Human Polymorphisms
cont’d & Forces of evolution

Professor Janaki Natalie Parikh
jnparikh@verizon.net
Lactose intolerance

- Words ending in “ose”, meaning?
- Sugar, lactose= milk sugar
- lactose $\xrightarrow{\text{lactase}}$ glucose & galactose
- Lactase is an enzyme (type of protein), proteins are coded for where?
- DNA: genetic component to lactose intolerance
  LL: lactose tolerant
  LI: mildly lactose tolerant
  II: severely lactose intolerant (rare combo, explain)
Lactose intolerance

• If it’s rare, why are so many ppl lactose intolerant?
• 2 reasons. 1st: oft. misdiagnosed, many are not lactose intolerant, but simply allergic to milk or additives in our dairy
• http://paskamansettfarms.com/
• 2nd reason: in mammals, regulatory gene turns off production of lactase post toddlerhood
• likely selected for due to need to ensure mom’s reproductive fitness
While a ♀ is lactating, another process does not take place, which ↓ her evolutionary fitness (process?)

Ovulation, thus decreasing her overall fertility (word of caution, not foolproof!)

If the regulatory gene switches off prod. Of lactase, this encourages mom to cease nursing, known as?

Weaning. Lactose intolerance post childhood is the default (normal) condition for all mammals
• In humans, a mutation occurred on the regulatory gene (creating a junk sequence)
• This may have initially provided a dietary advantage
• This is why (human) adults in some populations can continue to drink milk as adults, however, it is by far the exception & not the norm in mammals
• Cultural solution to the prob. of delayed Lactose intolerance (dairy food item w/o lactose?)
• Yogurt!
Forces of Evolution

- 1. Natural selection: review, mechanism?
- Reproduction. More specific, sexual selection: mating preferences, anything that makes ya more...
- Sexy!
- http://www.youtube.com/watch?v=5Fjh5Ss5bZw&feature=related
2. Gene flow: the movement of genetic information (alleles) from 1 population to another

As humans groups have migrated throughout the world, so have our genes, in other words...

Face it, when you get folks together, they make whoopie!

3. Gene drift: 2 types:

3a. Intergenerational & 3b. founder effect

General: random loss of genetic information (an allele) in a population, requires a small population
Gene Drift: Intergenerational

- Intergenerational g. drift: sample pop. Of 10 ppl
- For any 1 gene, how many alleles are in the pop.?
- 20: ea. Person has 2 alleles
- 9 ppl homozyg Blood type A, 1 person blood type AB
- How many copies of A allele?
- 9 ppl x 2 = 18, + 1 from AB person = 19 total
- If AB person is ♀ she mates w/ 1 of the ♂s
- Her mate’s blood type would be?
- AA, the only option
Gene Drift

- Punnett square of their mating:
- AB x AA:

<table>
<thead>
<tr>
<th></th>
<th>A</th>
<th>B</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>AA</td>
<td>AB</td>
</tr>
<tr>
<td>A</td>
<td>AA</td>
<td>AB</td>
</tr>
</tbody>
</table>

- Interpret: If they have 2 kids, is it guaranteed that one will inherit AB? No, & if neither does, then what happens to the B allele in the next generation?
- Lost from the population. Why?
- Chance occurrence
Gene Drift: Founder Effect

- Founder Effect: g. drift that occurs when a small group breaks off from the larger group
- Founding Grp won’t take a representative sample
- Native Amer. didn’t bring over B blood type allele
Forces of Evolution

• Yet, B blood type is present in Nat. Americans today, albeit @ somewhat lwr levels, how?
• Gene flow reintroducing the allele

• 4. genetic mutation: source of new variation, random occurrences, however, other forces of evolution (natural selection) can operate on the trait
• http://www.sciencedaily.com/releases/2008/01/080130170343.htm
Hardy Weinberg Model

- Hardy Weinberg Model of Genetic Equilibrium
  1. population is large (infinitely)
  2. equal # of
  3. all individuals mate (no celibacy please!)
  4. all matings are random (woohoo!)
  5. all matings occur within the population
  6. all matings produce same # of offspring
  7. no forces of evolution @ work

- What good is it?
- Degree & direction of changes, estab. the fact that evolution is always occurring!
GATTACA

- Genoism, de jure & de facto discrimination,
- What is the standard method of conception in GATTACA?
- Specific examples of genoism
- 3 main social classes in GATTACA & basis
- Message: relationship between gene & effect, not clear cut, a major factor to be considered?
- Environment! Individual choices! Determination!
- The science of GATTACA today:
  - [http://online.wsj.com/article/SB123439771603075099.html](http://online.wsj.com/article/SB123439771603075099.html)