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ON THE RELATIONS FOR LIMITING CASE OF SELECTION WITH EQUILIBRIUM AND MUTATION OF DIPLOID MODEL

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Abstract. Assume that at a certain locus there are three genotypes and that for every one progeny produced by an I^AI^A homozygote, the heterozygote I^AI^B produces. Choi find the adapted partial equations for the model of selection against heterozygotes and in case that the allele frequency changes after one generation of selection when there is overdominance. Also he find the partial differential equation of general type of selection at diploid model and it also shall apply to actual examples. This is a very meaningful result in that it can be applied in any model ([1], [2]).

In this paper, we start with the limiting case of selection against recessive alleles. For the time being, assume that the trajectories of p_t and q_t at time t can be approximated by paths which are continuous and therefore we have a diffusion process. We shall find the relations for time t, p_t and q_t and apply to equilibrium state and mutation.

1. Introduction

Assume that at a certain locus there are three genotypes and that for every one progeny produced by an I^AI^A homozygote, the heterozygote I^AI^B produces. To calculate a genotypic frequency, we add up the number of individuals possessing a genotype and divide by the total number of individuals in the sample N. The gene pool of a population can be represented in terms of allelic frequencies. Allelic frequencies can be calculated from the numbers or the frequencies of the genotypes. To calculate the allelic frequency from the numbers of genotypes, we count the number of copies of a particular allele present among the genotypes and divide by the total number of all alleles in the sample. ([5])

For a locus with only two alleles, the frequencies of the alleles are usually represented by p and q, respectively. The selection coefficien is the relative intensity of selection against a genotype([4]). We usually note of selection for a special genotype. When selection is for one genotype, selection is automatically against at least one other genotype.
The related variable is the fitness. The fitness is defined as the relative reproductive success of a genotype in case of natural selection. The natural selection takes place

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when individuals with adaptive traits produce a greater numbers of offspring than that produced by others in the population. If the adaptive traits have a genetic basis, they are inherited by the offspring and appear with greater frequency in the next generation ([4], [5]). The fitness is the reproductive success of one genotype compared with the reproductive success of other genotypes in the population.

Suppose that the allelic frequencies of a population do not change and the genotypic frequencies will not change after one generation in the proportion p^2 (the frequency of I^AI^A), $2pq$ (the frequency of I^AI^B) and q^2 (the frequency of I^BI^B). Here p is the frequency of allele I^A and q is the frequency of allele I^B. When genotype are in the expected proportions of $p^2,\ 2pq,\ q^2$, the population is said to be in Hardy-Weinburg equilibrium([4], [5]).

Choi defined the density and operator for the value of the frequency of one gene and found adapted equations as a follow-up for the frequency of alleles and applied this adapted equations to several diploid model and it also applied to actual examples ([1], [2]). He found the partial differential equation of general type of selection at diploid model and it also apply to actual examples. This is a very meaningful result in that it can be applied in any model ([1]).

In this paper, we start with the limiting case of selection against recessive alleles. For the time being, assume that the trajectories of p_t and q_t at time t can be approximated by paths which are continuous. We shall find the relation for time t, p_t and q_t and apply to equilibrium state and mutation.

2. The Main Results

We begin with the probability about the frequency of allele;

Lemma 1. Assume that the selection coefficients for each genotype I^AI^A, I^AI^B and I^BI^B are $\alpha_1,\ \alpha_2,\ \alpha_3$ respectively. The mean change for allele I^A is

$$pq\frac{p(\alpha_1-\alpha_2)+q(\alpha_2-\alpha_3)}{\hat{\alpha}},$$

and the mean change for allele I^B is

$$pq\frac{p(\alpha_2-\alpha_1)+q(\alpha_3-\alpha_2)}{\hat{\alpha}}.$$

Here $\hat{\alpha}=p^2(1-\alpha_1)+2pq(1-\alpha_2)+q^2(1-\alpha_3)$.

Proof. See Choi ([1]). □

Consider the limiting case of selection against recessive alleles. Recessive lethals occur when the recessive homozygotes have selection coefficient $\alpha=1$. We represent the frequency of I^A and I^B in the intial and following generation by $p_0, p_2, \cdots, p_t$ and $q_0, q_1 \cdots, q_t$, respectively. For the time being, assume that the trajectories of p_t and q_t can be approximated by paths which are continuous. Therefore we have a diffusion process.

Theorem 2. Assuming that recessive lethal occurs, we have following equation

$$t=\frac{p_0-p_t}{(1-p_0)(p_t-1)}=\frac{1}{q_t}-\frac{1}{q_0}.$$

Proof. Since $\alpha = 1$, the frequency of allele I^A after one generation of selection is

$$\frac{p_0^2 + p_0 q_0}{1 - \alpha q_0^2} = \frac{p_0}{1 - \alpha q_0^2} = \frac{p_0}{1 - q_0^2} = \frac{1}{2 - p_0}$$

and the mean change in allele frequency for I^A is

$$\frac{\alpha p_0 q_0^2}{1 - \alpha q_0^2} = \frac{p_0 q_0^2}{1 - q_0^2} = \frac{q_0^2}{1 + q_0}$$

Since

$$p_1 = \frac{1}{2 - p_0}, \; p_2 = \frac{1}{2 - p_1}.$$

We substitute p_1 in the equation for p_2. Then we have

$$p_2 = \frac{1}{2 - p_1} = \frac{1}{2 - \frac{1}{2 - p_0}} = \frac{2 - p_0}{3 - 2p_0}.$$

After t generation, we have

$$p_t = \frac{t - (t-1)p_0}{(t+1) - tp_0}.$$

We can find the number of generation t using the following formular;

$$t(1 - p_0)(p_t - 1) = p_0 - p_t, \; t = \frac{p_0 - p_t}{(1 - p_0)(p_t - 1)}.$$

On the other hand, the frequency of allele I^B after one generation of selection is

$$\frac{q_0^2 - \alpha q_0}{1 - \alpha q_0^2} = \frac{q_0 - q_0^2}{1 - q_0^2} = \frac{q_0}{1 + q_0}$$

and the mean change in allele frequency for I^B is

$$\frac{-\alpha p_0 q_0^2}{1 - \alpha q_0^2} = \frac{-p_0 q_0^2}{1 - q_0^2} = \frac{-q_0^2}{1 + q_0}.$$

Since

$$q_1 = \frac{q_0}{1 + q_0}, \; q_2 = \frac{q_1}{1 + q_1} = \frac{\frac{q_0}{1+q_0}}{1 + \frac{q_0}{1+q_0}} = \frac{q_0}{1 + 2q_0},$$

we have

$$q_t = \frac{q_0}{1 + tq_0}.$$

Therefore

$$q_t(1 + tq_0) = q_0, \; t = \frac{1}{q_t} - \frac{1}{q_0},$$

and we have equation

$$\frac{p_0 - p_t}{(1 - p_0)(p_t - 1)} = \frac{1}{q_t} - \frac{1}{q_0}.$$

□

Remark. Equations $p_t - p_0 = q_0 - q_t$ that we commonly know can be derived using the Theorem 1, and this fact tells us that the Theorem 1 is a reasonable result.

We define that equilibrium occurs when there is no net change in gene frequency. Then we have ;

Theorem 3. Assume that the allele frequency changes after one generation of selection with the selection coefficient $\alpha = 1$ of heterozygote when there is overdominance and equilibrium occurs. The equilibrium frequency of the I^A allele is

$$p_0 = \frac{t}{1+t}$$

and the equilibrium frequency of the I^B allele is

$$q_0 = \frac{1}{1+t}.$$

Proof. The frequency of the I^A allele after one generation of selection is

$$p_1 = \frac{p_0 q_0}{1 - p_0^2 - tq_0^2} = \frac{p_0 q_0}{1 - p_0^2 - tq_0^2}$$

and the mean change is

$$\frac{p_0 q_0}{1 - p_0^2 - tq_0^2} - p_0 = \frac{p_0 q_0 (tq_0 - p_0)}{1 - p_0^2 - tq_0^2}.$$

Therefore the equilibrium frequency for the I^A allele is

$$p_0 = \frac{t}{1+t}$$

Similarly, the mean change is

$$\frac{p_0 q_0 + q_0^2(1-t)}{1 - p_0^2 - tq_0^2} - q_0 = \frac{p_0 q_0 (p_0 - tq_0)}{1 - p_0^2 - tq_0^2}$$

for frequence of alleles I^B. Therefore the equilibrium frequency for the I^B allele is

$$q_0 = \frac{1}{1+t}.$$

□

We can apply equilibrium to general cases.

Corollary 4. Assume that the selection coefficients for each genotype $I^A I^A, I^A I^B$ and $I^B I^B$ are $\alpha_1,\ \alpha_2,\ \alpha_3$ respectively. The equilibrium occurs when $\alpha_1 = \alpha_2 = \alpha_3$.

Proof. From Lemma 1, the mean change for allele I^A is

$$\begin{aligned}\frac{p_0^2(1-\alpha_1) + p_0 q_0 (1-\alpha_2)}{\hat{\alpha}} - p_0 &= \frac{p_0 q_0 (1-\alpha_2)(q_0 - p_0) + p_0^2 q_0 (1-\alpha_1) - p_0 q_0^2 (1-\alpha_3)}{\hat{\alpha}} \\ &= p_0 q_0 \frac{p_0(\alpha_1 - \alpha_2) + q_0(\alpha_2 - \alpha_3)}{\hat{\alpha}}\end{aligned}$$

and the mean change for allele I^B is

$$\begin{aligned}\frac{p_0 q_0 (1-\alpha_2) + q_0^2(1-\alpha_3)}{\hat{\alpha}} - q_0 &= \frac{p_0 q_0 (1-\alpha_2)(p_0 - q_0) + p_0 q_0^2 (1-\alpha_3) - p_0^2 q_0 (1-\alpha_1)}{\hat{\alpha}} \\ &= p_0 q_0 \frac{p_0(\alpha_2 - \alpha_1) + q_0(\alpha_3 - \alpha_2)}{\hat{\alpha}}.\end{aligned}$$

Here

$$\hat{\alpha} = p_0^2(1-\alpha_1) + 2p_0 q_0 (1-\alpha_2) + q_0^2(1-\alpha_3).$$

In order for allele I^A to remain in equilibrium, we have $\alpha_1 = \alpha_2$ and $\alpha_2 = \alpha_3$ and In order for allele I^B to remain in equilibrium, we have $\alpha_2 = \alpha_1$ and $\alpha_3 = \alpha_2$. □

Assume that mutation from I^A to I^B occurs at a rate a and mutation from I^B to I^A occurs at a rate b at a given time t.

Theorem 5. Assume that the selection coefficients for each genotype I^AI^A, I^AI^B and I^BI^B are $\alpha_1,\ \alpha_2,\ \alpha_3$ respectively and that the equilibrium state occurs. The mean change for allele I^A is

$$\frac{1}{\hat{\alpha}}\frac{ab^2(\alpha_1-\alpha_2)+a^2b(\alpha_2-\alpha_3)}{(a+b)^3}$$

and the mean change for allele I^B is

$$\frac{1}{\hat{\alpha}}\frac{ab^2(\alpha_2-\alpha_1)+a^2b(\alpha_3-\alpha_2)}{(a+b)^3}.$$

Here

$$\hat{\alpha}=\frac{1}{(a+b)^2}\{b^2(1-\alpha_1)+2ab(1-\alpha_2)+a^2(1-\alpha_3)\}.$$

Proof. The product ap_{t-1} for allele I^A changes to I^B and the product bq_{t-1} for allele I^Bchanges to I^A. If at a given time t, the frequencies of I^A and I^B are p_t and q_t, respectively. then after next generation the frequency of I^A is

$$p_t=p_{t-1}-ap_{t-1}+bq_{t-1}.$$

If we let

$$\Delta p=p_t-p_0,$$

we have

$$\Delta p=(p_{t-1}-ap_{t-1}+bq_{t-1})-p_{t-1}=bq_{t-1}-ap_{t-1}.$$

Since the equilibrium occurs in the procedure of mutation and the equilibrium state occurs when I^A alleles changing to I^B alleles is the equal to I^B alleles changing to I^A alleles, we have

$$aP=bQ,\ aP+bP=b$$

where and P and Q are the equilibrium allele frequencies of p_t and q_t, respectively. Therefore

$$P=\frac{b}{a+b},\ Q=\frac{a}{a+b}.$$

Since the mean change for allele I^A is

$$PQ\frac{P(\alpha_1-\alpha_2)+Q(\alpha_2-\alpha_3)}{\hat{\alpha}}$$

from Lemma 1, we have

$$PQ\frac{P(\alpha_2-\alpha_1)+Q(\alpha_3-\alpha_2)}{\hat{\alpha}}=\frac{1}{\hat{\alpha}}\frac{ab^2(\alpha_1-\alpha_2)+a^2b(\alpha_2-\alpha_3)}{(a+b)^3}.$$

Here

$$\hat{\alpha}=\frac{1}{(a+b)^2}\{b^2(1-\alpha_1)+2ab(1-\alpha_2)+a^2(1-\alpha_3)\}.$$

Similarly, since the mean change for allele I^B is

$$PQ\frac{P(\alpha_2-\alpha_1)+Q(\alpha_3-\alpha_2)}{\hat{\alpha}},$$

from Lemma 1, we have

$$PQ\frac{P(\alpha_2-\alpha_1)+Q(\alpha_3-\alpha_2)}{\hat{\alpha}}=\frac{1}{\hat{\alpha}}\frac{ab^2(\alpha_2-\alpha_1)+a^2b(\alpha_3-\alpha_2)}{(a+b)^3}$$

□

Denote $x(t,p)$ be the probability that I^A become fixed in the population by time t-th generation, given that its initial frequency is p.

Corollary 6. Assume that the selection coefficients for each genotype I^AI^A, I^AI^B and I^BI^B are α_1, α_2, α_3 respectively and that the equilibrium state occurs. The adapted partial differential equations

$$\frac{\partial x}{\partial t}=\frac{ab}{4N(a+b)^2}\frac{\partial^2 x}{\partial p^2}+\frac{1}{\hat{\alpha}}\frac{ab^2(\alpha_2-\alpha_1)+a^2b(\alpha_3-\alpha_2)}{(a+b)^3}\frac{\partial x}{\partial p}$$

for frequence of alleles I^A and

$$\frac{\partial x}{\partial t}=\frac{ab}{4N(a+b)^2}\frac{\partial^2 x}{\partial p^2}+\frac{1}{\hat{\alpha}}\frac{ab^2(\alpha_2-\alpha_1)+a^2b(\alpha_3-\alpha_2)}{(a+b)^3}\frac{\partial x}{\partial p}$$

for frequence of alleles I^B. Here

$$\hat{\alpha}=\frac{1}{(a+b)^2}\{b^2(1-\alpha_1)+2ab(1-\alpha_2)+a^2(1-\alpha_3)\}.$$

Proof. This result follows easily from the result of Choi([1]) and M. Kimura.([3]). □

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