Human Polymorphisms

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Malaria

• Anthropologist’s “fave” diseases: Malaria & sickle cell anemia
• How does one contract malaria?
• Bite of a mosquito. Will any mosquito do?
• Mosquitoes of the Genus Anopheles
• Mosquito is actually a vector (carrier) of something that makes us sick.

What?
Malaria

- Protozoan, specifically *Plasmodium falciparum*
- Where is this endemic (widespread problem)?
- Tropical regions: Central & W. Africa, India, S.E. Asia, C. America, Northern America, Pac.

[Map of Malaria around the world]
Malaria

• How bad is the problem? (#3 killer in world)
  • [Link](http://www.msgpp.org/malaria.shtml)
  • [Link](http://www.sciencedaily.com/releases/2009/02/090203090708.htm)
  • [Link](http://ecoworldly.com/2008/09/18/potential-cure-for-malaria-discovered-in-rainforests-of-costa-rica/)

• What can we do? Currently no actual cure avail., prophylactic medicine is (prophylaxis?)
  • Preventive med.
  • Prophylaxis anecdote
Mosquitoes & Breeding

• Anopheles mosquito breeding requirements
• Similar to mosquitoes in general?
• Sunwarmed, stagnant (still) water
Sickle Cell Anemia (SCA)

• A genetically inherited disease: affects the shape of hemoglobin protein
• What does it mean to be anemic?
• Shortage of red blood cells (rbc’s)
• SCA is chronic & specific to hemoglobin: a type of protein that binds oxygen in rbc’s
• Originated as a single mutation on 1 amino acid

result of point on 1
Genetics behind SCA

- AA: 100%
- AS: 60%
- 40%
- SS: 100%

Mosaicism: neither of these alleles is truly dominant
Heterozygote advantage

- Thus a heterozygote does not have the disease but is has “sickle cell trait” (carrier)
- [http://www.mayoclinic.com/print/sickle-cell-anemia/DS00324/DSECTION=all&METHOD=print](http://www.mayoclinic.com/print/sickle-cell-anemia/DS00324/DSECTION=all&METHOD=print)
- [http://sickle.bwh.harvard.edu/sickle_trait.html](http://sickle.bwh.harvard.edu/sickle_trait.html)
- If this allele has such lethal potential, why didn’t natural selection “weed it out”?
- heterozygotes are relatively immune to malaria
Mechanisms for SCA heterozygote advantage

- Lower incidence of infection, lower frequency of transmission
- Infected cells slow process if sickled & lysed
- Sickled cells cause damage to protozoan
- Infected rbc’s “leak” K rapidly, protozoan requires ample K to reproduce
Ecology of SCA & malaria

- ~7000 yrs ago, pt. mutation gives rise to sickle allele
- No reproduct. Advantage conferred, allele is maintained @ very low levels
- ~5000 yrs ago, ppl in forested areas of W. & C. Africa transition to agriculture (requires?)
- Cutting down trees
- This inadvertently encourages?
Ecology of SCA cont’d

• ↑ mosq. breeding, meaning ↑ in malaria rates
• S allele still present @ low levels, individuals w/ S allele are spared malaria, creating?
• Reproductive advantage, natural selection operates on S allele, making it more abundant in subsequent generations
• Selection continues until a balanced polymorphism is attained
**Balanced Polymorphism**

- Balanced polymorphism: $AS \times AS$

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- In a malarial environment, this process will continue with each generation, bringing about stable allele frequencies, meaning that nearly all adults will be heterozygous.
Transient polymorphism

- In a non-malarial region: AS x AS

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Transient polymorphism: loss of one allele over time
(it’s level is changing, albeit, extremely slowly)

Environment:
http://ipsnews.net/news.asp?idnews=42976
Rate of natural selection on alleles

- Deleterious dominants & recessives exist, however, deleterious dominants are weeded out rather quickly.
- Ex: Achondroplastic dwarfism, inhibits growth in long bones of an individual, coded for by a mutated dominant allele: A, a
  - AA = death
  - Aa = a dwarfism
  - aa = "normal" (unaffected)
Late onset deleterious dominant

- Huntington’s chorea: H, h
  Genotypes:
- HH: death
- Hh: Huntington’s chorea
- hh: unaffected
- Doesn’t set in until an individ. is in their 40s or 50s
- thus natural selection cannot operate on this trait, why?
- Post reproductive yrs. Remember, natural selection operates through reproduction!
- Other late onset diseases?
Deleterious recessive

- Degenerative nerve disorder, Tay-Sach’s disease: T, t
  - TT: unaffected
  - Tt: unaffected, carrier
  - tt: Tay-Sach’s disease
- Heterozygote advantage exists in this case as well
- Check out “Curse & Blessing of the Ghetto” article for details
Summary/Myths

• Traits do not emerge due to heterozyg. advantage, but advantage may make a trait more common
• SCA is not a racial, nor an African disease
• Natural selection cannot operate quickly/efficiently upon recessive alleles since they are hidden
• Natural selection cannot operate upon late onset diseases since they do not impact reproduction
• N.S. solutions are far from ideal, but rather “messy” solutions