
Defining genetic diversity based on genomic tools

Second chapter in the book:

“Genomic management of animal genetic diversity”

Jesús Fernández, INIA and Jörn Bennewitz, Hohenheim University



➤ **You can not maintain what you can not measure!**

✓ **Degree of endangerment**

⇒ **prioritisation**

✓ **Management**

✓ **Monitoring**

⇒ **check for success**

➤ **Keep phenotypic features**

✓ **Morphological**

⇒ breed standard

✓ **Productive**

⇒ profitability

$$V_P = V_G + V_E$$

✓ **Adaptation to particular environment**

➤ **Classical approach through the concept of variance**

✓ **Good recording scheme (standardised and accurate)**

⇒ avoid confounding errors with high variability

➤ **Look for high levels of phenotypic diversity through high levels of genetic diversity**

PEDIGREES

➤ Absent or unreliable

✓ Especially between breeds

⇒ prioritisation

➤ Assume founders unrelated and non inbred

Genomics: Mendelian sampling visible by DNA analysis

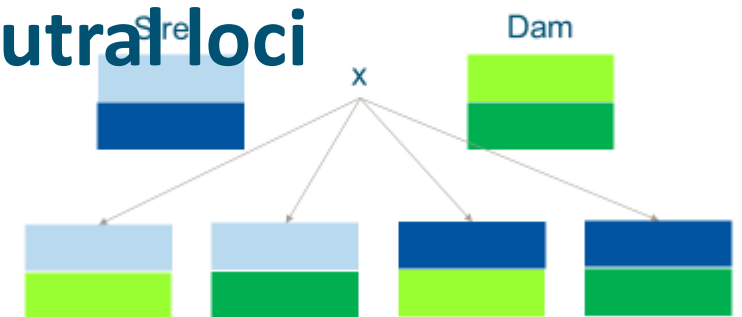
DNA

- Present in two copies (pairs of chromosomes)
- Always 50% submitted by the dam and 50% by the sire
- A random process determines which part from the sire and from the dam

➤ Average 'expected' value for neutral loci

✓ No Mendelian sampling

⇒ no all sibs are equal



Genomics determines which part from the sire and which part from the dam!

MOLECULAR INFORMATION

➤ Deal with 'realised' values

✓ percentage of polymorphic sites

✓ distribution of allelic frequencies

✓ observed and expected heterozygosity

✓ allelic diversity

⇒ detect relevant
individuals or
populations

- **dense coverage by SNPs**
 - ✓ **every locus in LD with one marker**
 - ⇒ more precise measure
 - ✓ **measure non-neutral genetic diversity**
 - ⇒ account for productivity or fitness
 - ✓ **separate analysis of particular regions**
 - ⇒ instead of global picture
 - ✓ **finer determination of relationships between individuals/breeds**
 - ⇒ crucial in management

➤ **Close SNPs inherited together**

✓ **use haplotype (kinship)**

⇒ **detect selection signatures**

✓ **Runs Of Homozygosity (ROHs)**

⇒ **reflect IBD if they are long enough**

⇒ **but still ‘realised’ IBD**

‘ ... long stretches of two homologous chromosomes within the same individual that are identical (homozygous for all the loci within) ...’

➤ **whole sequence**

- ✓ **detect other types of markers, e.g. Copy Number Variants (CNV)**
- ✓ **causal mutations for important traits are present**
⇒ **not depending on LD with the SNPs**
- ✓ **easier to detect rare variants**
- ✓ **efficient way of detecting SNPs for rare breeds**
⇒ **avoid ascertain bias from commercial SNP chips**

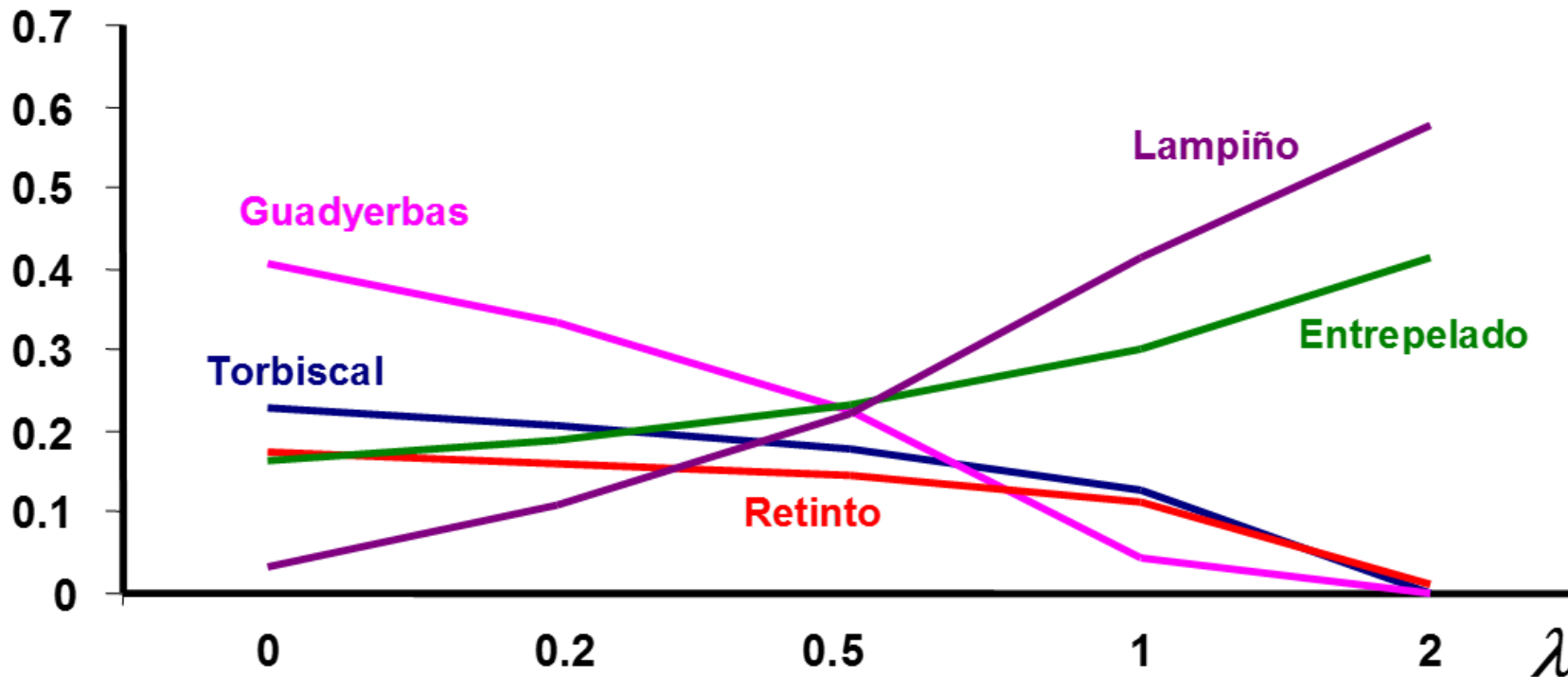
➤ **Partition of diversity within and between breeds**

✓ better description of genetic structure

✓ prioritisation of breeds

$$GD = \lambda GD_W + GD_B$$

⇒ $\lambda = 0$ ⇒ Weitzman
⇒ $\lambda = 0.5$ ⇒ TGV
⇒ $\lambda = 1$ ⇒ Exp. Het.



- **trait-based adaptive diversity measures**
 - ✓ **excess of variance in genotypic values relative to the variance expected in the absence of selection**
 - ✓ **adaptivity coverage of a set of subpopulations**
 - ✓ **how well the subpopulations could adapt to a large range of environments**