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## Iterative procedure for estimating gene frequencies in genetics data analysis

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### Abstract

Genetics is the mother of breeding and statistics is the only tool to analysis genetic data to arrive at conclusions which hold good in general with a high degree of accuracy and precision. Biometrical genetics is a branch of genetics that uses various statistical concepts and methods to study the genetical research problems. This may be divided into two major parts namely, population Genetics and Quantitative Genetics. Population genetics deals with the gene frequencies, genotypes and phenotypes in the Mendelian population.

The heredity units which are transmitted from one generation to the next generation are called 'Genes'. In the population genetic, individual genes have no meaning. It is the gene pool consists of population that matters. The gene pool consists of all the genes in all their allele forms in the reproductive gametes of the population. It is described as total genetic information possessed by a population which is available to next generation.

One of the basic Idea of population genetics is the Gene Frequency. If the gene frequencies of certain genes are estimated, then the expected genotypic frequency of the next generation can be calculated in a Random Mating.

**Keywords:** Iterative procedure, estimating gene frequencies

### 1. Introduction

Bio-statistical Techniques have a wide number of practical applications in Genetics and plant breeding.

One may define Biostatistics, Bio statistical Genetics, Quantitative Genetics and Population Genetics as follows:

- Biostatistics or Biometrics:** It is a branch of either Biology or Applied statistics which deals with the application of statistical techniques to study various research problems in the Biology.
- Biostatistical genetics or biometrical genetics:** It is a branch of Genetics that uses various statistical concepts and methods to study the genetical research problems sometimes it is known as statistical genetics or mathematical genetics. This may be divided into two major parts namely (i) Quantitative Genetics and ii) Population Genetics
- Quantitative Genetics:** It can be considered as a branch of Biostatistical Genetics which deals with the study of polygenic or quantitative characters.

### The main features of Quantitative genetics are

- Quantitative genetics is an extension of Mendelian genetics firmly based on Mendelian principles of heredity;
- It provides ways and means for the study of polygenic variation, which is not possible by Mendelian genetics;
- It deals with continuous variation.
- Its estimates are based on means, variances and covariances;
- It helps in better understanding of genetic principles and better planning of crop improvement programmes; and
- R.A. Fisher laid the foundation of quantitative genetics in years 1918.

i) **Population Genetics:** It is a branch of Bio statistical Genetics which deals with the Gene frequencies, and Genotypes and phenotypes in the mendelian population. These populations have two main features namely a) Random mating and b) Equal survival of all genotypes.

**The main features of population Genetics are**

(i) It deals with oligogenic variation or discontinuous variation; (ii) Its estimates are based on frequencies and ratios; (iii) It cannot be used for analysis of polygenic variation; and (iv) Hardy and Weinberg laid the foundation of population genetics in year 1908.

**2. Iterative Procedure for Estimating the Gene Frequencies**

The estimation of population frequencies of alleles at a given genetic locus or the estimation of the population frequencies of Haplotypes is referred to as ‘Gene Frequency Estimation’. A Haplotype, derived from the term ‘Haploid Genotype’ refers to the particular set of alleles present at a series of linked loci on a chromosome i.e., alleles at loci that are present relatively close together on a continuous strand of DNA. Each human being has two haplotypes for any given series of linked autosomal loci; one inherited maternally, and the other, paternally. The haplotype transmitted to an offspring of that individual may be identical to one of these two, or may reflect reshufflings due to recombination events. Allele and haplotypic frequencies may be estimated from various types of human data, including information from pedigrees, parent child dyads, or individuals; however, estimation based upon random samples of individuals is emphasized in this article.

Good estimates of gene frequencies are needed for a number of purposes. They are helpful in planning genetic studies. Marker allele frequencies are required for the application of certain types of linkage analysis.

If a random sample of N individuals is collected and there are  $X_i$  copies of allele  $A_i$  among the  $2N$  alleles, then the estimator of the allele frequency  $q_i$  is  $X_i/2N$ , the maximum likelihood estimator. The variance of  $q_i$  can be obtained by using Binomial

distribution as  $\frac{q_i (1 - q_i)}{2N}$ . Irrespective of the nature of the relationships the maximum likelihood estimation method may be used

to estimate the gene frequencies.

Suppose that a random sample of individuals is taken from the relevant population and that there are k distinguishable phenotypes associated with the locus of interest indexed by  $k = 1, 2, \dots, K$ . Usually, an assumption may be made in order to set up the correspondences between genotype and phenotype, expressed via the allele frequencies. By assuming the Hardy-Weinberg equilibrium law, the population frequency  $\phi_k$  of each phenotype can be expressed as a function of the allele frequencies.

For  $n_k$  individuals with the  $k^{th}$  phenotype, the Likelihood function (L) of the sample is given by

$$L \propto \prod_{k=1}^K \phi_k^{n_k}$$

The logarithmic likelihood function is given by

$$\ln L = \sum_{k=1}^K n_k \ln(\phi_k) + \text{constant}$$

For instance, consider a locus with a co-dominant alleles  $A_1, A_2, \dots, A_i, \dots, A_a$  plus a null allele  $A_x$ , which cannot be detected, with population allele frequencies  $q_1, q_2, \dots, q_a, q_x$ .

One may have  $\binom{k}{2}$  phenotypes of the type  $A_i A_j$ , which occur with population frequency  $2q_i q_j$ , and k phenotypes of the type that

express only a single allele  $A_i$ , which may be either  $A_i$  homozygotes or heterozygotes involving a null allele and occur with population frequency  $(q_i^2 + 2q_i q_x)$ , and a phenotype expressing no allele  $A_x$ , which occurs with population frequency  $q_x^2$ .

If one defines the sample counts of these phenotypes analogously, one may obtain the log likelihood.

$$\ln L = \sum_{i < j} n_{ij} \ln(2q_i q_j) + \sum_i n_i \ln(q_i^2 + 2q_i q_x) + n_x \ln(q_x^2)$$

The estimates of the allele frequencies are obtained using standard iterative methods based upon the maximum likelihood method, and estimates of the standard errors can be derived numerically by using the Fisher’s amount of information. In the application of iterative process any of the Bernstein estimates can be taken as the initial estimates. If  $f_i$  denotes the proportion of individuals expressing the  $i^{th}$  allele, one of the Bernstein estimator for  $q_i$  is

$$q_i^* = 1 - (1 - f_i)^{1/2}$$

$q_x$  is generally estimated by subtraction.

### 3. Conclusions

Under the inferential aspects of Genetic parameters, one may consider the problems of estimation of genetic parameters, for instance, segregation ratios, Gene frequencies, Recombination values, Heritability coefficients, Repeatability coefficients, Genetic correlation coefficients and so on. Then one may turn to the examination of goodness of fit with the estimated genetic parameters and to the comparison of sets of data.

An Iterative procedure for estimating the Gene Frequencies has been proposed in the present research work. The maximum likelihood estimation methods has been used by taking Bernstein estimates as initial estimates for the iterative procedure.

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