

A population genetics online calculator with adjustable parameters for the estimation of the risk of inheriting Mendelian diseases

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ABSTRACT

The population genetics online calculator is a tool that allows users to estimate frequencies of alleles and genotypes in a given population of interest. It provides several options of input information, including dominant allele, recessive allele, homozygous dominant and homozygous recessive frequencies. Additionally, it also allows the input of autosomal recessive disease frequency or the proportion of the population that has the disease. As output, it shows all allele and genotype frequencies (including heterozygotes), as well as the option for the user to verify the probability that a couple will have the disease, given the input information. It can be accessed through the following website: <https://bioinfo.azurewebsites.net/calculator.html>.

KEYWORDS;- Population Genetics, Hardy Weinberg, Calculator

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I. INTRODUCTION

The Hardy-Weinberg Law, created by Wilhem Weinberg and Godfrey Harold Hardy, is a useful way to verify if natural selection or other evolutionary factors have an impact on a given population [1]. Their creation represented an important development in the field of Population Genetics, which aims to investigate the dynamics of genes in natural populations, searching to elucidate the mechanisms that alter their genetic composition. Although there are factors that possibly affect the equilibrium of the Hardy-Weinberg equation in human populations, it is still useful, as this population rapidly reaches a new genetic equilibrium whenever it is exposed to evolutionary factors [2, 3].

The validation of the Hardy-Weinberg equation for Mendelian disorders has a notable practical application. For instance, if we know the population frequency for individuals with a recessive trait in a given population, it is possible to use this equation in order to estimate allele frequencies. Therefore, we can also calculate the homozygous and heterozygous frequencies in the same population.

Here we constructed a website which can be used to calculate population frequencies given different options as input. It will save users from making mistakes when doing these calculations by hand.

II. METHODS

The calculator was built with Python 3.0 language, and its code is freely available at GitHub (<https://bit.ly/3qEiiIu>). It can be accessed through the following web page: <https://bioinfo.azurewebsites.net/calculator.html>.

Users can choose among the following parameters in order to start the calculation:

- Dominant allele frequency
- Recessive allele frequency
- Homozygous dominant frequency
- Homozygous recessive frequency
- Disease frequency in the population of interest
- Proportion of population that has the disease

Additionally, users can also specify if they would like to know the probability that a couple will have a child with the disease. In this case, information about the parents being affected or not by the disease can also be provided.

All calculations are based on the following equations:

$$P^2 + 2pq + q^2 = 1$$

$$p + q = 1$$

where:

p: dominant allele frequency

q: recessive allele frequency

p²: dominant homozygous genotype frequency

2pq: heterozygous genotype frequency

q²: recessive homozygous genotype frequency

III. RESULTS

Opening Page, where users select the information that they have:

Population Genetics Calculator
Frequency Calculations of Autosomal Recessive Disease using Hardy-Weinberg Equations

What information do you have?

- ✓ -- Please choose an option --
- Dominant allele frequency (A)
- Recessive allele frequency (a)
- Homozygous Dominant frequency (AA)
- Homozygous Recessive frequency (aa)
- Disease frequency in the population of interest
- Proportion of population that has the disease

Example output, showing allele and genotype frequencies:

What information do you have?

Disease frequency in the population of interest

What is the frequency of the disease in the population of interest (example: 6.4%)?

8.8

The dominant allele frequency (A) is 70.34%

The recessive allele frequency (a) is 29.66%

The homozygous dominant frequency (AA) is 49.47%

The homozygous recessive frequency (aa) is 8.80%

The heterozygous frequency (Aa) is 41.73%

In case the user wishes to view the probability that a couple will have a child with the disease, the following output will be shown:

Would you like to know the probability that a couple will have a child with this disease (yes/no)?
 Yes No

Is the mother or the father affected by the disease (yes/no)?
 Yes No

The probability that their child will have the disease is 4.35%.

In case one of the members of the couple is known to be a carrier of the recessive allele, the probability will be 10.43%.

In case both mother and father are known to be carriers of the recessive allele, the probability will be 25.00%

IV. CONCLUSION

We presented a population genetics calculator with a variety of input parameter options in order to estimate genotype and allele frequencies for a given population. Additionally, it can also estimate the inheritance risk of a Mendelian disease. This easy-to-use tool should be helpful for students and professionals who are interested in these data, but who do not have experience in making the calculations by hand.

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