

Educational Items Section

Short Communication

Mutations Effect on the Genetic constitution of a population

Robert Kalmes

Institut de Recherche sur la Biologie de l'Insecte, IRBI - CNRS - ESA 6035, Av. Monge, F-37200 Tours, France (RK)

Published in Atlas Database: March 2005

Online updated version: <http://AtlasGeneticsOncology.org/Educ/Mutation30041ES.html>

DOI: 10.4267/2042/38232

This work is licensed under a Creative Commons Attribution-Noncommercial-No Derivative Works 2.0 France Licence.

© 2005 Atlas of Genetics and Cytogenetics in Oncology and Haematology

I- Mutation rate

II- Recurrence law

II-1. Example

II-1.1. Genetic determinism and allele frequencies

II-1.2. Two hypothesis

III- Equilibrium state

I- Mutation rate

Consider an allele couple A_1 et A_2 of frequencies p_t and q_t respectively in generation t ; consider u as the direct mutation rate of A_1 towards A_2 in each generation and v the reverse mutation rate of A_2 towards A_1 .

We define the mutation rate as the probability for a mutation to appear per gamete and per generation.

For example, suppose a population only composed of individuals of the genotype $A_1 A_1$ which contribute to the production of $2N$ gametes in the next generation.

If u represents the mutation rate of A_1 locus towards all the other possible alleles in a diploid population of N individuals, as we have $2N$ concerned genes, the number of new mutants that we get are $2N \times u$, in a given generation. In the population, on $2N$ genes, only one gene among them ($1/2N$) will be fixed. Thus, the probability for a mutant allele to appear in the generation with a fixed gene, is $2N \times u \times 1/2N = u$.

Therefore, the probability for one allele to be substitute by a muted allele is equal to the mutation rate, per gamete and per generation, in a given locus.

II- Recurrence law

In the presence of mutations, the evolution of allele frequencies depends on p_i and q_i frequencies in generation i , but also on the mutation rates of u and v .



The frequency of an allele A_1 , one generation after mutation ($p_i + 1$), corresponds to the frequency of an allele A_1 in the preceding generation (frequency p_i) which would not have muted (probability equal to $1-u$), or to an allele A_2 (frequency q_i) which would have muted in A_1 (probability v).

$$p_i + 1 = (1-u) p_i + v q_i$$

$$q_i + 1 = (1-v) q_i + u p_i$$

II-1. Example

The peppered moth (with the kind authorisation of Pr. Georges Périquet, University of Tours)

The peppered moth, *Biston betularia* (Order: Lepidoptera) is common in Northern Europe. The individuals fly during the night and in the day lay down against the light-coloured bark of trees. Since the 19th century, this species has been often studied; it is represented by two morphs, one light-coloured (typica) and another dark-coloured, or melanic (carbonaria).

II-1.1. Genetic determinism and allele frequencies

The genetic determinism of this coloration is monogenetic and autosomic. The allele carbonaria (C) is dominant on the allele typica (c). The allele frequencies C and c will be named p and q respectively.

Phenotypes	[Melanic form]	[Light form]
Genotypes	CC ou Cc	cc

In the middle of the 19th century, among the British populations, the typica form was largely majority. It is only in 1848 that an individual carbonaria capture had been reported in Manchester area. This form frequency had strongly increased and other melanic individuals, afterwards, had been captured in others industrial areas of England. The frequency increase of the carbonaria form was very rapid in Manchester region; it reached 98 %, in 1895. In less than 50 years, this form had become the great majority into the region. As the variations always occurred in the way of a carbonaria form increase of frequency, it could not be due to a random phenomenon.

II-1.2. Two hypothesis

Two hypothesis are possible; the effect of recurrent mutations, or the action by selection.

The hypothesis of recurrent mutations:

We could think that the carbonaria allele was obtained frequently enough by mutation and thus replace the typica allele. Therefore, we can calculate the mutation rate v from c towards C, which would corresponds to an evolution as rapid as the one observed in the nature.

Under mutations effect alone, the recurrence relation for the allele frequencies c, between two successive generations, can be written as.

$$q_1 = q_0 - vq_0$$

The c allele frequency will be the same as the preceding generation (q_0) reduced by the muted allele frequency (vq_0). Here, We consider that the allele C does not mute in c, which correspond to the most rapid possible evolution under the mutation effect.

As the process is repeated with the course of the generations, we deduce:

$$q_t = q_0 (1-v)^n$$

and

$$v = 1 - (q_t/q_0)^{1/n}$$

Where n represents the generation number.

If we consider that the melanic form frequency, in the Manchester region, was 0,01 in 1848 and 0.98 in 1895, and knowing that the peppered moth gives one generation per year in this region, it is possible, therefore, to calculate the v value which better express the rapidity of the observed evolution.

Thus, in 47 generations, we obtain:

$$v = 1 - (0,141/0,995)^{1/47} = 0,041.$$

Then, the calculate value obtained was about 4 %. That means, to obtain a rapid evolution, as fast as what we can observe in the nature, about 4 gametes per 100 at each generation should be muted from typica to carbonaria. In fact, it is an incompatible rate mutation in respect with the spontaneous mutation rate observed in reality (about 10^{-5} to 10^{-6}). Furthermore, we have to explain the brutal change of mutation rate since 1848!

If the rate of $4 \cdot 5 \cdot 10^{-2}$ really exists, it could be explained by the existence of Transposons (doubtful) or by the effect of other factors of evolution such as the selection.

III- Equilibrium state

The equilibrium state in which performed direct and reverse mutations.

$$q_1 = (1-v) q_0 + u p_0$$

$$q_1 = q_0 - v q_0 + u p_0$$

Δq = the difference between 2 successive generations

$$\Delta q = q_{n+1} - q_n$$

Following the sign Δq we know if:

$$\Delta q > 0 \text{ ----> } q_{n+1} > q_n \text{ ----> } A_2 \text{ increase}$$

$$\Delta q < 0 \text{ ----> } q_{n+1} < q_n \text{ ----> } A_2 \text{ decrease}$$

$$\Delta q = 0 \text{ ----> } q_{n+1} = q_n = q_e \text{ Equilibrium state}$$

If we pose at this level of equilibrium state

$$q_e = q_e - v q_e + u p_e$$

$$u p_e = v q_e$$

$$u p_e = v (1 - p_e)$$

$$u p_e + v p_e = v$$

$$p_e = v / (u + v)$$

and

$$q_e = u / (u + v)$$

The equilibrium state is function of the mutation rate u and v.

$$q_e \in] 0 , 1 [$$

Example:

$$\text{If } u = v \text{ ----> } q_e = 1/2$$

$$\text{If } u = 10^{-5} \text{ and } v = 10^{-6} \text{ ----> } q_e = 0.91$$

What ever the mutation rates are, we can have any equilibrium state.

To pass of $q_0 = 0.51$ to $q_n = 0.71$ and $q_e = 0.91$

Mutations can produce equilibrium states, but they will be reached very slowly through the time. The mutation process does not have major effect on the genetic structure of populations; the variation of allele frequencies is very low through the time. This evolution of allele frequencies varies according the equation:

$$p_{n+1} = p_e + (p_0 - p_e) (1-u-v)^n$$

$$q_{n+1} = q_e + (q_0 - q_e) (1-u-v)^n$$

Study by simulation:

The proposed program permits to simulate the evolution of the frequency q through the generations, from q_0 to q_e , for different values of mutation rate parameters u and v and of q_0 .

Translation: Maureen Labarussias

This article should be referenced as such:

Kalmes R. Mutations Effect on the Genetic constitution of a population. Atlas Genet Cytogenet Oncol Haematol. 2005; 9(3):269-271.
