



Université catholique de Louvain
Faculté d'ingénierie biologique, agronomique et environnementale

Methodological aspects of the mapping of disease resistance loci in livestock

Pierre Tilquin

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Promoteurs : P. Baret et E. Le Boulengé

Glossary

ALLELE

An allele, or allelic variant, is a form that can take a gene, or locus. In diploid organisms, each individual carry two "copies", or alleles, of each gene. These two alleles can but do not need to be identical.

BACKCROSS DESIGN

A backcross design is a type of crossing design used in QTL mapping studies in which one examines marker-trait associations in the progeny formed by the backcrossing of the result of a cross between two inbred lines (F_1 or hybrid) to one of the parental lines. As the F_2 design, the objective a such a cross is to generate LINKAGE DISEQUILIBRIUM in the resulting population, i.e. variation in terms of allelic association between loci that would not be encountered in nature or in parental lines.

CANDIDATE GENE

A candidate gene is a sequenced gene of known biological action which is involved in the development or physiology of a quantitative trait (Rothschild and Soller, 1997). In the candidate gene approach, polymorphisms (allelic variations) at candidate genes are used to test for effects on a trait of interest, instead of typing several anonymous markers spread out over the whole genome, as used in the QTL mapping approach.

Glossary

DAUGHTER YIELD DEVIATION (DYD)

The DYD is defined as a weighted average of yield deviations (YD) of a bull's daughters adjusted for merit of their dams (milk, fat, protein); although DYD were first used for milk yield (as indicated in their name), the acronym DYD is now used for many traits as well as for somatic cells in milk.

EPISTASIS

The epistasis is a type of interaction between different loci, in which one locus has an action on another locus either by modifying its expression or by preventing its expression. If there is no epistasis between loci, one can say that loci act additively.

F₂ DESIGN

The F₂ design is a design used in QTL mapping studies to examine marker-trait associations in the progeny from a cross (or selfing) of F₁ individuals, i.e. hybrids that are the results of a cross between two parental lines. As the backcross design, the objective of such a cross is to generate LINKAGE DISEQUILIBRIUM in the resulting population, i.e. variation in terms of allelic association between loci that would not be encountered in nature or in parental lines.

GRANDDAUGHTER DESIGN

In the granddaughter design (Weller *et al.*, 1990), a number of sons of a proven sire are scored for genetic markers and granddaughters are evaluated for the quantitative trait. Then, the trait value for each son is taken to be the mean value of the trait in their daughters (rather than direct measures of the son itself). This design was developed in dairy cattle for milk-production traits, for which the sires are evaluated by means of trait measurements on their daughters or granddaughters.

HALF-SIB DESIGN

The basic idea of the half-sib design, also referred to as the daughter design, is to trace marker alleles from the sire to his offspring and to determine whether progenies that inherited alternative sire alleles differ with respect to the quantitative trait of interest. Due to the extensive use of the artificial insemination in the breeding industry, data from such a design can be found in current breeding populations.

HARDY-WEINBERG EQUILIBRIUM

A population is said to be in Hardy-Weinberg equilibrium when the frequency of each gene and genotype is stable from one generation to the other. Furthermore, in a population in Hardy-Weinberg equilibrium, the frequency of each genotype is a function of the frequency of each allele in the population (Lints, 1991).

HERITABILITY (h^2)

The heritability is the proportion of total variance that is attributable to genetic variance; its most important function in the genetic study of metric characters is its predictive role, expressing the reliability of the phenotypic value as a guide to the genetic value (Falconer & Mackay, 1996).

INFORMATION CONTENT

The information content is a measure of the information contained in marker genotypes; it is computed at each position as the ratio of the observed and expected variance of the conditional probabilities of inheriting one QTL allele at the position being tested.

INNATE RESISTANCE

See NATURAL RESISTANCE.

Glossary

INTERFERENCE

When there is interference between two adjacent region of a chromosome, the presence of a crossover (recombination) in one region has an effect on the frequency of crossover the adjacent region.

LIABILITY

The liability is a concept used for understanding the mode of inheritance of THRESHOLD CHARACTERS. It is the underlying continuous variable that is assumed to govern the expression of a discontinuous characters. When this underlying variable is below a threshold level, the individual has one form of phenotypic expression, and when it is above the threshold the individual has the other phenotypic expression (Falconer & Mackay, 1996).

LINKAGE DISEQUILIBRIUM (LD)

First let us describe what the terms allelic association and linkage disequilibrium (LD) refer to. If there are two tightly linked loci, with two alleles each (A,a at locus 1; B,b at locus 2), then there are four possible combinations of alleles that could exist on the same chromosome, AB, Ab, aB and ab. If allele A has frequency p_A , and allele B has frequency p_B , then the haplotype AB would have frequency $p_A p_B$, for example, in the absence of linkage disequilibrium (*i.e.* the alleles occur independently on haplotypes). If alleles A and B are associated, the frequency of haplotype AB would be $p_A p_B + \delta$, where δ is a measure of the strength of LD between the two loci (Terwilliger & Weiss, 1998).

MICROSATELLITES

Microsatellites are DNA markers that are short arrays of simple repeated DNA sequences (of one to six base pairs) and have the advantages of being abundant, multiallelic, highly polymorphic and codominant (as heterozygotes can be distinguished from homozygotes) and are detected using PCR (polymerase chain reaction).

MOLECULAR MARKER

A molecular marker is a locus with a known position on the genome and that can take various forms (ALLELES) identifiable at the molecular level/

NATURAL RESISTANCE

Natural resistance refers to the inherent capacity of an animal to resist disease when exposed to pathogens, without prior exposure or immunisation (Adams & Templeton, 1998). Such a resistance can also be referred to as innate resistance.

PREDICTED TRANSMITTING ABILITY

The PTA is an estimate of the genetic superiority (or inferiority) that an animal will transmit to its offspring; it is used for both males and females, and includes the deviation of the daughter performance from the population mean adjusted for the genetic merit of the mate (*i.e.* DYD) and the genetic merit of the grandsire and granddam (VanRaden & Wiggans, 1991).

PLEIOTROPY

The pleiotropy is the fact that some genes may have an action on several different traits, and are therefore not governing the expression of only one trait.

PRION

Prion is the a shortened and altered form of *proteinaceous infectious* particle. Prions are agents thought to be responsible for a group of degenerative brain disorders, such as the BSE in cattle, scrapie in sheep and Creutzfeldt-Jakob disease in humans.

Glossary

QUANTITATIVE TRAIT LOCUS (QTL)

A quantitative trait locus is defined as a region of the genome (i.e. segment of chromosome) that harbours one or more genes affecting a quantitative trait (Geldermann, 1975).

QTL MAPPING

The QTL mapping strategy is defined as the methodology aimed at testing for – and positioning on chromosomes – putative QTL involved in the determination of quantitative traits, owing to their linkage with molecular markers.

RECOMBINATION FREQUENCY

The recombination frequency between two loci on a same chromosome is defined as the proportion of gametes produced in a meiosis that will be the result of a chiasma happening between the two loci of interest and resulting in an exchange of chromosomal material between two homologous non-sister chromatids. It is linked to the map distance between loci by what are called mapping functions (e.g. Haldane, Kosambi).

RESILIENCE (OR TOLERANCE)

The resilience is the ability of a host to maintain a relatively undepressed production while subjected to infection, and therefore to withstand the effects (symptoms) of the disease (Bisset & Morris, 1996; Bishop *et al.*, 2002).

RESISTANCE

The resistance is the ability of a host to initiate and maintain responses to prevent the establishment of an infectious agent and/or eliminate the load of this agent.

SEGREGATION

The segregation of genes is defined as the separation, during the meiosis, of different alleles from an allelic pair, which implies that the allelic pair is heterozygote

SELECTIVE GENOTYPING

Selective genotyping is a procedure used to lessen the cost of an experiment by genotyping only individuals from the high and low tails of the phenotypic distribution of the entire sample population. It can markedly decrease the number of individuals genotyped for a given power at the expense of an increase in the number of individuals phenotyped. However, such a procedure has the disadvantage of leading to an overestimation of the QTL effect (Darvasi & Soller, 1992).

THRESHOLD TRAITS

Thresholds traits are characters which vary in a discontinuous manner but are not inherited in a simple Mendelian manner. Familiar example are susceptibility to disease, where there are two phenotypic classes – affected or not affected. Such traits are said to have an underlying continuity with a threshold, which imposes a discontinuity on the visible expression. This underlying continuous variable is called the LIABILITY (Falconer & Mackay, 1996).

TOLERANCE

See RESILIENCE.