

Modeling Human Genetics – The Hardy-Weinberg Equilibrium

William Clemson, Dan Crawford, Rafia Darwish, Rebecca Miesle, Melinda Schall

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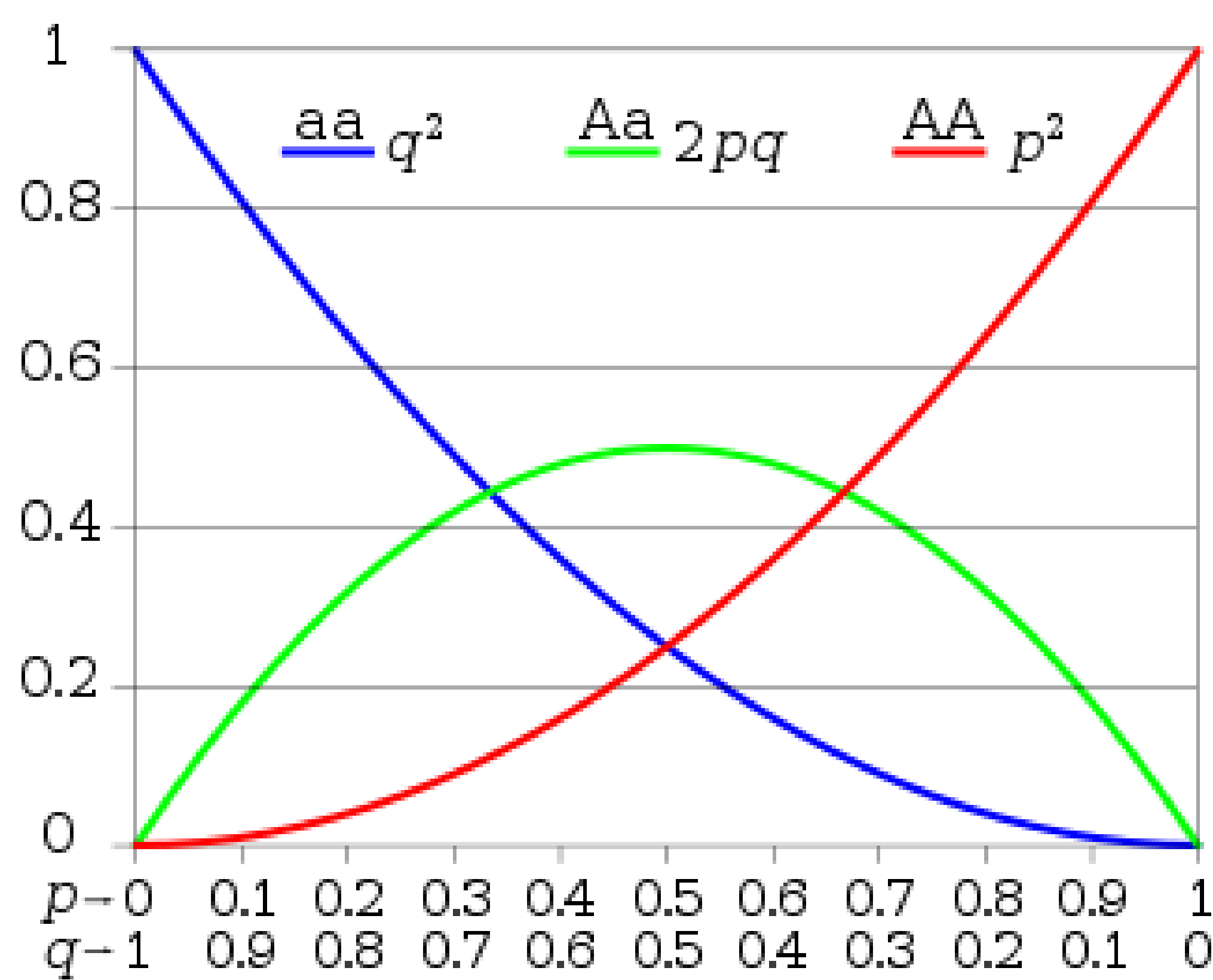
Introduction

Hardy-Weinberg Equilibrium is a null model used to find allele frequencies from phenotype frequencies.

We can collect information from a gene pool and use it to find hypothetical genotype frequencies.

Genotype frequencies might change through subsequent generations, but they will eventually return to Hardy-Weinberg equilibrium values.

The equation used is $p^2 + 2pq + q^2 = 1$, where p represents the frequency of dominant allele and q represents the frequency of recessive allele. By using the known frequency of the recessive phenotype, the genotype frequencies can be determined within the sample population.



In the above graph, the horizontal axis shows respective values of p and q , and the vertical axis shows the frequency for a specific genotype to occur under those conditions.

Application of Hardy-Weinberg

217 participants were surveyed for presence/absence of three traits: widow's peak, tongue rolling, and colorblindness. Parental data was collected (when possible) to use for further statistical tests. Traits were assumed to be controlled by one locus and to follow simple Mendelian rules for inheritance:

- Presence of widow's peak and tongue rolling – assumed autosomal dominant
- Colorblindness – assumed sex-linked recessive

Hardy-Weinberg for Autosomal Traits

Using Widow's peak (autosomal dominant trait),

- With widow's peak: 61 (29.469% of population)
- Without widow's peak: 146 (70.531% of population)
- $p^2 + 2pq + q^2 = 1$, $p + q = 1$
- $q^2 = .70531$
- $q = .8396$
- $p = 1 - q = .1603$
- With these values, the genotype frequencies can then be calculated:
- $p^2 = .0257$, $2pq = .2691$, $q^2 = .7053$

Thus, within this population, 2.57% are homozygous dominant, 26.91% are heterozygous dominant, and 70.53% are homozygous recessive for a widow's peak.

The same methods were used to find frequencies for tongue rolling: 26.88% were homozygous dominant, 49.93% were heterozygous dominant, and 23.18% were homozygous recessive.

Hardy-Weinberg for Sex-Linked Traits

The same methods as used for autosomal traits, but the phenotype frequencies of males are used to find q .

In this population, 92.52% of the males were dominant, and 7.47% of males were recessive for colorblindness. Since males only have one X chromosome, this allele is the only one that is expressed.

For females, 85.60% were homozygous dominant and 13.82% were heterozygous dominant. No females were homozygous recessive for this trait, but a larger sampling size might have provided data to match this model.

Assumptions

Certain assumptions must be made for a population in order to use the Hardy-Weinberg equation. If any of these assumptions (aside from random mating) are not held true, genotype frequencies will fluctuate between successive generations.

Mating must be random. Inbreeding decreases heterozygosity and increases homozygosity, which influences the distribution of the alleles. Also, small populations are more likely to exhibit genetic drift (random changes in frequencies between successive generations).

No migration from or immigration into the population is acceptable. No mutations may occur within the genome. Natural selection may not happen, as that leads to specific genes being preferred over others.

Experimental Error

There is potential for subjectivity if participants were uncertain about various traits, especially regarding traits of their parents.

The sample size was small. However, if the population was made to be too large, the Hardy-Weinberg model could not be used.

Limitations of the Hardy-Weinberg

It is a less accurate way to find genotype frequencies than counting alleles or using pedigrees. These are not used since this data is generally not available.

Individuals are just one in a huge population.

Hardy-Weinberg is a null model; these assumptions are regularly broken, so populations never stay in Hardy-Weinberg equilibrium - frequencies will change from one generation to the next.

It cannot account for genetic complexities, such as:

- Reduced penetrance, which occurs when a seemingly dominant trait is not always expressed in offspring generations
- The control of traits by multiple loci
- Environmental influence on phenotype

Observations

Even though it is a dominant trait, the presence of a widow's peak was less common than the absence of a widow's peak.

It was very common in those surveyed that if a trait was present, both parents also had it. Likewise, if a trait was absent, most often neither parents had it.

As anticipated, colorblindness was the rarest trait within the population surveyed. Furthermore, only males or fathers had this trait, which can easily be explained by the fact that it is a sex-linked trait in which males only receive one allele.

In some of the surveys collected, contradictions to what would be normally expected were present in the data. For example, a dominant trait would be present for the individual surveyed but absent in the parents. This can probably be explained by human error, but it could also be a result of some of the complications to simple genetics as previously describes.

Collected Data

| | Present | Absent | Mother with trait | Father with trait | Both parents with trait | Neither parent with trait |
|------------------------------|------------|------------|-------------------|-------------------|-------------------------|---------------------------|
| Widow's peak - Men | 39 | 68 | 11 | 20 | 3 | 58 |
| Widow's peak - Women | 22 | 78 | 6 | 18 | 3 | 63 |
| Widow's peak - Both | 61 | 146 | 17 | 38 | 6 | 121 |
| Tongue rolling - Men | 79 | 28 | 17 | 6 | 27 | 17 |
| Tongue rolling - Women | 80 | 20 | 20 | 6 | 33 | 12 |
| Tongue rolling - Both | 159 | 48 | 37 | 12 | 60 | 29 |
| Colorblindness - Men | 8 | 99 | 1 | 6 | 0 | 91 |
| Colorblindness - Women | 0 | 100 | 0 | 3 | 0 | 93 |
| Colorblindness - Both | 8 | 199 | 1 | 9 | 0 | 184 |
| Total men surveyed | 107 | | | | | |
| Total women surveyed | 100 | | | | | |
| Total participants | 207 | | | | | |