

Every human is a carrier of one of the three genotypes AA, Aa, or aa. The genotypes are occurring with the probabilities  $(1 - p)^2$ ,  $2 * p * (1 - p)$  and  $p^2$  whereas  $0 < p < 1$  and testing of  $n$  persons yielded

- $x$  persons had the genotype AA
- $y$  persons had the genotype Aa
- $z$  persons had the genotype aa

Describe the corresponding statistical model and determine the Maximum Likelihood Estimator for  $p$ .

Solution:

The likelihood function is given by [1]:

$$P(x, y, z / p) = \binom{x+y+z}{x} * (1 - p)^{2 * x} * \binom{y+z}{y} * (2 * p * (1 - p))^y * \binom{z}{z} * p^{2 * z} \dots \dots \dots (1)$$

Taking log likelihood of (1) we get,

$$\ln(P(x, y, z / p)) = \ln\left(\binom{x+y+z}{x} * (1 - p)^{2 * x} * \binom{y+z}{y} * (2 * p * (1 - p))^y * \binom{z}{z} * p^{2 * z}\right)$$

$$\ln(P(x, y, z / p)) = \ln\left(\binom{x+y+z}{x}\right) + \ln((1 - p)^{2 * x}) + \ln\left(\binom{y+z}{y}\right) + \ln((2 * p * (1 - p))^y) + \ln\left(\binom{z}{z}\right) + \ln(p^{2 * z})$$


$$\ln(P(x, y, z / p)) = \text{constant}_1 + 2 * x * \ln(1 - p) + \text{constant}_2 + y * \ln(p) + y * \ln(1 - p) + \text{constant}_3 + 2 * z * \ln(p) \dots \dots \dots (2)$$

We set the derivative equal to zero:

$$\frac{2 * z + y}{p} - \frac{y + 2 * x}{1 - p} = 0 \dots \dots \dots (3)$$

Solving equation (3) we got the value of  $p$ .

$$\begin{aligned} \frac{2 * z + y}{p} - \frac{y + 2 * x}{1 - p} &= 0 \\ \rightarrow \frac{(1-p)*(2*z+y)-p*(y+2*x)}{p*(1-p)} &= 0 \\ \rightarrow 2 * z - 2 * z * p + y - y * p - y * p - 2 * x * p &= 0 \\ \rightarrow 2 * z + y - 2 * z * p - 2 * y * p - 2 * p * x &= 0 \\ \rightarrow (2 * z + y) - p * (2 * z + 2 * y + 2 * x) &= 0 \\ \rightarrow p = \frac{2 * z + y}{2 * x + 2 * y + 2 * z} \end{aligned}$$

*check maximum* 

The corresponding statistical model is “multinomial distribution model”. An extension of the binomial distribution is the multinomial distribution. The multinomial distribution is used to simulate the results of  $n$  experiments, where each trial's outcome has a categorical distribution [3].

Exactly one of the fixed finite number  $k$  of possible results with probabilities  $p_1, p_2, \dots, p_k$  (here  $p_i \geq 0$  for  $i = 1, \dots, k$  and  $\sum_{i=1}^k p_i = 1$ ), and there are  $n$  independent trials. Next, the random variable  $X_i$  indicates the number of times outcome number  $i$  was observed over the  $n$  experiments. Then  $X = (X_1, X_2, \dots, X_k)$  follows a multinomial distribution with the parameters  $n$  and  $p$ . Where  $p = (p_1, p_2, \dots, p_k)$  [2].

The PMF of the multinomial distribution is given by

$$P(X_1 = x_1, X_2 = x_2, \dots, X_k = x_k) = \frac{n!}{x_1! x_2! \dots x_k!} * p_1^{x_1} * p_2^{x_2} \dots p_k^{x_k}$$

with,  $\sum_{i=1}^k x_i = n$ , and  $\sum_{i=1}^k p_i = 1$

Reference:

- [1]. <https://math.mit.edu/~dav/05.dir/class10-prep.pdf>
- [2]. Sinharay, Sandip. "Discrete Probability Distributions." (2010): 132-134.
- [3]. Multinomial distribution, [https://en.wikipedia.org/wiki/Multinomial\\_distribution](https://en.wikipedia.org/wiki/Multinomial_distribution)

