



**Mutation “rates”:  
How often do mutations happen?**



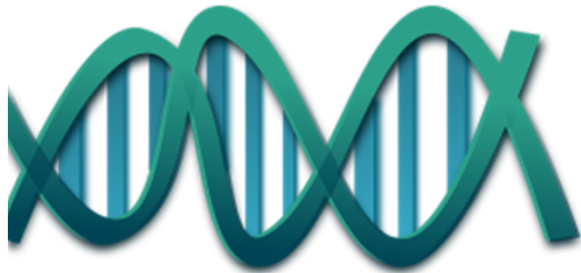
# **All the genetic variation started as new mutations...**

- How often do new mutations arise?
- How often do “bad” new mutations arise?



# Mutation “rate” per generation

- Depends on type of mutation
  - Specific base to specific base (A → G)
  - Specific base to “any different” base (A → nonA)
  - Other types (inversions, translocations, deletions)
  - Anything in a gene that yields a phenotype



# Mutation “rate” per generation

- Many use base to “any other” base
  - Measured by
    - Get sequence of whole genome after known number of generations (and taking average)
    - Comparing sequences from existing species and estimating number of generations
  - $2.1 \times 10^{-8}$  mutations per base per generation in worms
  - Human genome size is  $3.1 \times 10^9$  bases



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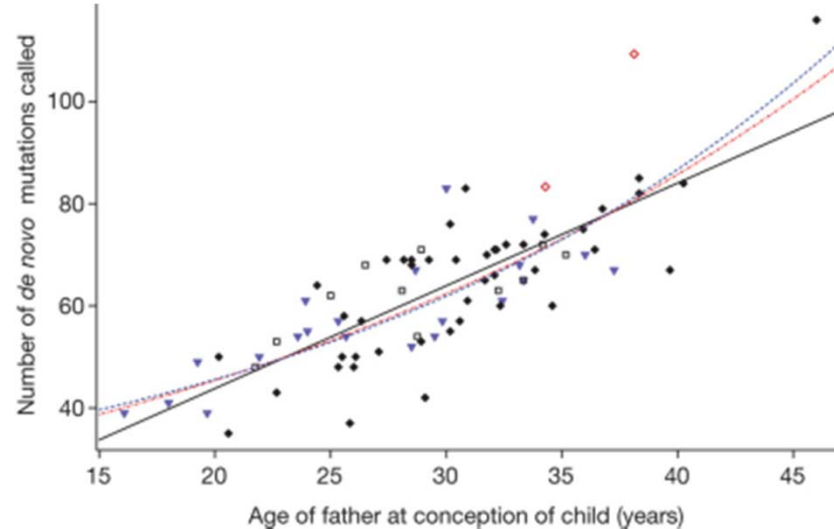
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65 new mutations!!!

## ... confirmed!

- 2012 study in Iceland found, on average, 63 new mutations
- **STRONG** effect of father's age



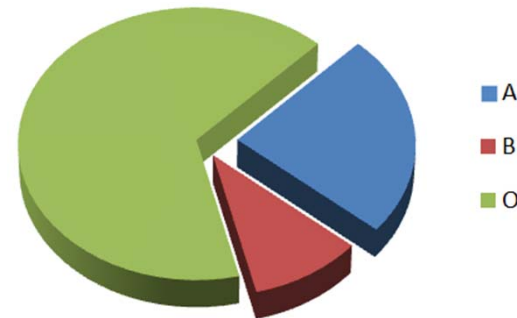
# So, your brother is a mutant!

- A “mutant” is what you call your brother when he’s being annoying...
- ... but so are you...
- **Mutations introduce A LOT of variation into populations**



# Once mutations arise...

- New mutations may spread or be lost (via natural selection or other forces)
- While present, new variations will have a “frequency” in the population:
  - Allele frequencies of blood types:
    - 24% A; 10% B; 66% O in USA
  - Genotype frequencies of blood types:
    - 6% AA; 32% AO; 5% AB; 44% OO; etc.





**but again, all the genetic variation  
started as new mutations...**

- How often do new mutations arise? VERY!
- How often do “bad” new mutations arise?



# “Deleterious” mutation rate

- If chromosomal anomalies, may have immediate effect on offspring
  - Often result in miscarriage
- A lot of other bad mutations are either more subtle, or are masked because recessive
  - Current estimates, ~1-2 per generation
  - Sorry... you’ re probably a “bad” mutant, too...



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