

NOTE ON A PAPER BY C. W. COTTERMAN AND L. H. SNYDER

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C. W. Cotterman and L. H. Snyder [1] gave a method to test simple Mendelian inheritance in randomly collected data. From a population assumed to be at equilibrium a sample is taken. The number of homozygous recessives in the sample is known. We wish to estimate the number of heterozygous individuals in the sample.

Let α be the proportion of recessive genes among all genes in the population; π, ρ, τ the proportion in the population of homozygous recessives, heterozygous and homozygous dominant individuals respectively and p, r, t the sampling values of π, ρ, τ . Then

$$(1) \quad \pi = \alpha^2, \rho = 2\alpha(1 - \alpha), \tau = (1 - \alpha)^2, p + r + t = 1.$$

Cotterman and Snyder use as an estimate of r' the quantity $2\sqrt{p}(1 - \sqrt{p})$. It is the purpose of this note to show that this estimate is for all practical purposes equivalent to the maximum likelihood estimate of r .

The joint distribution of p, r and t in samples of n is given by

$$(2) \quad P(p, r, t) = \frac{n! \pi^{np} \rho^{nr} \tau^{nt}}{(np)!(nr)!(nt)!} = \frac{n! \alpha^{2np} [2\alpha(1 - \alpha)]^{nr} (1 - \alpha)^{2nt}}{(np)!(nr)!(nt)!},$$

where $P(p, r, t)$ is the probability of obtaining the values p, r, t in samples of n .

We wish to maximize $P(p, r, t)$ for fixed values of p with respect to α and r .

Maximizing first with respect to α one easily obtains

$$(3) \quad 2\alpha = 2p + r.$$

We can regard α as a continuous parameter and hence (3) must hold at any maximum of $P(p, r, t)$. For any maximum of $P(p, r, t)$ we must further have

$$\frac{n! \pi^{np} \rho^{nr} \tau^{nt}}{(np)!(nr)!(nt)!} \geq \frac{n! \pi^{np} \rho^{nr+1} \tau^{nt-1}}{(np)!(nr+1)!(nt-1)!}$$

and

$$\frac{n! \pi^{np} \rho^{nr} \tau^{nt}}{(np)!(nr)!(nt)!} \geq \frac{n! \pi^{np} \rho^{nr-1} \tau^{nt+1}}{(np)!(nr-1)!(nt+1)!}.$$

This leads to the inequalities

$$(4) \quad \frac{\tau}{nt} \geq \frac{\rho}{nr+1}, \quad \frac{\rho}{nr} \geq \frac{\tau}{nt+1}.$$

Substituting $t = 1 - p - r, \tau = 1 - \pi - \rho$ one easily obtains from (4)

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