Inbred, consanguineous marriages and their risks

Inbred means crossing between *related individuals*. Related individuals are those who have at least "one common ancestor", at most at the level of a great-grandparent.

Introduction

If inbreeding occurs, **the number of heterozygotes decreases'** and **the number of homozygotes increases'** (applies to the population, as well as to individual families). To investigate inbreeding, an allele of common origin was introduced - **ibd allele'** (Identical By Descent) - it is such an allele that an individual inherited from a *common ancestor.*

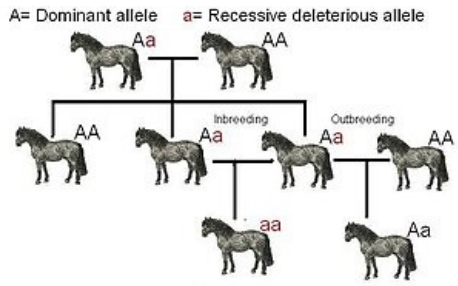
- **Coefficient of inbreeding – F** = determines the probability that in an individual both alleles of a given locus are ibd alleles

$$F = \left(\frac{1}{2}\right)^{n+1}$$

← where **n** = number of generations (connecting lines of the family tree diagram)

Template:Hint

- **Kinship coefficient – f** = is defined as the probability that a randomly selected allele of a given locus of one individual is ibd with an allele randomly selected from the same locus in another individual
- **Relationship coefficient – r** = probability that a randomly selected allele in two related individuals is an ibd allele, i.e.:



Inbred between related individuals.

$$r = 2 \cdot f$$

$$r = \left(\frac{1}{2}\right)^{n}$$

Consanguineous marriages

- From a clinical point of view, they represent an *increased risk* of having a child with autosomal recessive (AR), or a polygenically inherited disease.
- Assuming the disease is rare, then the gene frequency is low and the probability of being homozygous for ibd alleles is relatively high.
- The most common marriages are cousin and second cousin marriages.
- The proportion of AR children born from first-cousin marriages depends on the *gene frequency* and the *amount of marriages* of this type in the population. This is the so-called **Dahlberg relationship** = the dependence of the relative proportion of AR homozygotes born from cousin marriages on the gene frequency and frequency of these marriages in the population.

Inbred in the population - risks

The model population with inbred is based on the assumption that a *relative part of the population (F)* is fully **inbred'** and a *other part of the population (1 – F)* then **panmictic**

- it can be proven that the distribution of genotypes in the population is:

Genotype	AA	Oh	aa
Frequency	p (p + Fq)	2pq (1 – F)	q (q + Fp)

- in the population in which inbreeding takes place, there are no changes in gene frequencies, but there are **changes in the frequency of genotypes'** (heterozygotes decrease and homozygotes increase)
- **inbreeding coefficients** for human populations are usually very low
  - the exception may be so-called "isolates" - they may be geographical (islands, mountain valleys) or social

(nationality, religious sect)

Example

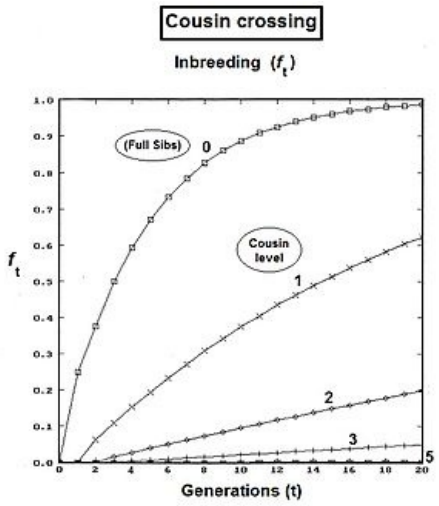
- Disorders of the development of the locomotor system, malformation of the CNS associated with impairment of mental functions (high risk of mental retardation), albinism, hemophilia.

Links

Related Articles

- Relationship coefficient
- Coefficient of inbreeding
- CNS malformation

Source



Dependence of risk on inbreeding