

9 March 2022

deleterious  
recessive  
alleles

MENDELIAN genetics

why do deleterious alleles  
persist?

- tradeoff ( $G \times E$  interaction)
- linkage to an advantageous allele

↳ \* heterozygote advantage

$\{1, 1, s\}$

$\{1, 1+s, s\}$

MUTATION -  
SELECTION

DRIFT

↳ bottleneck  
founder effect

# TAY - SACHS disease

$1/300$  in US population

$1/30$  E European (Ashkenazi)  
French Canadians

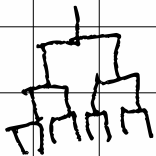
→ DRIFT

↳ tb resistance? intelligence?

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- pop frequency
- pop distribution  
geographic  
pop history
- mechanism / where expressed?
- PHYLOGENETIC info

↳ age of allele



- COALESCENT

↳ shape of the tree

Population history

selective sweep

# phenylketonuria (PKU)

strongly deleterious if  
you have phenylalanine  
in your diet

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- many different mutations
- alleles are OLD
  - Irish gene pool ~ 2500 BC.
  - pop size 100K - 200K
  - 0.6% 2x from drift

→ heterozyg adv  $\approx$  1.5%

phenotypic effects.

higher birth weight  
nicotinic resistance?  
starvation resistance?