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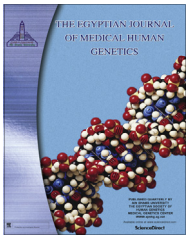

Estimation of allelic frequencies for ABO and Rh blood groups

Dear Editor

Estimation of the allelic frequencies for genetic markers is very important in genetic studies. Also investigation of the concordance between observed and expected value based on the Hardy–Weinberg equilibrium (HWE) is strongly recommended by STrengthening the REporting of Genetic Association studies (STREGA) [1]. Today, many investigators used the polymerase chain reaction (PCR) based methods for genotyping. In all of these methods, heterozygote genotypes may be distinguished from both homozygote genotypes. Although, the allelic frequencies may be easily estimated using the counting of the alleles, several mistakes were reported by investigators [2–4]. However, for some other genetic markers such as ABO and Rh blood groups in humans, we observed more problems in studies [5–7]. Here I am going to mention how we can estimate the allelic frequencies in these two polymorphic traits.

Consider the ABO locus with three alleles, 'O', 'A' and 'B'. The frequency of the alleles 'O', 'A' and 'B' will be designated by r, p and q, respectively. Hardy–Weinberg shows that the allelic and genotypic frequencies will remain stable from generation to generation, provided that there is no mutation, no migration, and no natural selection in a very large population with random mating. Based on the HWE the frequencies of the O, A, B, and AB phenotypes are r^2, $(p^2 + 2pr)$, $(q^2 + 2qr)$, and $2pq$, respectively.

We can estimate the allelic frequency using the following formulae:

$$r = \sqrt{(\text{frequency of O phenotype})}$$

$$p = \sqrt{(\text{frequency of A} + \text{O phenotypes})} - r$$

$$q = \sqrt{(\text{frequencies of B} + \text{O phenotypes})} - r$$

Considering that we do not use the AB phenotype for estimation of the frequencies of the O, A, and B alleles, we saved one degree of freedom. It is self-evident that summation of the allelic frequencies should be 1. However, it happens if the observed frequencies of the phenotypes did not show any deviation from the expected values based on HWE. Several sets of factors (such as random sampling error, non-random evolutionary forces and technical mistakes) are involved for occurrence some deviations from expected HWE. At this time we should correct our estimations. If

$$d = 1 - (p + q + r)$$

then the allelic frequencies should be corrected by:

$$p_c = p(1 + d/2)$$

$$q_c = q(1 + d/2)$$

$$R_c = 1 - (p_c + q_c)$$

Now the expected frequencies for A, B, O, and AB phenotypes should be estimated using the corrected allelic frequencies:

Expected numbers for A phenotype: $p_c^2 + 2p_c r_c$
Expected numbers for B phenotype: $q_c^2 + 2q_c r_c$
Expected numbers for O phenotype: r_c^2
Expected numbers for AB phenotype: $2p_c q_c$

Using χ^2 test we can compare the observed and expected numbers of the ABO phenotypes.

$$\chi^2 = \sum(\text{observed value} - \text{expected value})^2/\text{expected value}, \quad \text{df} = 4 - 3 = 1.$$

It should be mentioned that in articles we can report p_c, q_c, and r_c as p, q, and r, respectively.

Considering that in Rh blood groups, the d allele acts as a recessive allele compared with the D allele, we observed only two phenotypes (DD + Dd) and dd which are named Rh^+ and Rh^-, respectively. Assuming that the D and d alleles have p and q frequencies respectively, under HWE condition, the frequency of Rh^- is equal to q^2. The allele frequencies are now:

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Table 1 Estimation of allelic frequencies of ABO and Rh blood groups in three ethnic groups at Silte Zone, Ethiopia.

	Sodo population	Silte population	Meskan population
ABO blood group			
Allelic frequencies			
A	0.2213	0.1902	0.1529
B	0.1837	0.1572	0.1368
O	0.5950	0.6526	0.7103
Observed values			
A	47	42	35
B	38	34	31
O	54	63	73
AB	8	8	8
Expected values			
A	45.9	41.8	35.4
B	37.1	33.8	31.3
O	52.0	62.6	74.2
AB	12.0	8.8	6.1
χ^2	1.45	0.005	0.61
P-value	>0.05	>0.05	>0.05
Rh blood groups			
Rh^+	134	137	135
Rh^-	13	10	12
Allelic frequencies			
D	0.7026	0.7392	0.7143
d	0.2974	0.2608	0.2857

$q = \sqrt{\text{frequency of } Rh^-}$

and

$p = 1 - q$

However, we cannot investigate the deviation from HWE, because we have no degree of freedom.

I read the article of Tesfaye et al. [5] with great interest. Based on the data presented in Tables 1 and 2 of the above mentioned article [5], the allelic frequencies (means the corrected frequencies), and deviations of the observed values from the expected values based on HWE for ABO phenotypes are presented in Table 1. This table also shows the allelic frequency of Rh blood groups.

Conflict of interest

The author declares no conflict of interest. There is no financial and personal relationship with other people or organizations that could inappropriately influence this work.

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