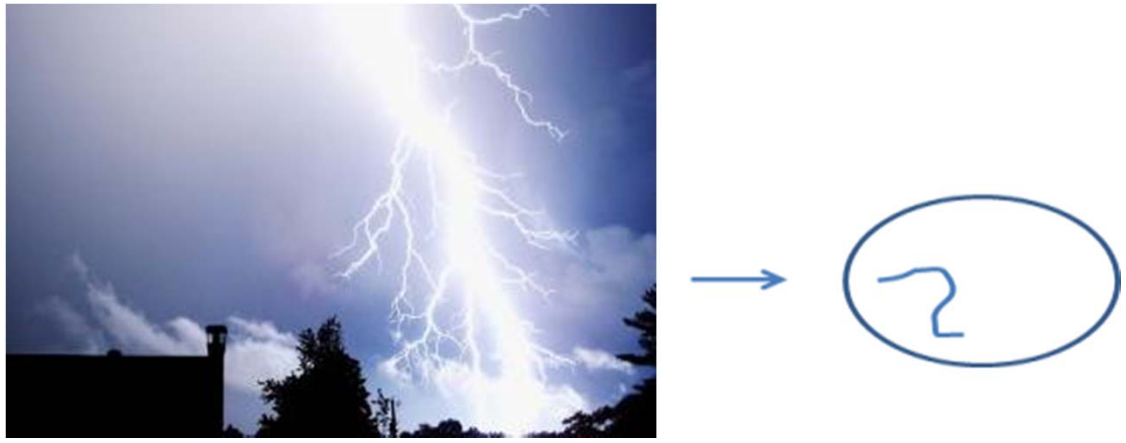
Mutations as the origin of genetic variation

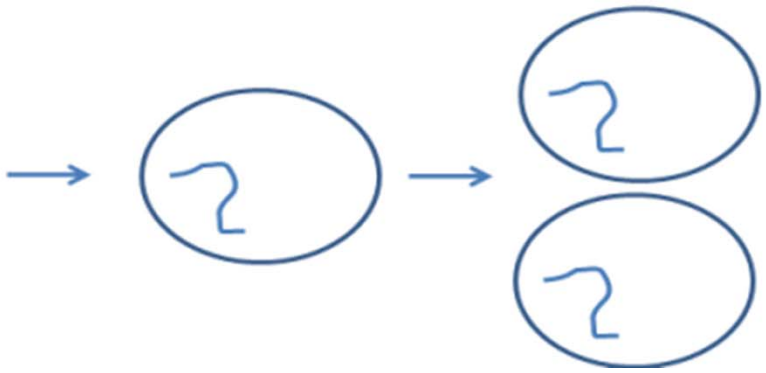
Life starts….

- Likely single haploid genome in single primitive cell
- What would define it as being “alive”? How would we decide it was “alive” rather than “inanimate”?

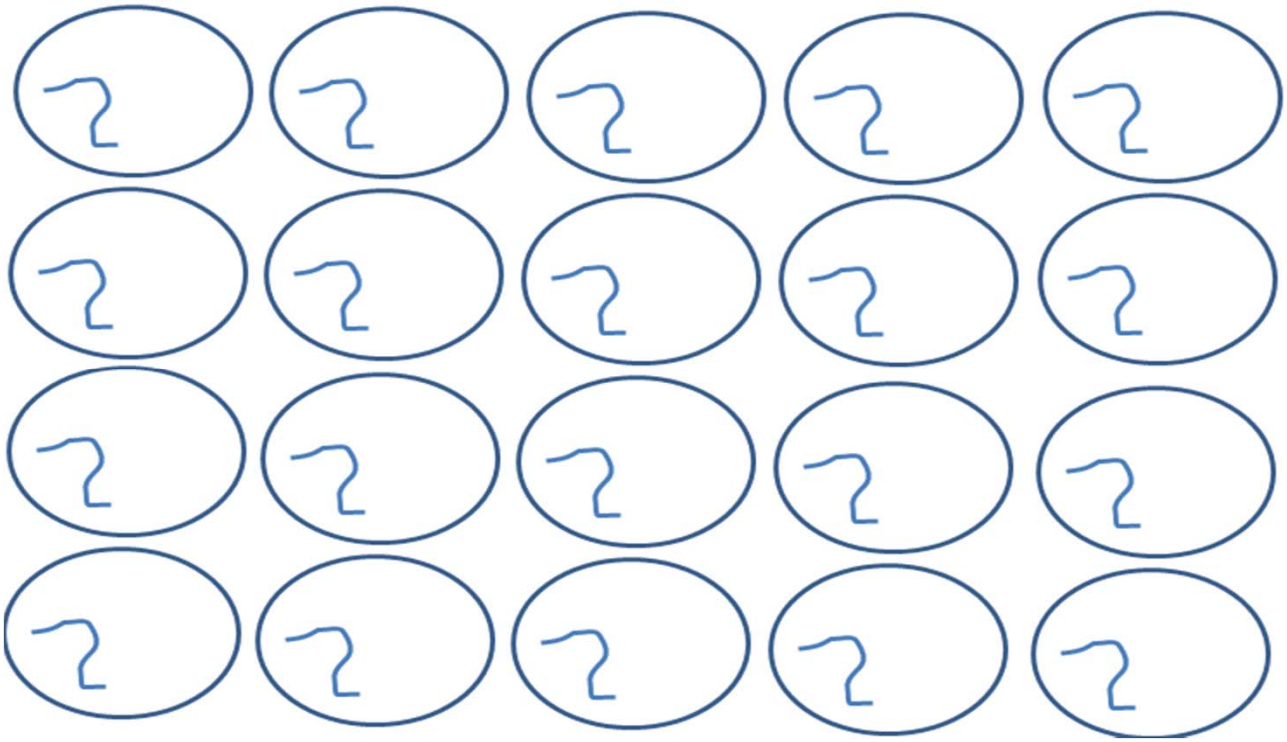


Life starts….

- Likely single haploid genome in single primitive cell
- Reproduces!
- What it can make is dictated by its genes
  - What if that genetic code never changes?



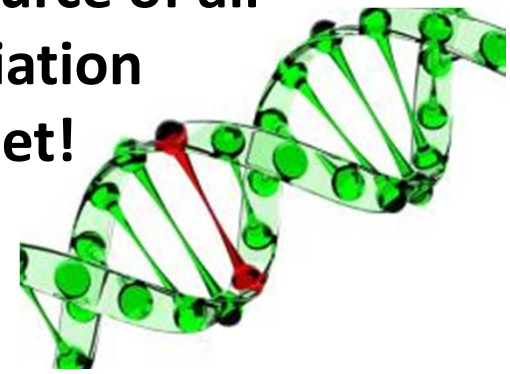
Life, 3.5 billion years later…



Where does variation come from?

(Theme for lecture)

- What is the source of those "changes in the code"?
  - "A change to the DNA sequence"
  - **<u>Mutations</u> are the ultimate source of all genetic variation on the planet!**



What is a mutation???

- A “mutant” is what you call your brother when he’s being annoying…

What is a mutation???

- A “mutant” in the comic world is “an individual who possesses a genetic trait called an X-gene that allows them to naturally develop Superhuman Powers and Abilities…”

What is a mutation???

- A “mutant” is what you might call someone who has a disfiguration, like extra fingers…

What is a mutation???

- There's a little bit of truth to all of those
  - (less-so for the X-Men version)
- Again, ultimate source of all variation
  - So, "MUTATIONS HAPPEN"
- One of most common types:
  - Error in replication/ meiosis leading to a change in a base
  - Mom's cells are homozygous for: **ATGCCAGCCTGA**
  - Her egg that makes the kid is: **ATGCCGGCCTGA**

What *isn't* a mutation???

- Mutations don't happen preferentially "when we need them"
  - Random when they happen relative to "need"

- Many mutations are "bad":
  - Why?

- Some mutations "don't matter" AAA -> AAG
- Rarely (though it happens), a mutation is "good"

After mutation, the new variant exists in the population

Alleles in gene pool at one gene

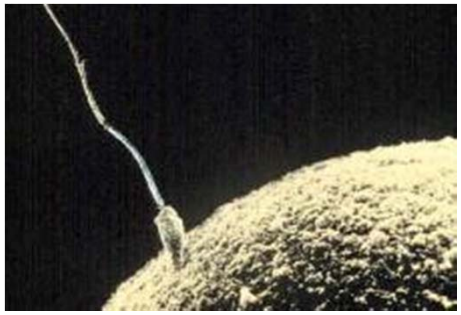
Generation 1		Generation 2
ATGCCAGCCTGA	**Birth of a SNP!**	ATGCCGGCCTGA
ATGCCAGCCTGA		ATGCCAGCCTGA
ATGCCAGCCTGA		ATGCCAGCCTGA
ATGCCAGCCTGA		ATGCCAGCCTGA
ATGCCAGCCTGA	**Meiotic or mitotic?**	ATGCCAGCCTGA
ATGCCAGCCTGA		ATGCCAGCCTGA

1 allele at base 6

2 alleles, 1 abundant

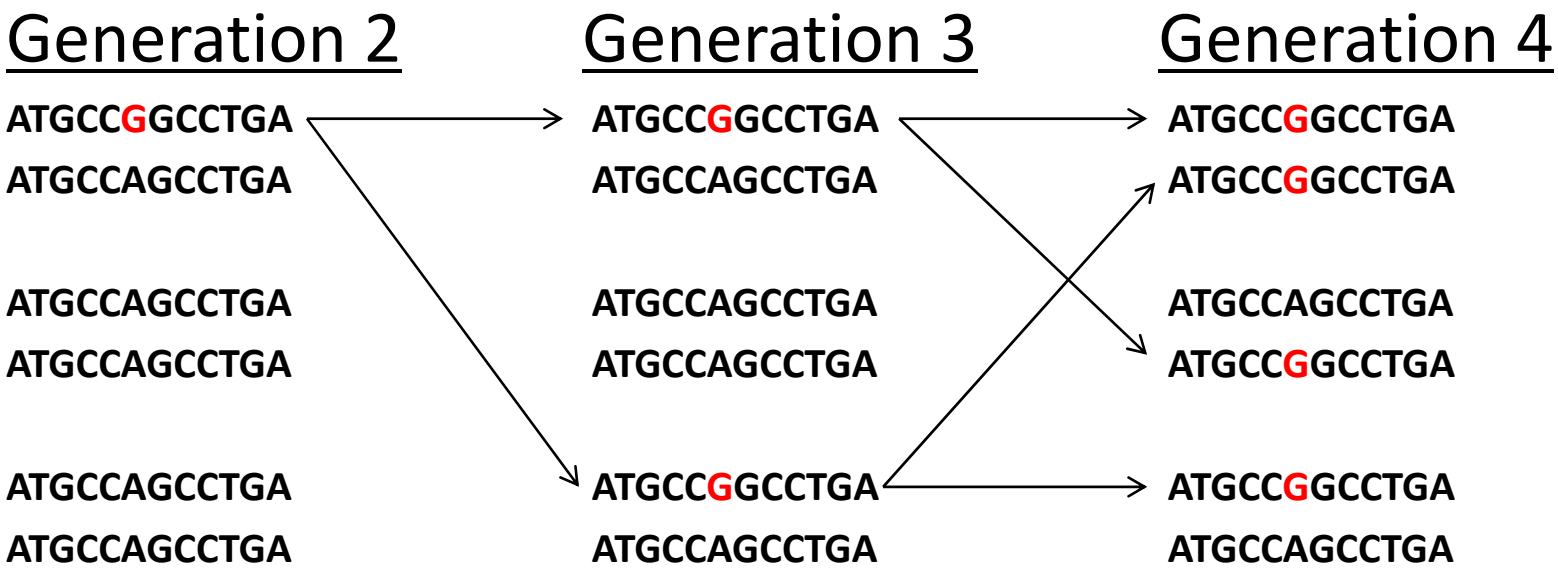
What happens then?

- … if **ATGCCGGCCTGA** causes death of the embryo?
- … if **ATGCCGGCCTGA** causes loss of fitness?
- … if **ATGCCGGCCTGA** causes no change in fitness?
- … if **ATGCCGGCCTGA** causes super-fertility?

After mutation, the new variant *may spread* in the population

Alleles in gene pool at one gene



Over time, mutation generates a lot of variation in natural populations

- A lot of mutations don't have an effect on fertility or life span
  - *May* still affect a phenotype
  - LOTS of genetic variation in human height

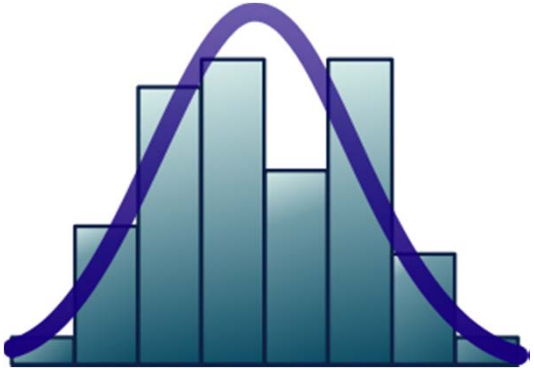


Fig 9-1 in Freeman & Herron

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