TU Turkish
UG Uygurs
UZ Uzbek
WS Western Samoa
YA Yakuts
A B C D ExE3b E3b F
G H I J K L M
N O P Q RxR1 R1a R1b Other
The data in this map is supposed to represent the situation before the recent European expansion beginning about 1500 AD. In some cases such as some Native American tribes and the Maori this can be done reliably because STR typing was done. In other cases, especially in America, it is guesswork. The "Other" sectors in America indicate this. Native American groups are labeled by language group as Amerind, Na-Dene (N-D), and Eskimo. F, K, L, and P are in some cases "catchall" groups because some researchers did not use enough markers for a full haplotype determination.
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