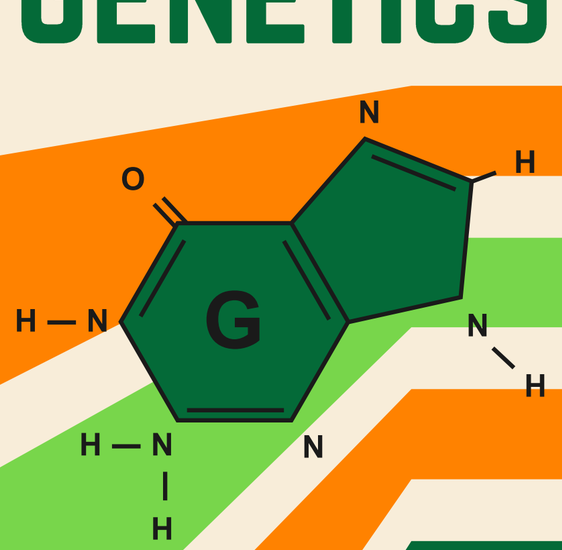
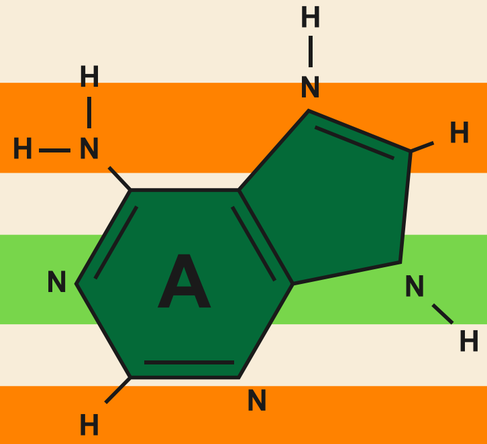
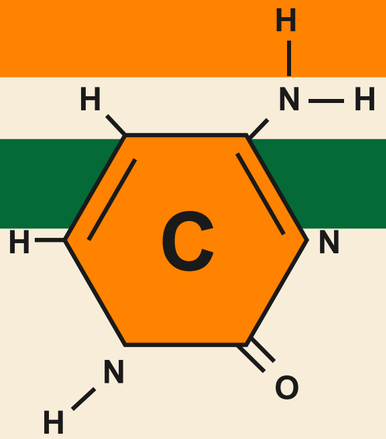
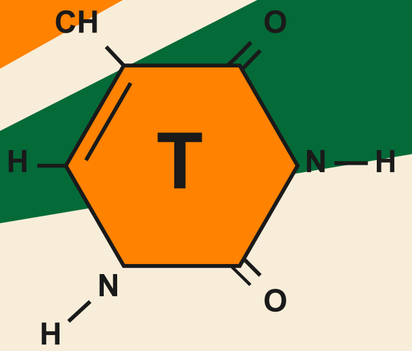
WBFSH General Assembly 2027

Studbook applications of Genetics & Genomics









Jennifer Doyle, BAgrSc, PhD
Senior Geneticist

HORSE SPORT IRELAND

(c)

DNA Testing of Irish Horses

- Pre 1990s → “Believed to be by”
- 1990 - 2000 → Blood Typing
- 2000 - Present → Microsatellites
- 2023 - Present → Single Nucletide Polymorphisms



Microsatellites (MS)

- AKA Short Tandem Repeats (STRs)
- Repeated segments of DNA
- The number of repeats varies by animal
- 11 to 17 markers for parentage verification
- Current ISAG approved method of equine Parentage Verification

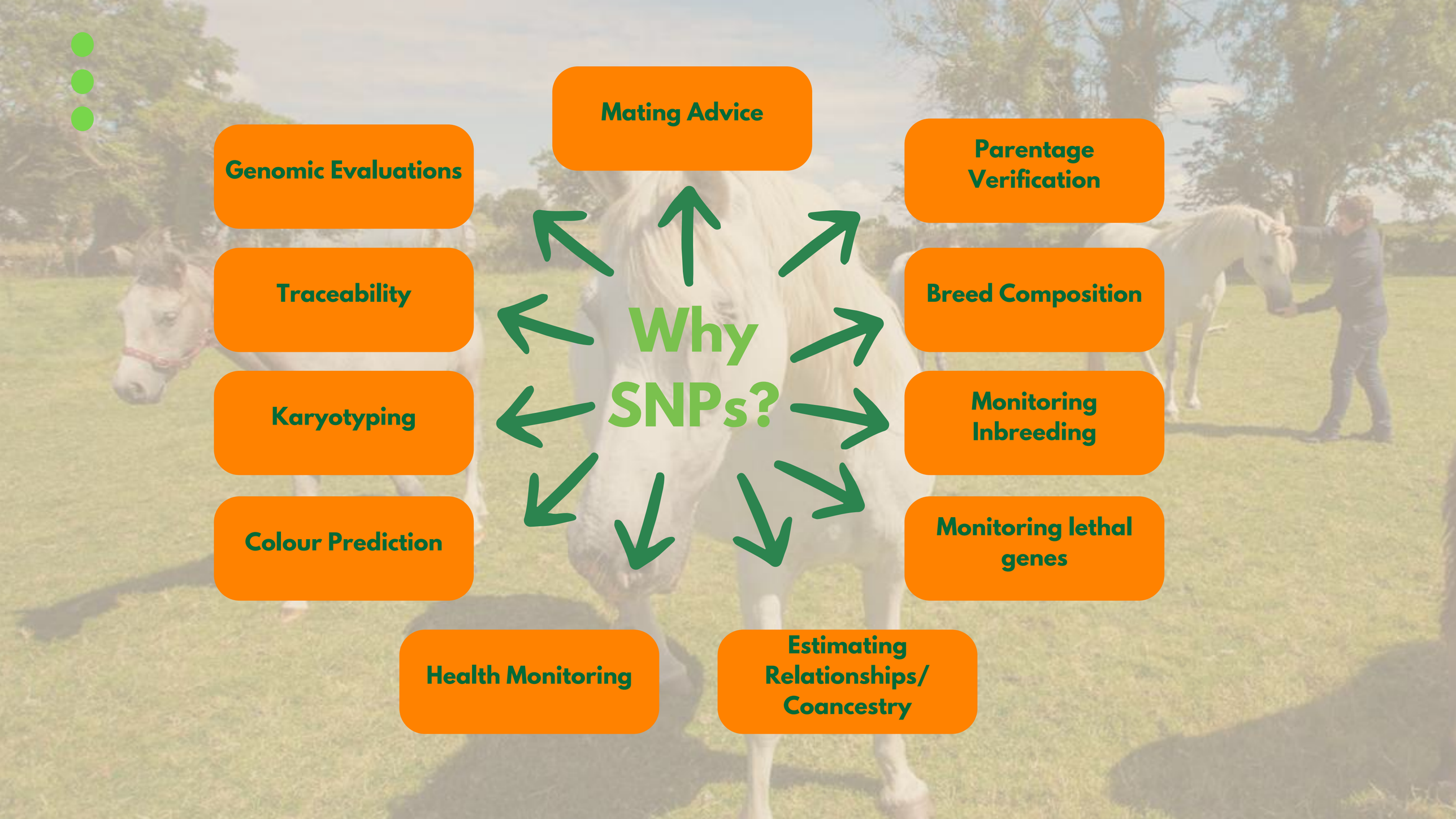
Single Nucleotide Polymorphisms

- Single base pair change in the DNA sequence
- Most common type of genetic variation
- Different arrays test for different number of SNPs
  - HSI = Illumina Equine80Select BeadChip
- Many uses beyond parentage verification
- Not yet ISAG approved - 2027?

ISAG

- International Society of Animal Genetics
- Set official marker panels and guidelines for parentage verification
- Equine SNP parentage comparison tests are ongoing
- Current Panel:
  - 381 autosomal SNPs (+397 backup markers)
  - 8 Y-chromosome SNPs
  - 76 X-chromosome SNPs
- HSI have contributed samples for comparison testing via Paul Flynn/ Weatherbys

ISAG
INTERNATIONAL SOCIETY FOR ANIMAL GENETICS


Why SNPs?
Mating Advice
Parentage Verification
Breed Composition
Monitoring Inbreeding
Monitoring lethal genes
Estimating Relationships/ Coancestry
Health Monitoring
Colour Prediction
Karyotyping
Traceability
Genomic Evaluations

Parentage Verification

- Ensures scientifically verified accurate pedigree recording
  - Removes uncertainty around horses origins
- Studbook Quality Assurance
- Can increase market value of horse
- Improves traceability



Parentage Verification

- Still get breeders making errors with parentage

Case Study

- ISH studbook received foal application for TIH foal
- The foal excluded from nominated sire during the SNP parentage
- Subsequently verified to a warmblood stallion
- Breeder not happy - new sample submitted - Foal excluded again
- Breeder states he “only breeds TIH”
- Breeder submitted a new sample that verified against original parentage
- Similarity of sample checked against database
- Breeder had sent in his yearlings sample as that of the foal

Monitoring Genes /Health

- SNP chips have built in pre-designed targets for monitoring SNPs of interest
- Different chips = different SNPs

- May have patents in place in your territory
  - Eg., FFS - no patent in ROI but patented in NI

- Illumina Equine80Select - Malformations, defects, diseases, coat colours

Fragile Foal Syndrome



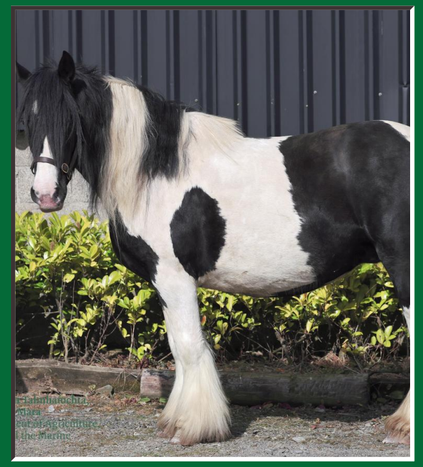
- SNP validated
- Prevalence being monitored
- Estimated that 2.8% of population are carriers
- Must have 2 copies of FFS gene to be affected
  - Breeding 2 carriers - 25% chance foal will have FFS

Hoof Wall Separation Disease



- SNP validated
- Prevalence being monitored
- Originally thought to just affect Connemara Ponies
  - Identified in ISH / ISP with CP in pedigree
  - Also appears in the Irish Cob
- Must have 2 copies to have HWSD
  - breeding 2 carriers - 25% chance foal will be affected



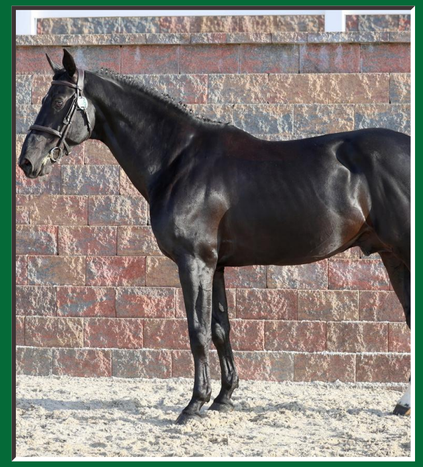




Colour Traits

- Undergoing validation
  - Extension
  - Agouti
  - Cream
  - Dun
  - Grey
  - Silver
  - Tobiano
  - Lethal White Overo
- Rollout 2027?







Karyotyping

- SNP analysis cheaper & easier than conventional karyotyping
- Uses variables already generated for the SNP Final Report file
- Can detect both autosomal & sex chromosome abnormalities

animals

MDPI

Article

Two Cases of Chromosome 27 Trisomy in Horses Detected Using Illumina BeadChip Genotyping

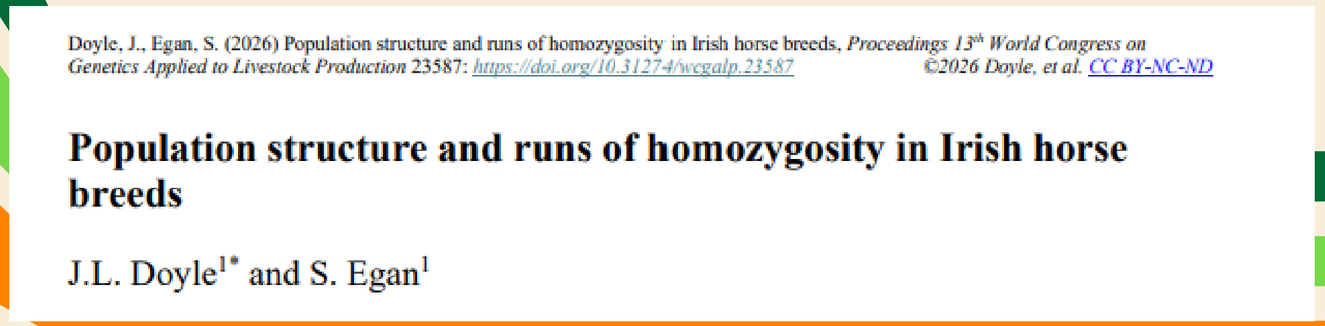
Cliona A. Ryan [1,*], Donagh P. Berry [1], Monika Bugno-Poniewierska [2], Mary-Kate Burke [3], Terje Raudsepp [4], Sonja Egan [5] and Jennifer L. Doyle [5]





Breed Composition

- Well established methods in other species
- Essential for conservation and sustainable management in smaller studbooks
- Has allowed HSI to identify Irish Draughts who otherwise would have remained outside the studbook
- Combination of:
  - Genomic Relationship Matrix to find relatives
  - PCA and admixture analysis to determine breed



Doyle, J., Egan, S. (2026) Population structure and runs of homozygosity in Irish horse breeds, *Proceedings 13th World Congress on Genetics Applied to Livestock Production* 23587: https://doi.org/10.31274/wcgalp.23587 ©2026 Doyle, et al. CC BY-NC-ND

Population structure and runs of homozygosity in Irish horse breeds

J.L. Doyle[1*] and S. Egan[1]



Genetic Resources

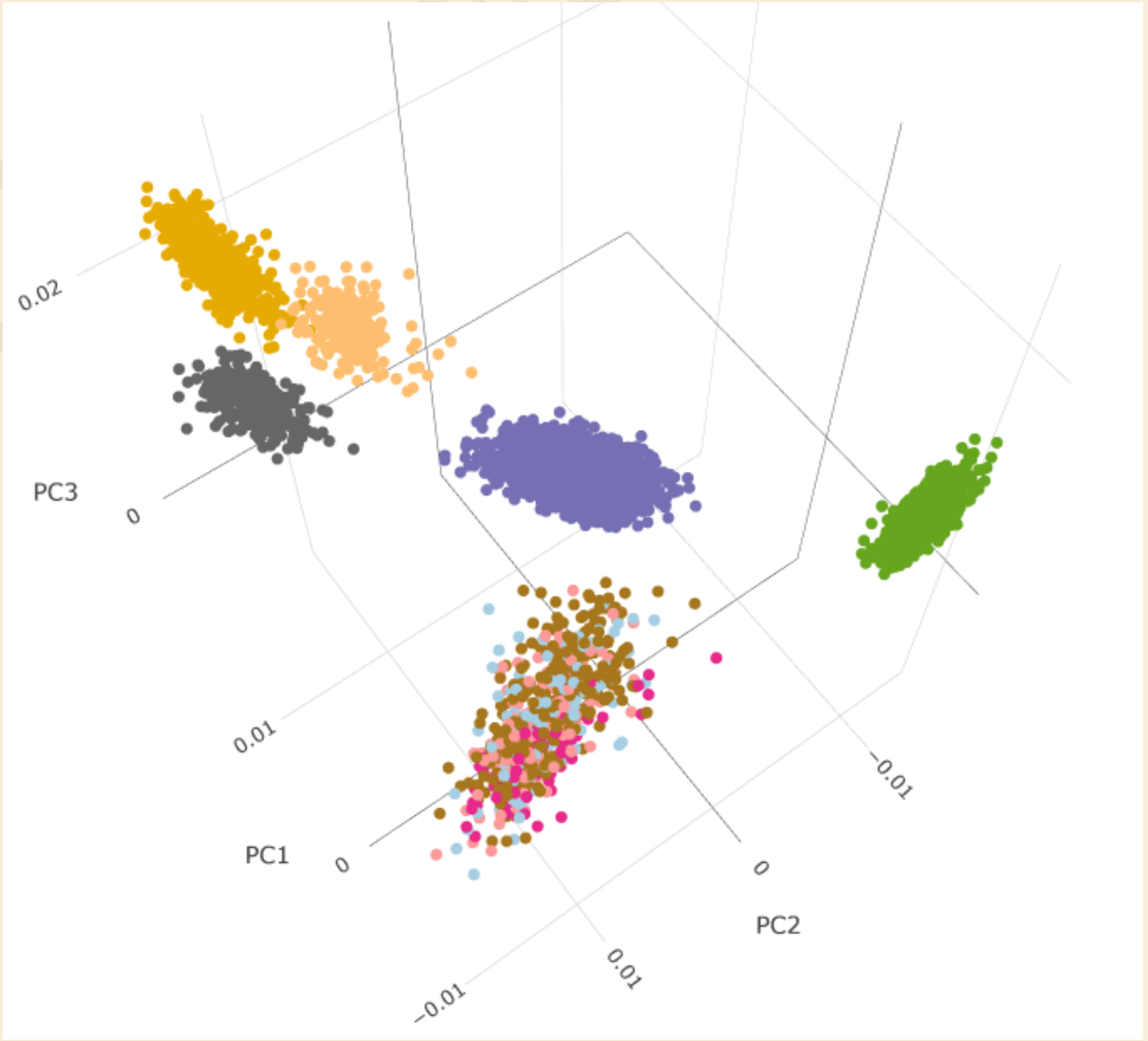
SHORT COMMUNICATION

Genetic Resources (2023), 4 (8), 29–36
DOI: 10.46265/genresj.VOOZ8371
https://www.genresj.org
ISSN: 2708-3764

The genetic composition of the Traditional Irish Horse – towards the development of a DNA-ancestry test for the preservation of traditionally bred Irish Sport Horses

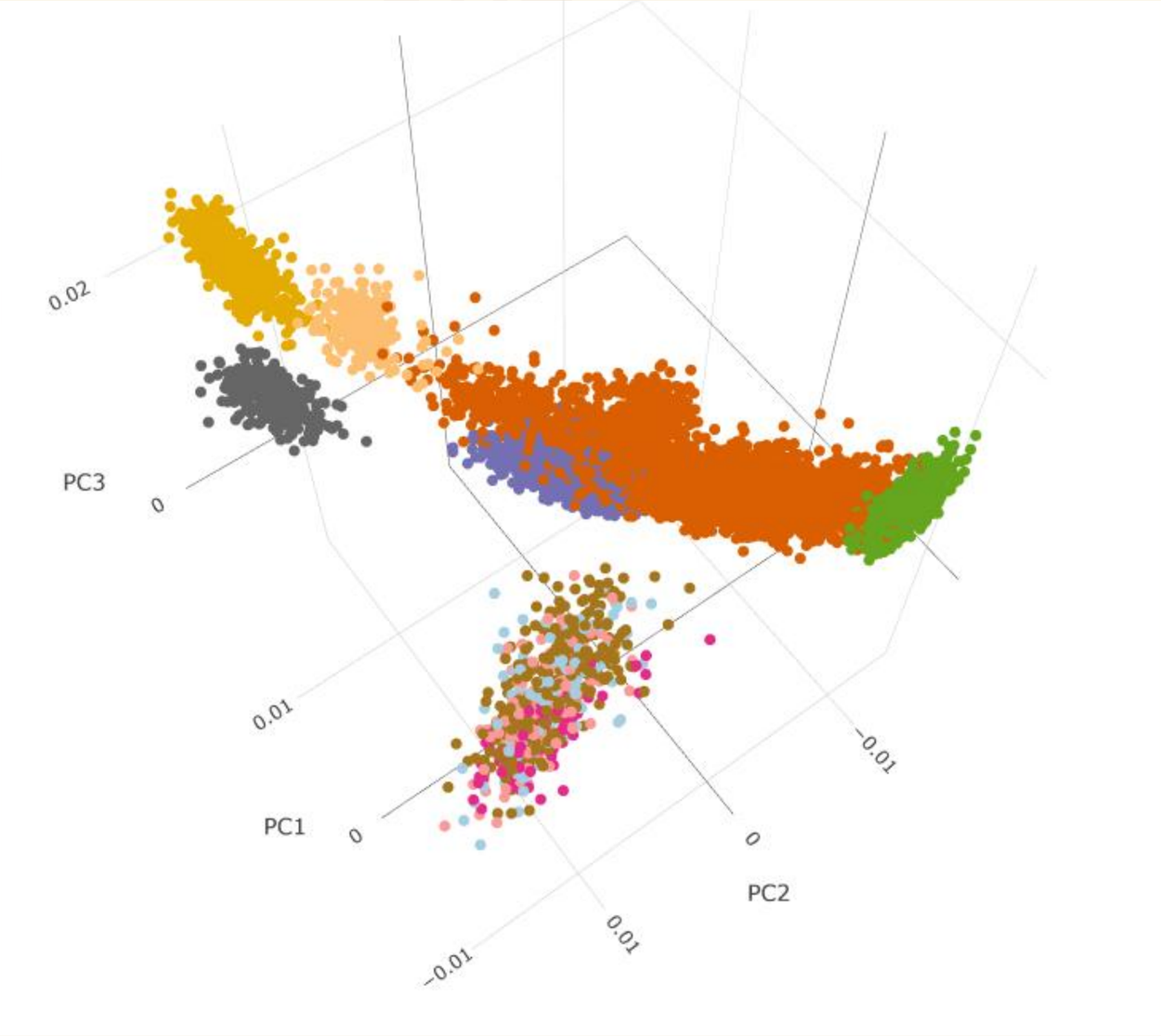
Beatrice A McGivney[a], Deirdre Harty[b], Alison Corbally[b] and Emmeline W Hill *,a,c

Principal Component Analysis



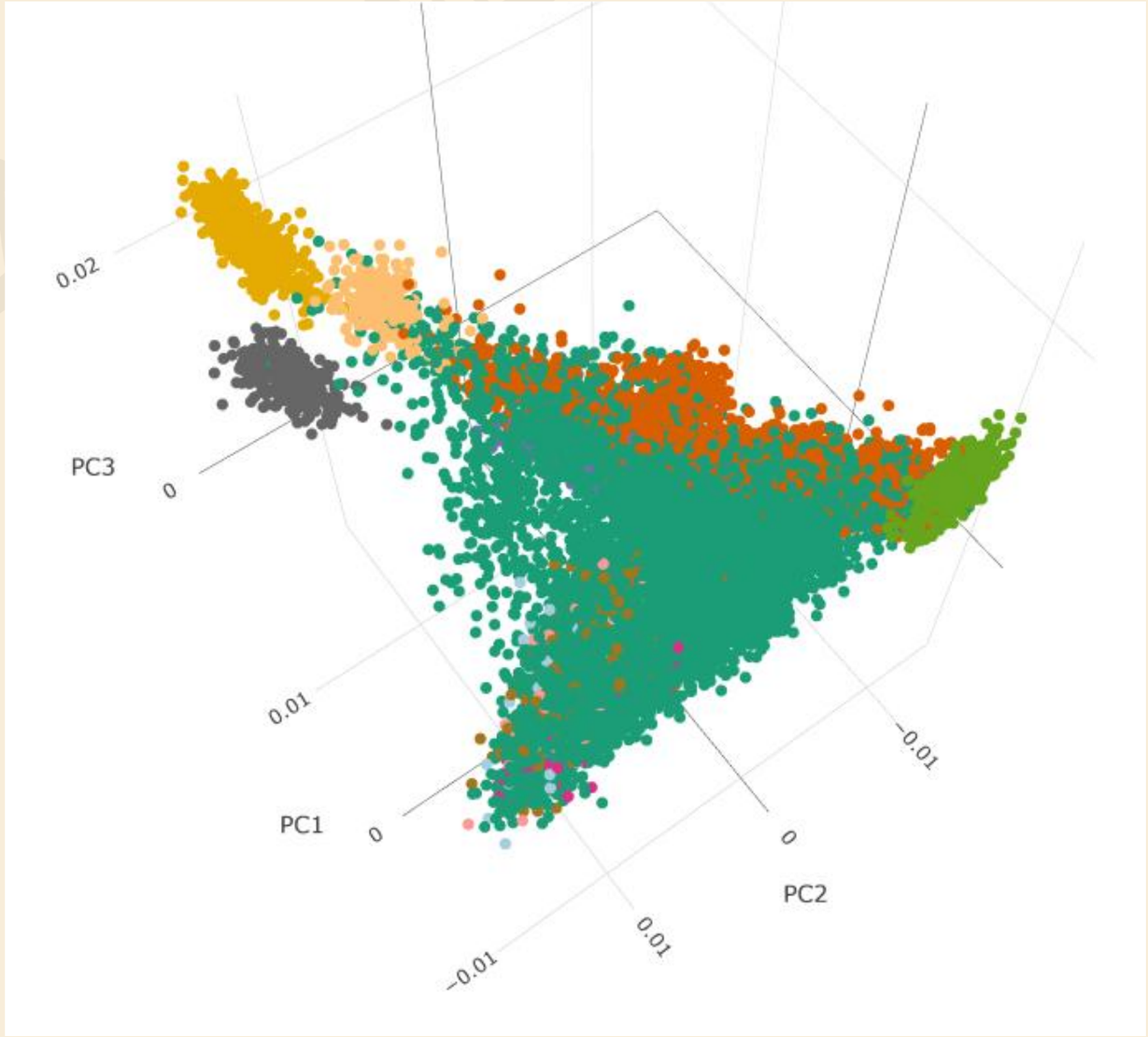
- Belgian Warmblood
- Connemara
- Dutch Warmblood
- Holsteiner
- Irish Cob
- Irish Draught
- Irish Sport Horse
- Kerry Bog Pony
- Thoroughbred
- Traditional Irish Horse
- Zangersheide

Principal Component Analysis



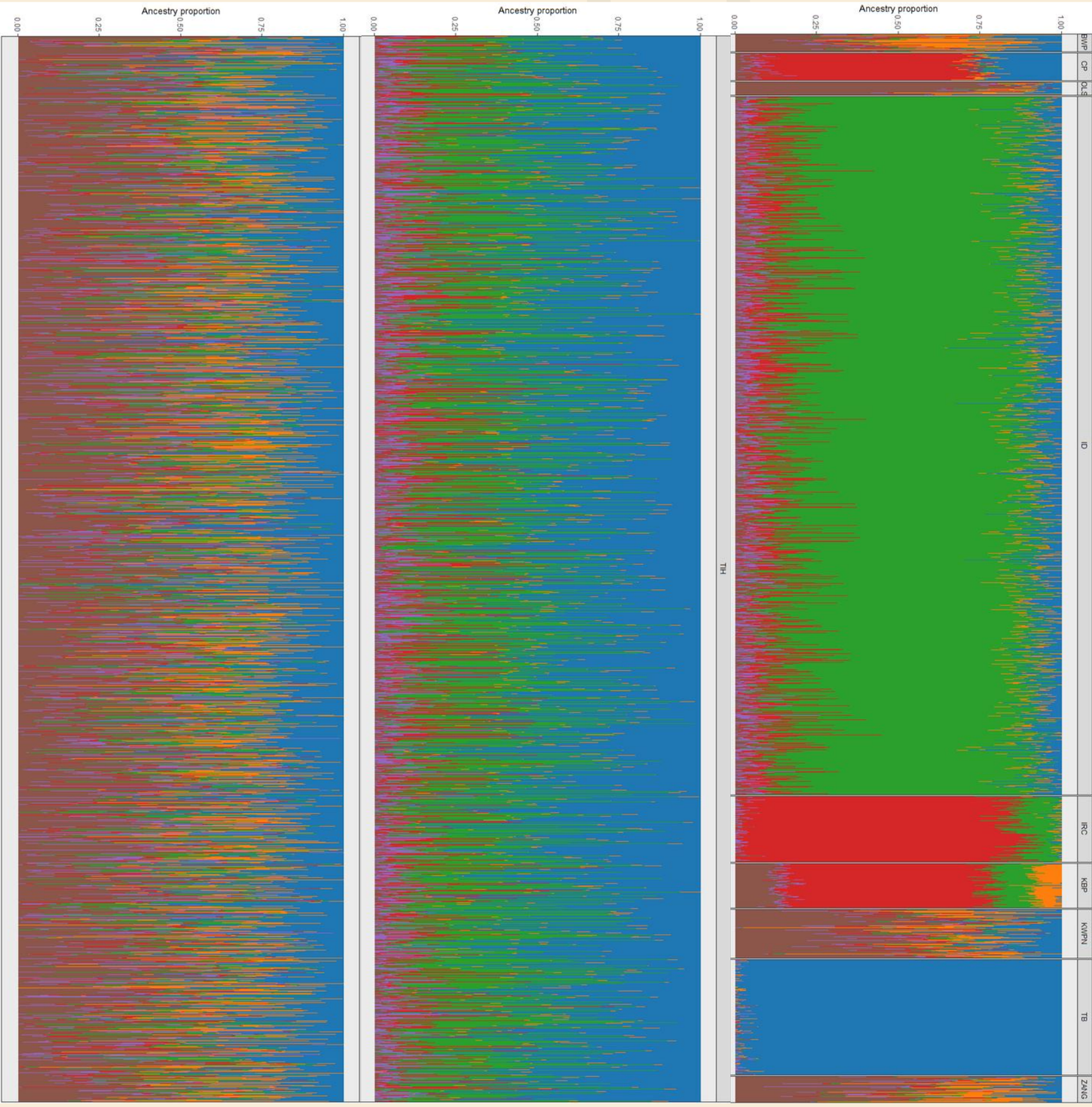
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Principal Component Analysis



- Belgian Warmblood
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Admixture Analysis

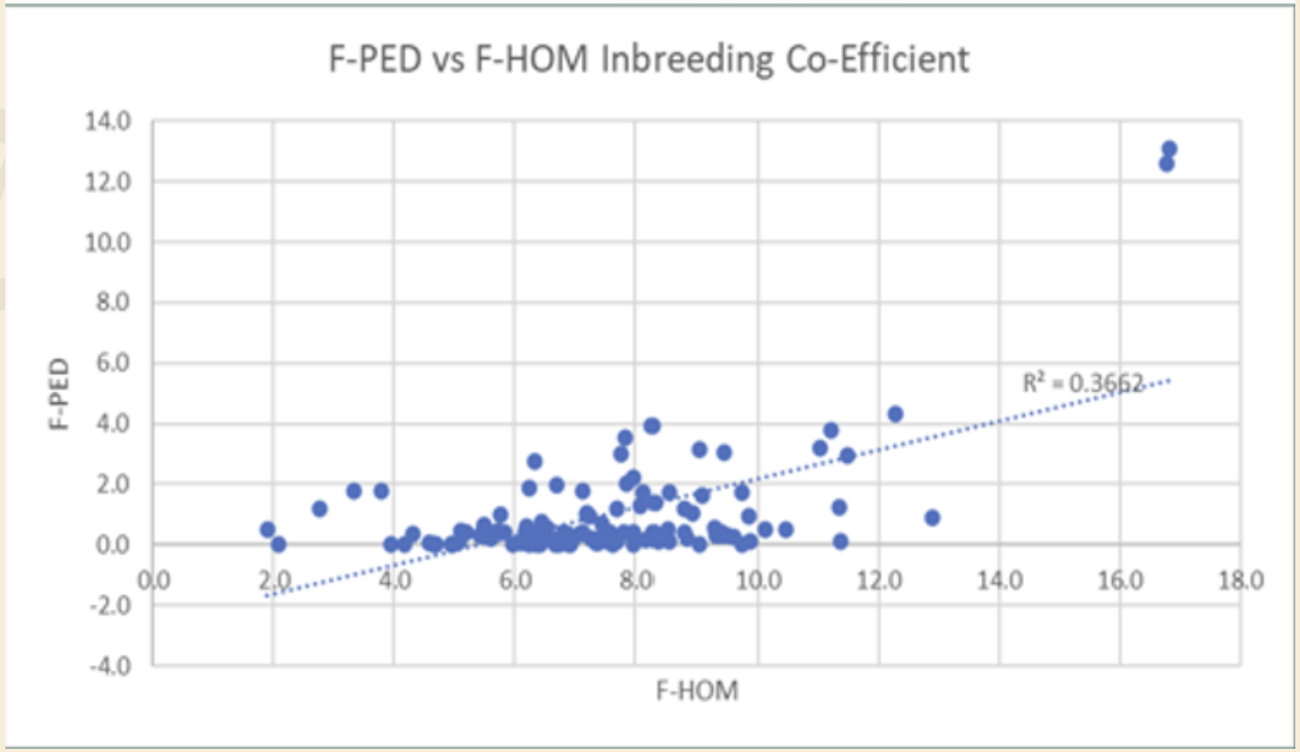


Inbreeding

Pedigree Inbreeding: calculates an expected statistical probability based on recorded pedigree

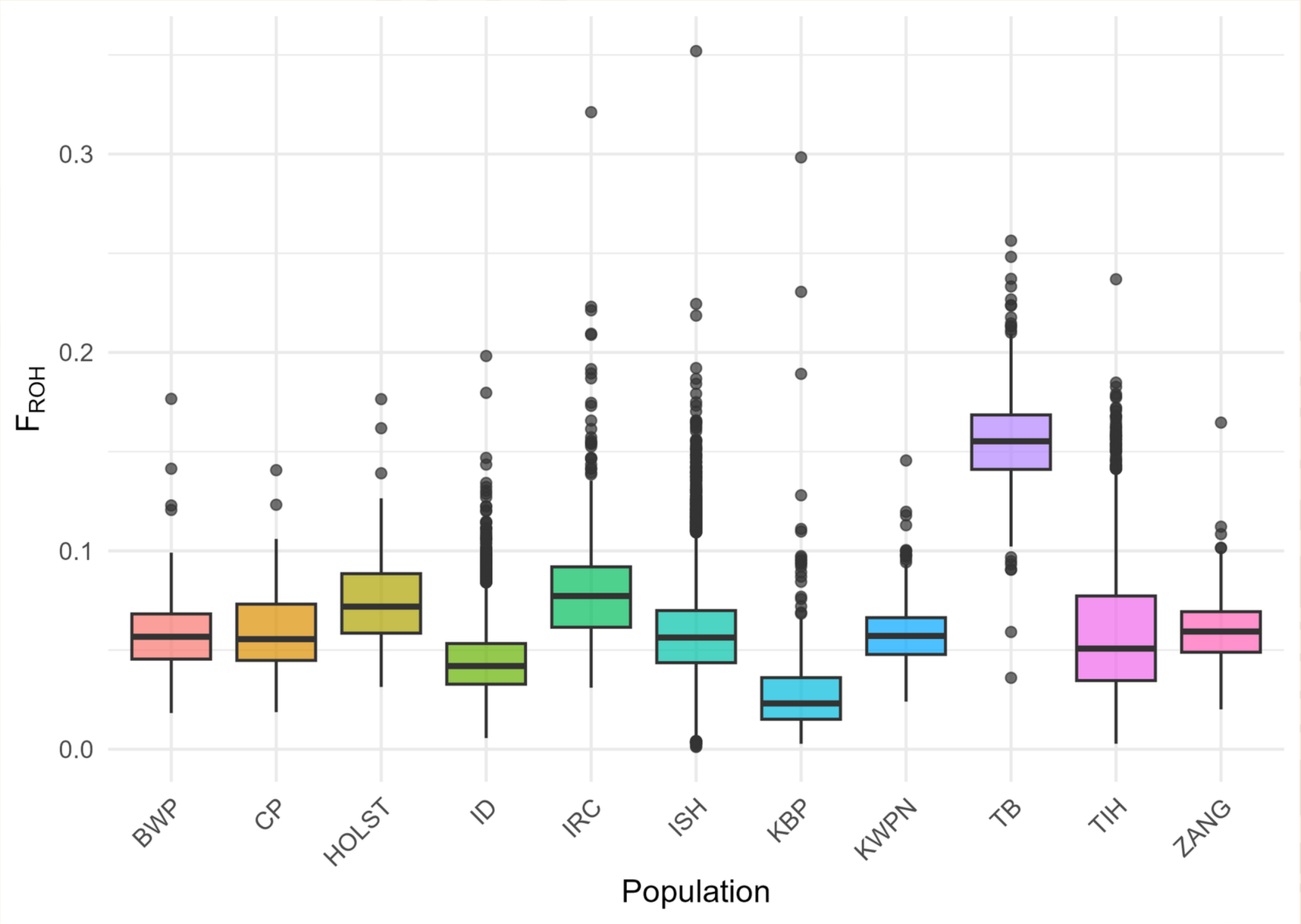
Genomic Inbreeding: measures actual DNA homozygosity independent of recorded pedigree.

Inbreeding



Time Period	Number of Animals	F-PED vs F-ROH R2 Correlation	Average Number of Pedigree Generations
Pre-1990	5	0.102	1
1990-2000	28	0.086	2
2001-2010	33	0.391	3
2011-2020	52	0.466	4
ALL	118	0.367	3

Inbreeding


0.3
0.2
0.1
0.0
F_{ROH}
BWP
CP
HOLST
ID
IRC
ISH
KBP
KWPN
TB
TIH
ZANG
Population

Genomic Evaluations

EBVs

- Relies on accurate pedigree records & performance records
- Full-siblings will have the same EBV
- Years of waiting for performance records or relying on parental averages
- Lower reliability for young animals

gEBVs

- Uses actual DNA (genotype) alongside EBV data
- Identifies distinct genetic differences between full siblings
- Enables earlier selection of genetically animals
- Much higher reliability at a younger age
- Improved insights for difficult to measure traits

Improved Traceability

- Microchips / passports can & have been altered
- Horse's can look similar and can get mixed up
- DNA is the gold standard for identifying any organism
  - a genomic ID provides a permanent, tamper-proof biological barcode

Case Study

- Irish mare exported to UK with passport
- Microchip in Passport matched the microchip in the mare.
- Had a foal - UK PIO contacted HSI for mares DNA
- Foal excluded from the mare
- Ran DNA comparison test and a discrepancy was identified between the DNA
- DNA identified who the mare in the UK really was

Genotype Management

- >78000 SNPs per animal x thousands of animals
- Phenotypic & pedigree data
- Problems?
  - Storage/management issues
  - Lack of computing power
  - Lack of knowledge on how to interrogate data



How is HSI handling the massive amount of data generated from SNP genotyping?

Helical

THANK YOU!

Jennifer Doyle, BAgrSc, PhD
Senior Geneticist

HORSE SPORT IRELAND

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