Population Differentiation Index

Towards a generic measure of genetic relatedness of populations

EAAP 2024, Jan ten Napel, Jack Windig, Mira Schoon, Mirjam Spoelstra, Sipke-Joost Hiemstra

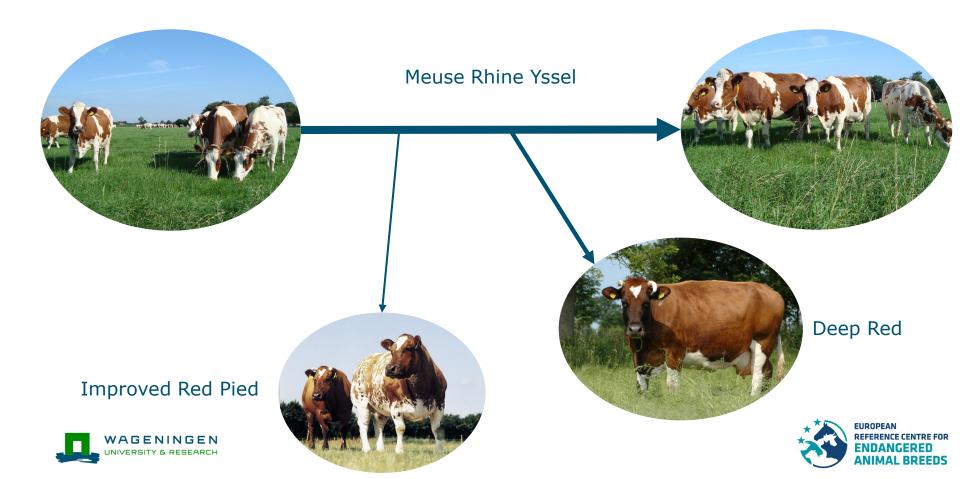








How much do populations overlap?



No clear guidelines to measure overlap

- National Coordinators, herdbooks, rare breed societies, breeding companies
 - Conserve separately or not?
 - Two breeds with a common origin, but no exchange
 - Two breeds with a low structural level of exchange
 - Transboundary breeds
 - What is a sustainable level of exchange?
 - Is there a risk of fragmentation of a population?





Current practice in population genetics

- Most commonly used measure of genetic differentiation between subpopulations is Fixation Index (F_{ST})
 - Developed by Sewall Wright (1951) and Gustav Malécot
 - Fraction of total expected heterozygosity (H_T) that is not a consequence of the average expected heterozygosity within subpopulations (H_S)
 - Affected by migration, drift and mutation
 - Captures changes in allele frequency
 - Can be used for individual SNP or microsatellite data

$$F_{ST} = \frac{H_T - H_S}{H_T}$$





Practical issues of F_{ST} for animal breeds

- Requires genomic information on representative & sufficiently large sample in each subpopulation
- Difficult to compare across species and studies
 - Dependent on good estimates of allele frequencies
 - Dependent on the marker set used
- Difficult to differentiate between historical and recent genetic relatedness





Objective

Develop a generic measure of genetic relatedness of subpopulations based on pedigree data that can be used to provide guidelines for conservation and management





Genetic drift within and between subpopulations

Considerations on Genetic Connectedness Between Management Units Under an Animal Model¹

B. W. Kennedy and D. Trus²

Centre for Genetic Improvement of Livestock, University of Guelph, Guelph, Ontario, Canada N1G 2W1

ABSTRACT: Connectedness among management units (e.g., herds or regions) is of concern in genetic evaluation. When genetic evaluation is under an animal model, connections occur through A, the numerator relationship matrix. It is argued that the most appropriate measure of connectedness is the average prediction error variance (PEV) of differences in EBV between animals in different management units. It is shown that PEV of differences is influenced by average genetic relationship between and within management units, which in turn affects the variances of estimates of differences between management unit effects. When PEV of differences cannot be computed,

use of one of three alternative measures is proposed; the gene-flow method that measures the exchange of genes between management units, measurement of genetic drift variance based on average relationships between and within management units, and measurement of the variance of estimated differences between management units effects. These were correlated with PEV of differences in a test simulation. The gene-flow method, which is simplest to compute, had the lowest correlation (.671). The drift variance and variance of management unit effects methods were highly correlated with PEV of differences (.924 and .995, respectively).

Key Words: Connectedness, Animal Models, Genetic Evaluation

J. Anim. Sci. 1993. 71:2341-2352





Population Differentiation Index

$$PDI_{i} = \frac{\overline{a}_{11,i} + \overline{a}_{22,i} - 2\overline{a}_{12,i}}{\overline{a}_{11,i} + \overline{a}_{22,i}} * \frac{\sigma_{a}^{2}}{\sigma_{a}^{2}} * 100\%$$

 $\bar{a}_{11,i}$ and $\bar{a}_{22,i}$ is average genetic relationship within subpopulations 1 and 2 in year of birth i (not including an individual with itself)

 $\bar{a}_{12,i}$ is the average genetic relationship between subpopulations 1 and 2 in year of birth i

Calculated from the pedigree relationship matrix

Captures expected change in allele frequencies due to genetic drift





Example: Belgian and Dutch Draught horse

Two populations with full pedigree available

Truncated at animals born in 1970 so two unrelated

populations

Low level of exchange of stallions

Size active breeding population

• Belgium: 449 mares (2020)

Netherlands: 249 mares (2021)

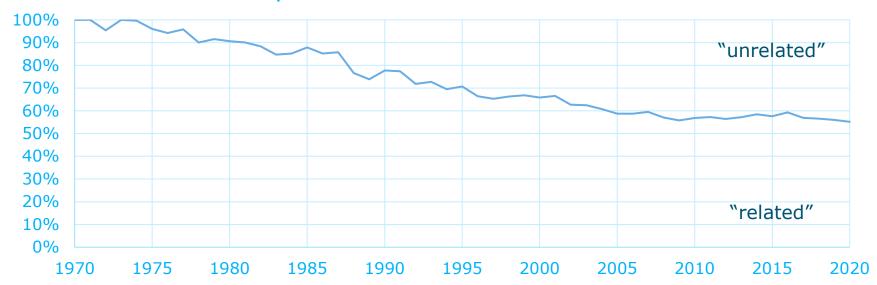






Genetic relatedness of Belgian and Dutch Draught horse over time

Population Differentiation Index





Properties of Population Differentiation Index

- The PDI is similar to the Fixation Index, but is not the same
- The scale (0-100%) is independent of species, population sizes, rates of inbreeding and pedigree depth
- It is relatively easy to calculate from the pedigree relationship matrix
- It is a measure for pairs of populations
- It allows to monitor genetic relatedness of populations over time
- If no pedigree available, it is possible to simulate both populations given demographic information and monitor expected PDI per year





Draft table for interpreting PDI

PDI	Interpretation
0-20% "hot"	Freely interbreeding populations
20-40%	Highly related populations
40-60%	Related but distinct populations
60-80%	Largely unrelated populations
80-100% "cold"	Two separate populations

Suggested use

PDI < 20%: populations do not need to be conserved separately

PDI > 80%: risk of fragmentation if subpopulations of the same breeding program

Need to build up experience for more detailed guidelines





Population Differentiation Index

A generic measure for monitoring genetic differentiation of subpopulations

It may contribute to harmonizing conservation and breeding programmes across countries and species





