

Homework April 2, 2015 (to be returned on April 9)

1) Consider the 64 codons of the standard genetic code. Each codon can undergo nine single-site mutations as each nucleotide can mutate to three other bases. The total number of mutations is then 576. How many of these mutations are silent, i.e. result in no change of the encoded amino acid (or stop codon)?

2) If individual I has an autosomal recessive disease (it appears only in homozygous for a particular mutation) and both parents are unaffected, what is the chance that the sibling of I is a heterozygous carrier? What is the chance that the sibling is also affected by the disease?

3) Phenylketonuria is an autosomal recessive form of severe mental retardation. About one in 10,000 newborn Caucasians are affected. Assuming random mating, what is the frequency of heterozygous carriers?

4) Suppose that the recessive disease in question 2) is not autosomal but linked to a locus on the sexual chromosome X . Will the disease be more common among males or females (assuming random mating)? How does the ratio of occurrence between males and females depends on the frequency q of the allele?