Video Tutorial 16.1: The Hardy-Weinberg equilibrium

With microarrays and other genomic tools, it is often feasible to directly determine or impute the frequency of a given haplotype among a population for a particular region of the genome or a particular allele. If we have the haplotype frequency, we can ask if the population meets at least one of the criteria for being at Hardy Weinberg equilibrium—that is, is the population mating at random for the trait in question?

Here is a specific example for a human population. The data are reported in an article by Kenny et al. in *Science* but the focus of the article is on the particular gene and not specifically on the population genetics, so we can ask a question that the authors did not address directly. Several more questions about this population are taken up in Study Question 16-19 at the end of the chapter.

The Solomon Islands are in the Pacific Ocean, about 1800 km north-east of Australia. You can see their location on the map here.

One of the most remarkable genetic features of the native peoples of the Solomon Islands is that, although the people are very dark-skinned, between 5-10% of them have blonde hair. Here are some examples. This is by far the highest frequency of blonde hair in any population other than those native to northern Europe.

The study found that the blonde hair was the result of a recessive allele in the TYRP1 gene, and that this particular allele is unique to the Solomon Islands. Polymorphisms in this gene have not been found in other human populations. Using a microarray analysis, the frequency of the haplotype and thus the allele was found to be 0.26.

Does this trait meet the definition for mating at random with respect to this trait?

Let's work it out. . The allele frequency q = 0.26. The allele is recessive, so the homozygotes would have blonde hair. If the population is at equilibrium, the expected frequency of blonde hair is q^2, or 0.26^2 or 6.8%. The expected frequency is very similar to the observed frequency so the population does appear that there is no preferential mating for or against blonde hair. If there were, the genotype frequencies would not agree with what is expected from the haplotype frequencies—that is, there would be either more or fewer people with blonde hair (that is, homozygous for the recessive allele) than would this.

What about the other assumptions of a Hardy-Weinberg equilibrium? Is there evidence for or against any of the other assumptions?

There is no suggestion that this is a recurrent mutation in the TYRP1 gene. The paper reports that the blonde hair people have the same molecular change in the gene, and share a haplotype, which is consistent with them being descended from a common ancestor.

There is no evidence for this allele elsewhere in the world, suggesting that the mutation arose in an ancestor and that, probably due to the remote location of the Solomon Islands, was limited to this population. There has been very little migration to the Islands.

© P Meneely, R Dawes Hoang, I N Okeke, K Heston 2017

The population has been small, so this is an ideal situation to observe the Founders Effect. It is now more than 600,000 people but even two generations ago, it is likely to have been much less than 100,000 people and a few centuries ago when this change might have arisen in the population, it was much smaller than that.

The study does not tell us if there are subpopulations—that is, if the haplotype is more common in some populations than others but 60% of the people now live in villages of less than 200 people, and there are hundreds of islands so there may be many regional differences in the frequency of this and other traits. There are no data that suggest that blonde have had more or fewer children over the past several centuries—that is, we don't know if natural selection is at work here. So this seems very likely to be a simple, but visually striking example of a Founders effect, and that there is random mating about the population at large.

© P Meneely, R Dawes Hoang, I N Okeke, K Heston 2017