

## Genetic Constitution of Consanguine Populations

Let us consider the case of a diallele gene, A1 and A2, with a frequency of p and q in the parents

Knowing that the children's consanguinity is equal to F,

If a child is selected at random, what is the probability that (s)he will be A1A1?

The two copies of gene A in this child may be:

the same by inheritance, an event with a probability of F.

- or not be the same by inheritance, an event with a probability of (1 - F).

In the first case, the probability that the first copy of gene A will be A1, is equal to p, the frequency of A, in the parents, and the probability that the second copy will also be A1, is equal to 1, as both copies of the gene are the same by inheritance. If the first one is A1, then the second will also be A1.

In the second case, the probability that the first copy of gene A will be A1 is equal to p, the frequency of A1 in the parents, and the probability that the second copy will also be A1 is still equal to p, because we are in a situation in which the two copies of the gene are not "the same by inheritance": if the first is A1, the second is not automatically A1: it will only be A1 if it is selected again, with a probability p.

Hence the frequency of the genotypes A1A1 in the children:

$$\text{frequency}(A1A1) = Fp + (1-F)p^2$$

The same logic, with A2 and q replacing A1 and p, gives us the expected frequency of the A2A2 genotype in the children:

$$\text{frequency}(A2A2) = Fq + (1-F)q^2$$

What is the frequency that a randomly selected child will be an A1A2 heterozygote?

The two copies of the gene A, present in this child may be:

the same by inheritance, an event with a probability of F;

or not be the same by inheritance, the opposite event, with a probability of (1-F).

In the first case, the probability that the first copy of gene A, A1, is equal to p, the frequency of A1 in the parents, but the probability that the second copy will be A2 is equal to 0, because we are in a situation in which both copies of the gene are "the same by inheritance"; if the first is A1, then the second is A1 too.

In the second case, the probability that the first copy of gene A is A1 is equal to p, the frequency of A1 in the parents, and the probability that the second copy will be A2 is equal to q, because here we are in a situation in which both copies of the gene are not "the same by inheritance": although it would also be possible for the first to be A2 and the second A1, again with a probability pq.

Hence the frequency of the A1A2 genotypes in the children:

$$\text{frequency}(A1A2) = (1-F)2pq$$

These genotype frequencies are, after being developed and factorized:

$$\text{frequency}(A1A1) = p^2 + Fp(1-p) = p^2 + Fpq$$

$$\text{frequency}(A1A2) = 2pq(1 - F)$$

$$\text{frequency}(A_2A_2) = q^2 + Fq(1 - q) = q^2 + Fpq$$

### Contributor(s)

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