Identifying genetic diversity within indigenous and highly commercialized breeds for improved performance and future preservation

Cornell CALS College of Agriculture and Life Sciences

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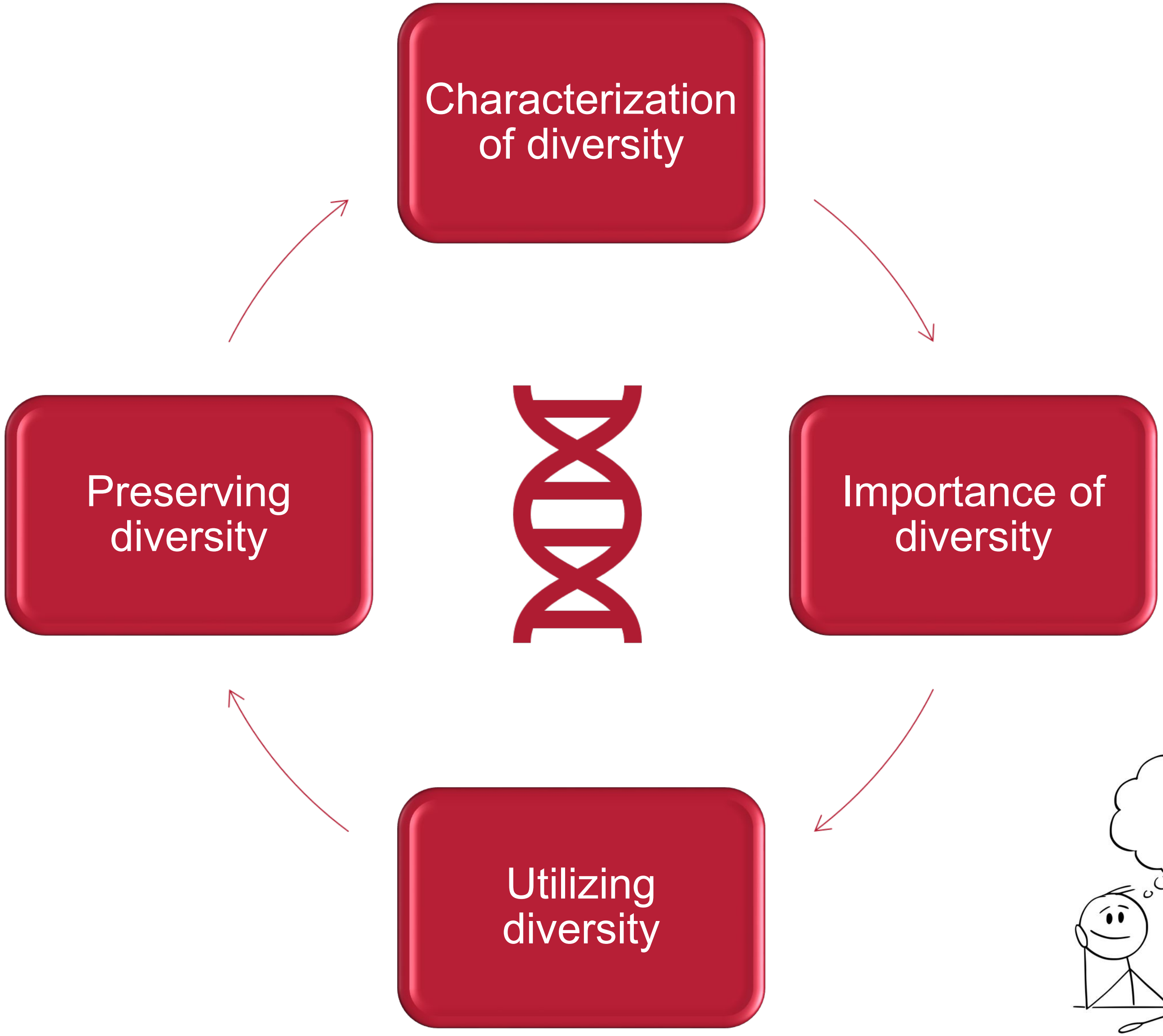
AMERICAN DAIRY SCIENCE ASSOCIATION®

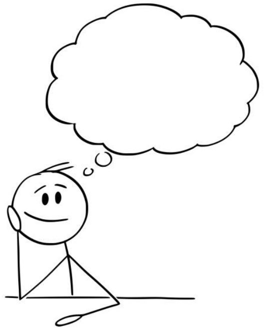


Characterization of diversity

Importance of diversity

Utilizing diversity

Preserving diversity



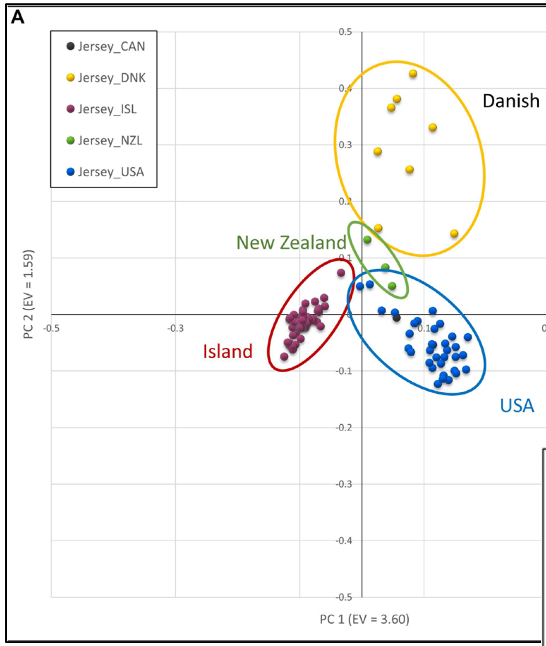
- Exceptionally good at characterizing diversity and identifying regions of the genome that are important for production
- Reasonably good at utilizing diversity in breeding programs but likely have opportunities to improve efficiency and use of data in breeding programs
- Increasing awareness of preserving diversity but often a lack of structured plans with long-term sustainability. Challenge in preserving indigenous diversity balanced with increasing production demands.

Interpretation of Results

- Different perspectives may lead to different interpretations
- Data use provides varying results → varying interpretations

Considerations when characterizing diversity

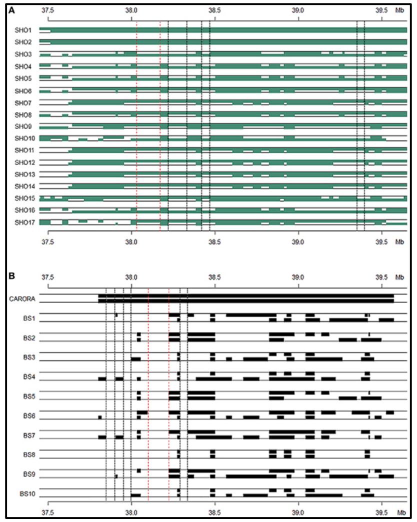
Within breed population structure and admixture

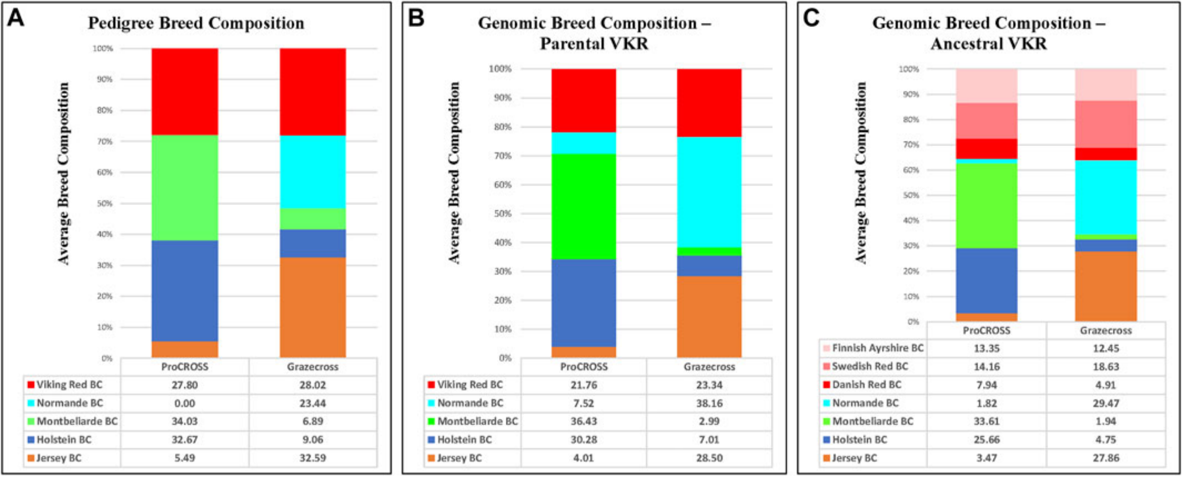


Huson, H.J. *et al.* 2020
Frontiers in Genetics

Data and reference population choice

Huson, H.J. *et al.* 2014
Frontiers in Genetics





	ProCROSS	Grazecross
🟥 Viking Red BC	27.80	28.02
🟦 Normande BC	0.00	23.44
🟩 Montbeliarde BC	34.03	6.89
🟦 Holstein BC	32.67	9.06
🟧 Jersey BC	5.49	32.59

	ProCROSS	Grazecross
🟥 Viking Red BC	21.76	23.34
🟦 Normande BC	7.52	38.16
🟩 Montbeliarde BC	36.43	2.99
🟦 Holstein BC	30.28	7.01
🟧 Jersey BC	4.01	28.50

	ProCROSS	Grazecross
🟥 Finnish Ayrshire BC	13.35	12.45
🟥 Swedish Red BC	14.16	18.63
🟥 Danish Red BC	7.94	4.91
🟦 Normande BC	1.82	29.47
🟩 Montbeliarde BC	33.61	1.94
🟦 Holstein BC	25.66	4.75
🟧 Jersey BC	3.47	27.86

Jaafar, M.A. *et al.* 2022
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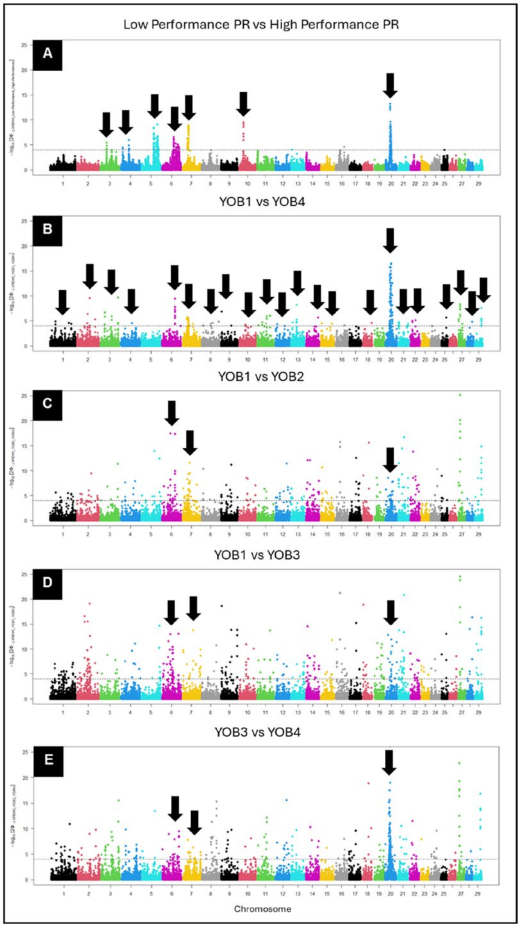
Strategic choice of population use and comparison



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Animals

Take home – Characterizing diversity

- How will your choice of animals effect your analysis and results?
- Are you creating bias in your results due to the animal or analysis choice?

Importance of genetic diversity

Indigenous cattle

The Pan African Review

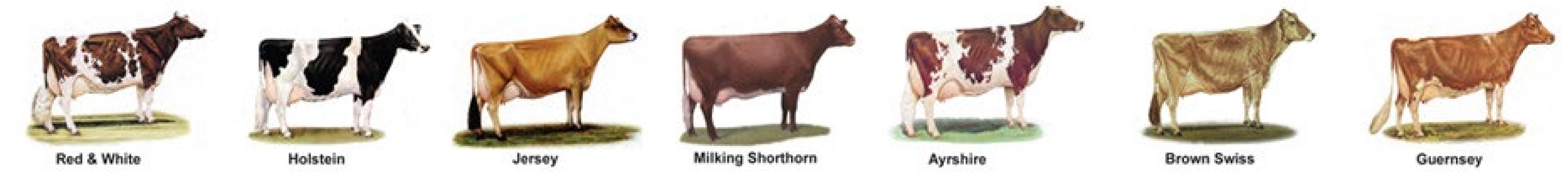
The Conscientious Perspective

The disappearance of Africa's indigenous cattle and the dangers of a single breed

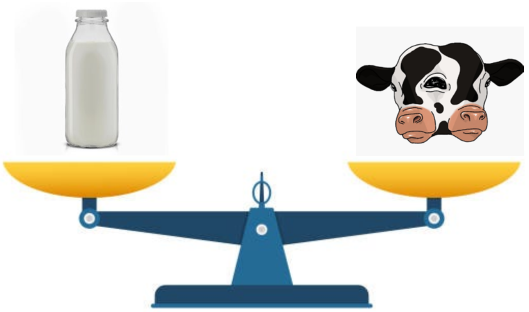
"Regrettably, Africa's drive towards increased food security has mainly focused on the single-story mindset approach of the West." by Chika Esiobu

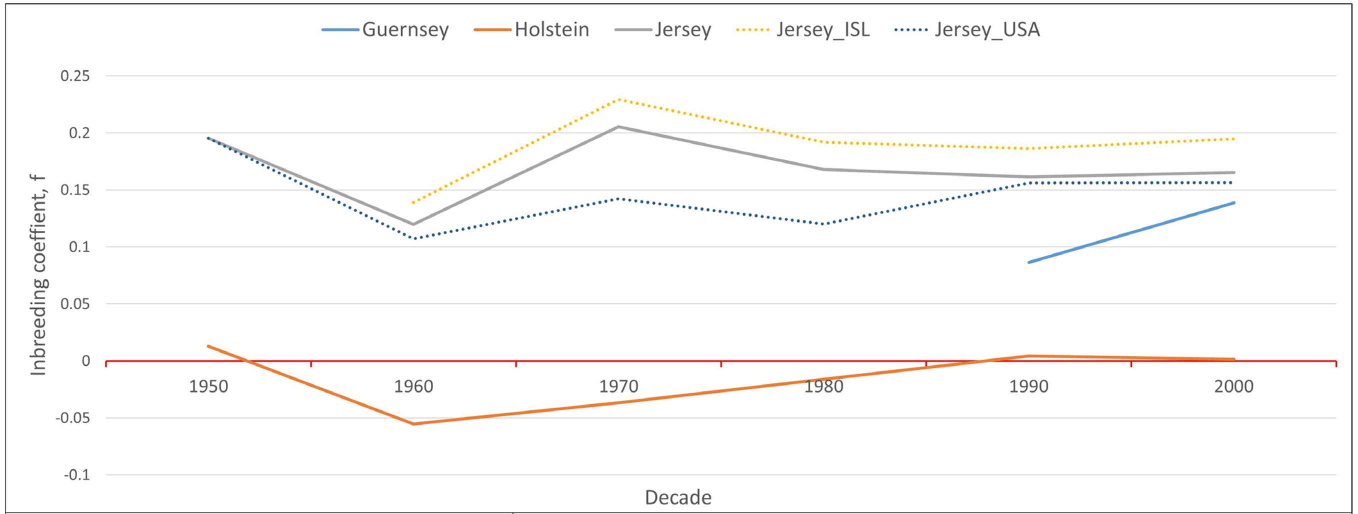
Importance of genetic diversity

Commercialized dairy breeds



Rutgers University – 4-H Animal Science Resource Blog





# individuals decade	JE (n=1) JE_ISL (n=0) JE_USA (n=1) HO (n=7)	JE (n=5) JE_ISL (n=2) JE_USA (n=3) HO (n=3)	JE (n=11) JE_ISL (n=8) JE_USA (n=3) HO (n=7)	JE (n=18) JE_ISL (n=10) JE_USA (n=5) HO (n=8)	JE (n=45) JE_ISL (n=21) JE_USA (n=18) HO (n=32)	JE (n=17) JE_ISL (n=8) JE_USA (n=5) HO (n=6)

Huson, H.J. *et al.* 2020
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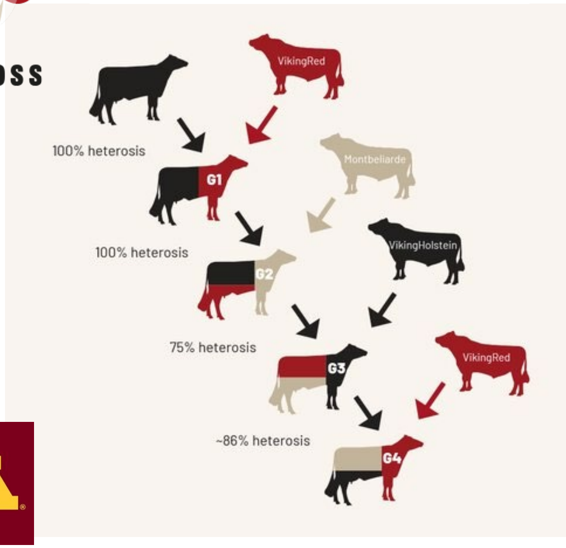
Functional importance of diversity

Breed influence

PROCROSS

VIKINGGENETICS®

breeding for what truly matters



Ancestry is significantly associated with performance

- Milk, fat, and protein yield – HOL & MON

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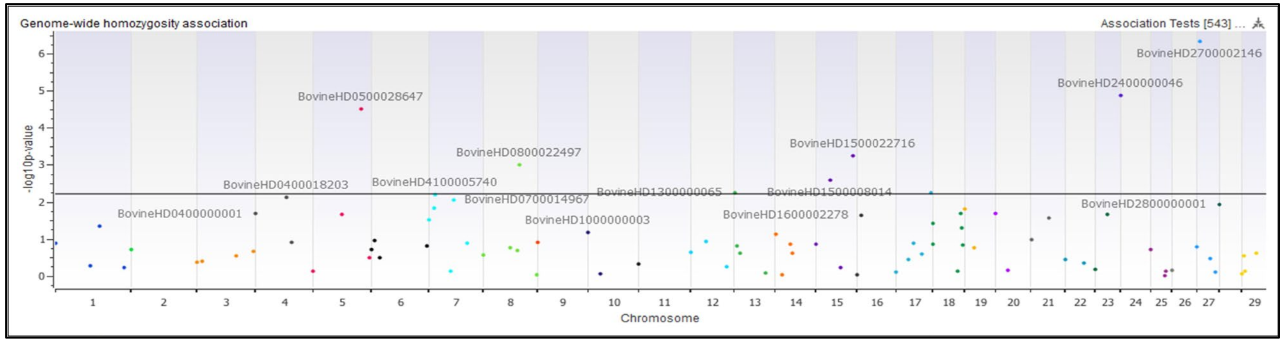
Signatures of selection

Runs of homozygosity within Isle or non-Isle Jersey cattle



Chromosome	P-value[1]	Population[2]	Panther Pathway3
5	4.01E-03	Island	Cholesterol metabolic processes
5	3.03E-05	Island	Complement activation; Blood coagulation
7	8.68E-04	Island	Anion Transport
8	8.56E-04	Island	Cell adhesion; Biological adhesion
13	6.15E-03	Island	Protein complex assembly
15	2.80E-03	Island	Blood circulation
15	1.51E-03	Non-Island	Fatty acid metabolic process
24	5.49E-06	Island	Sensory perception of sound
27	4.43E-07	Island	Lipid metabolic process
29	3.77E-03	Non-Island	Amino acid transport

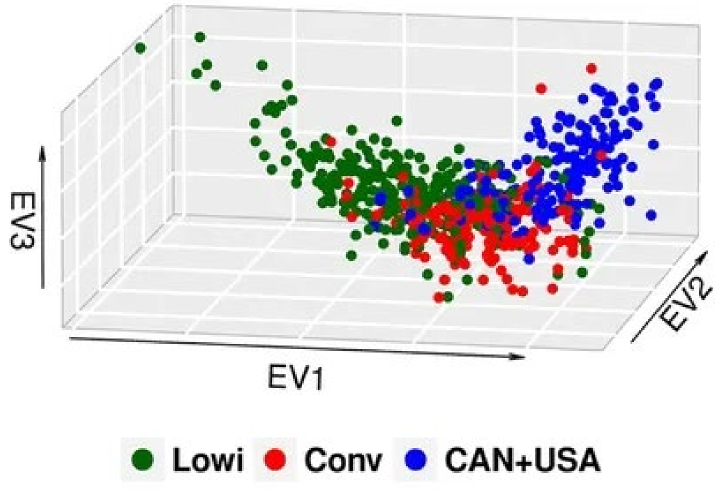
Huson, H.J. *et al.* 2020
Frontiers in Genetics

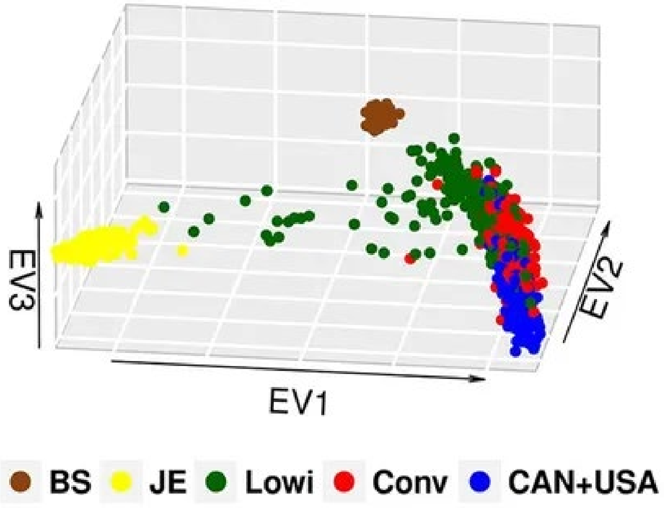
Take home – Importance of diversity

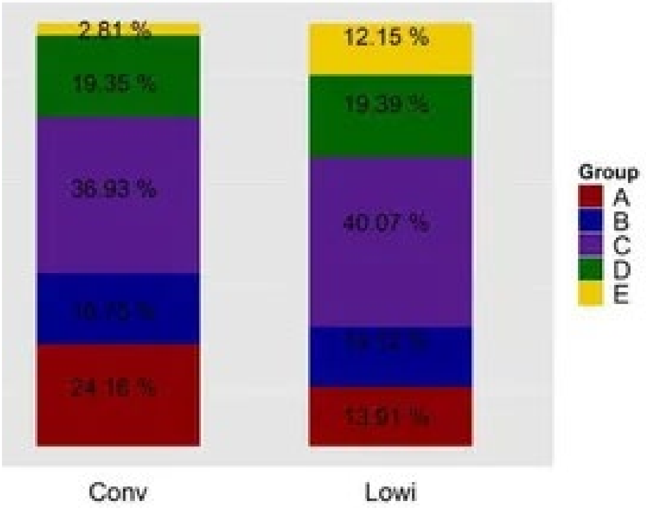
- Indigenous breeds
  - Establishing value of indigenous breeds
  - Intense natural selection & adaptation
  - Minimal commercial selection for production
- Commercialized dairy breeds
  - Intense commercial / production selection → Value recognized
  - High degree of breed uniformity → loss of genetic diversity
  - Preservation of genetic diversity ≠ capture of genetic progress
- Functional importance
  - Breed influence
  - Identification of genomic regions / genes associated with traits of interest

Utilizing diversity

You must understand your diversity before you can properly utilize it….







Garcia-Ruiz *et al.* 2015
Frontiers in Genetics

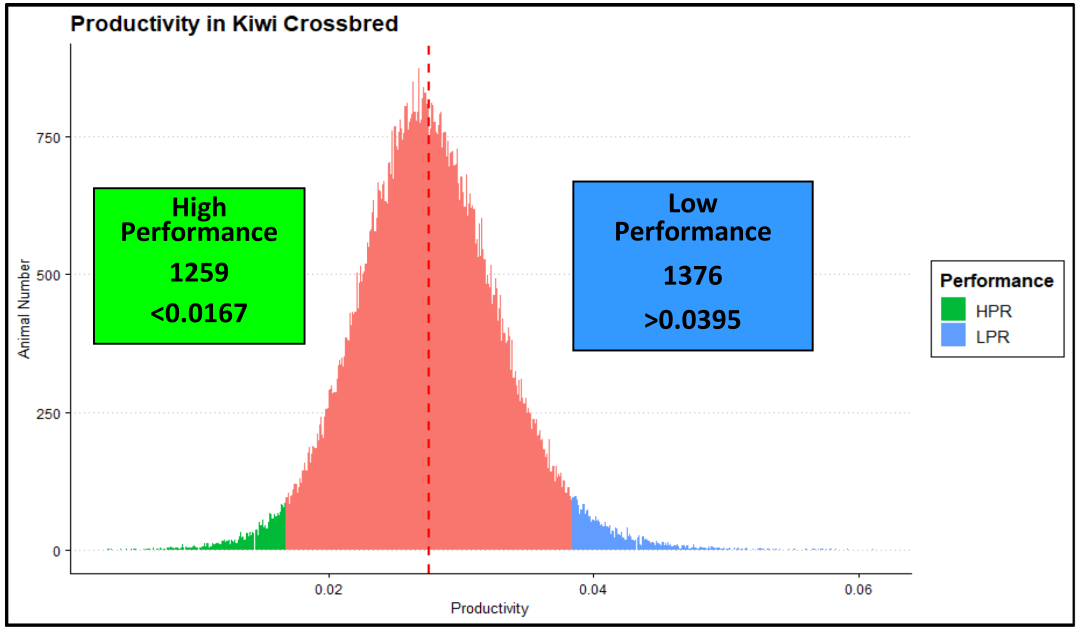
Understanding population structure gives insight into practical application → will the inclusion of U.S. & Canadian Holstein improve genetic prediction in Mexican Holstein?

Utilizing diversity

Relationship between phenotypic variation and genetic variation… it's complicated

KiwiCross®

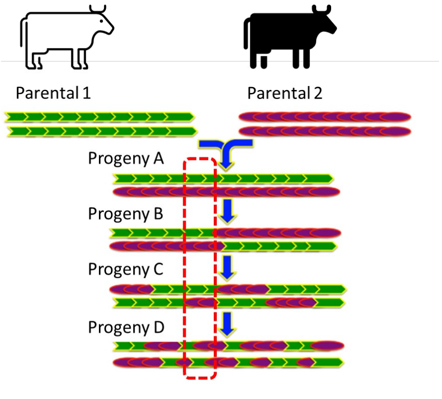
$$Production\ efficiency\ (PR) = milk\ solids \div mature\ weight$$



Challenge

- Significant association of global breed composition variation to traits…. BUT
    - Variation in breed composition is often minimal (ie. <10%)
    - Variation within performance group is substantial…. So that individuals in both high and low performance groups may have up to 90% HO or only 10% HO….

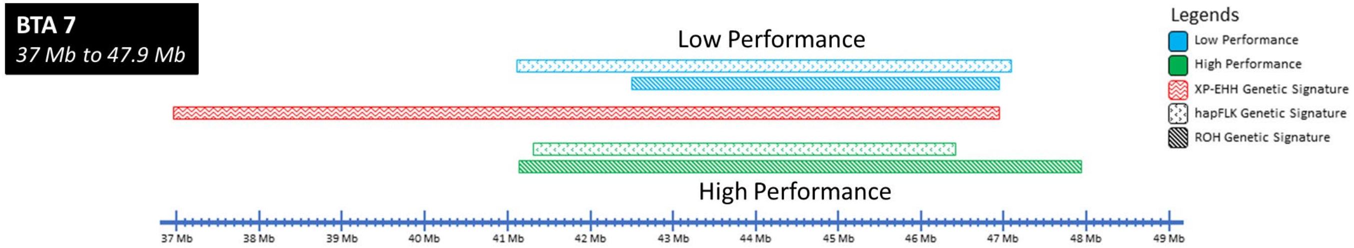
Hypothesis: Variation in chromosomal localization of ancestry influences trait → only specific regions of ancestral selection are important



Utilizing diversity

Localizing ancestry to chromosomal regions provides insight into performance and an opportunity for more targeted selection….

LIC KiwiCross®



Nine functionally annotated genes in *Bos taurus*: ***ABCA7, CLK4, GDF9, MADCAM1, NLRP3, PROP1, RMND5B, SLC25A48*** **and** ***SLC34A1*** **.**

<u>Genes have been associated with 3 trait types:</u>

- Conformation (5 genes – *CLK4, MADCAM1, NLRP3, RMND5B, SLC34A1*),
- Performance (Milk/Production) (3 genes – *ABCA7, PROP1, RMND5B*),
- Reproduction (4 genes – *GDF9, PROP1, RMND5B, SLC25A48*).

CattleQTLdb (Release 49); Wu et al., 1998; Pan et al., 2013; Honerlagen et al., 2021

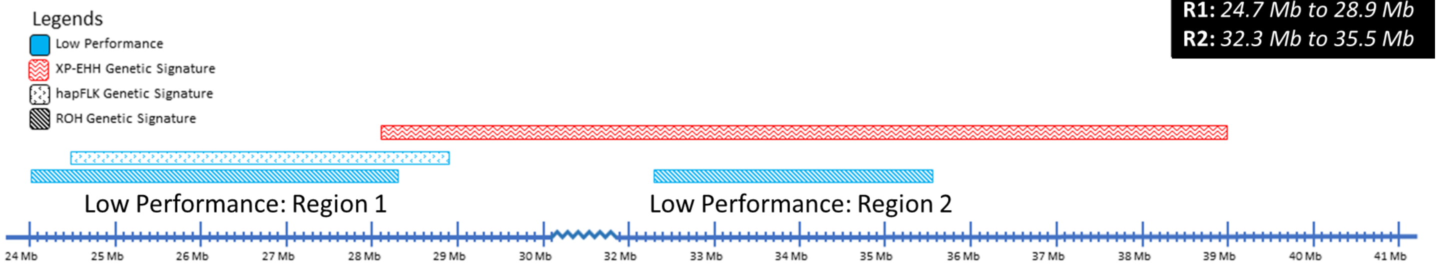
Selection for differing haplotypes derived from Holstein-Friesian ancestry at the same region on BTA 7 for both low- and high-performance efficiency cattle

Jaafar, M.A. *et al.* 2025
Animals

Utilizing diversity

Localizing ancestry to chromosomal regions provides insight into performance and an opportunity for more targeted selection….

LIC KiwiCross®



BTA 20
R1: *24.7 Mb to 28.9 Mb*
R2: *32.3 Mb to 35.5 Mb*

12 functionally annotated genes in *Bos Taurus*
Region 1 : ***ARL15, FST, ITGA1, NDUFD4, PARP8***, **and** ***PELO***
Region 2 : ***C6, C7, DAB2, PLCXD3, RICTOR*** **and** ***TTC33***

Genes have been associated with 4 trait types:

- Performance (Milk/Production) (11 genes – *C6, C7, DAB2, FST, ITGA1, NDUFD4, PARP8, PELO, PLCXD3, RICTOR and TTC33*),
- Reproduction (1 gene – *FST*).
- Meat (3 genes – *ARL15, FST and RICTOR*)
- Conformation (3 genes – *RICTOR, PLCXD3* and *TTC33)*

R1: *24.7 Mb to 28.9 Mb*

R2: *32.3 Mb to 35.5 Mb*

Holstein-Friesian ancestry haplotype is associated with low performance efficiency in region 1 while Jersey ancestry is associated with region 2

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KiwiCross® = Holstein x Jersey

ProCross = Holstein x
Montbeliarde x Viking Red

Identifying common localized Holstein ancestry selection across different crossbred dairy cattle populations

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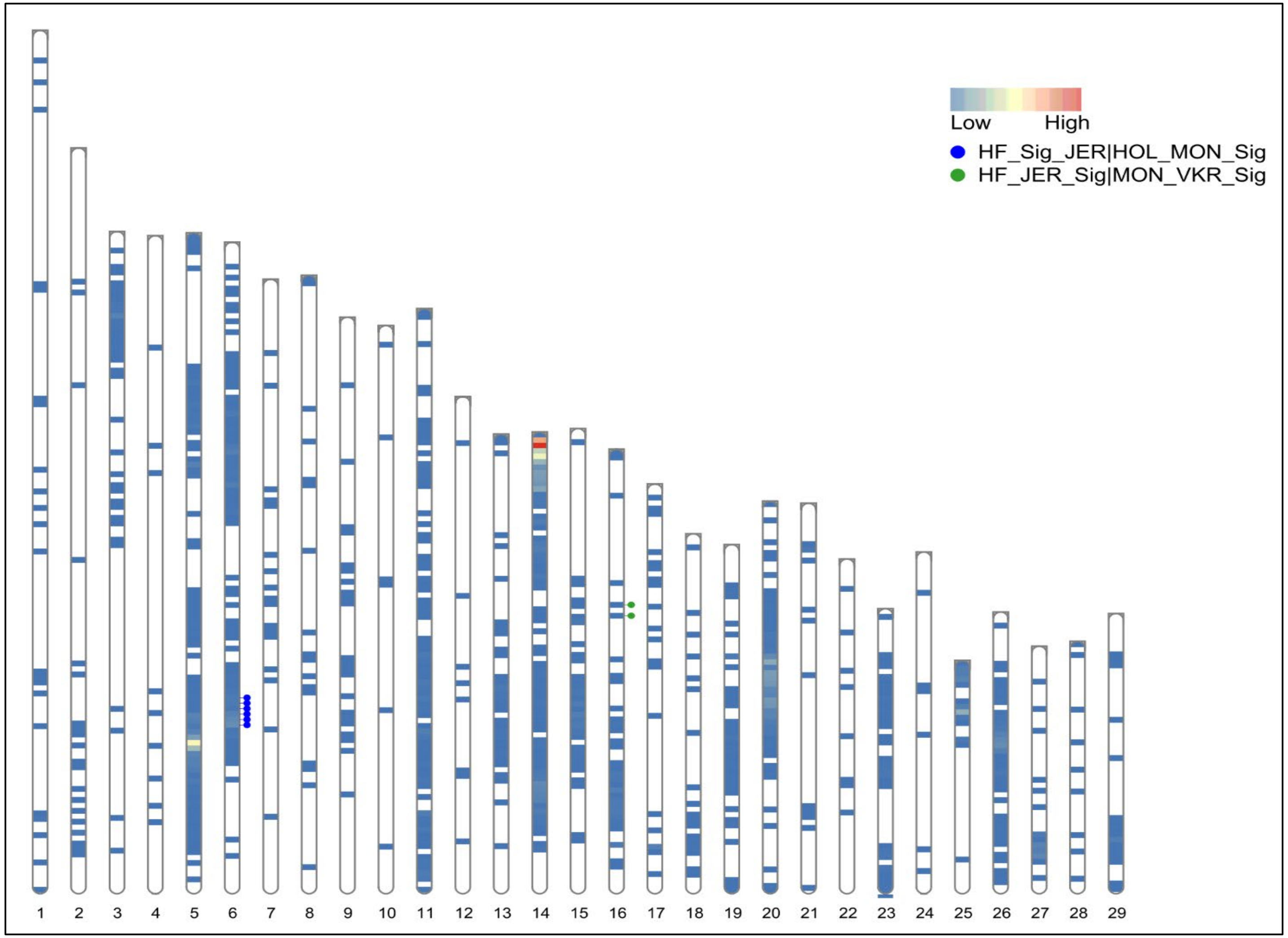
ProCROSS and KiwiCross® high-performance progeny showed common haplotypes descending from HOL/HF ancestors across their genome.



- **8 overlapping segments**
  - 6 on BTA6
  - 2 on BTA16
- **BTA 6 (83 Mb to 89 Mb)**
  - Holstein ancestry important in both crosses, likely associated with production traits
- **BTA 16 (28 Mb to 29 Mb and 30 Mb to 31 Mb)**
  - Holstein was disadvantageous for ProCross but advantageous for KiwiCross®

Unpublished
Chapter 4: Assessing Local Ancestry Inference Effect on Two Hierarchical Distinct Crossbreeding Systems
Jaafar, M.J., Harris, B.L., Heins, B.J., Dechow, C., Huson, H.J.

Preserving diversity


A limited number of Y chromosome lineages is present in North American Holsteins

Xiang-Peng Yue *†, Chad Dechow *, Wan-Sheng Liu *

Journal of Dairy Science
Official Publication of the American Dairy Science Association

Neptune H
1880

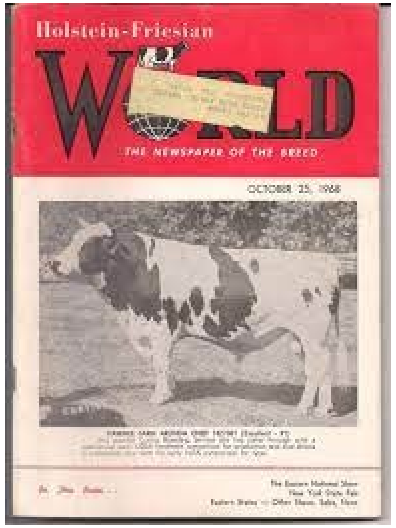
Round Oak Rag Apple Elevation

Hulleman
1881

Pawnee Farm Arlinda Chief

Penn State Ivanhoe Star

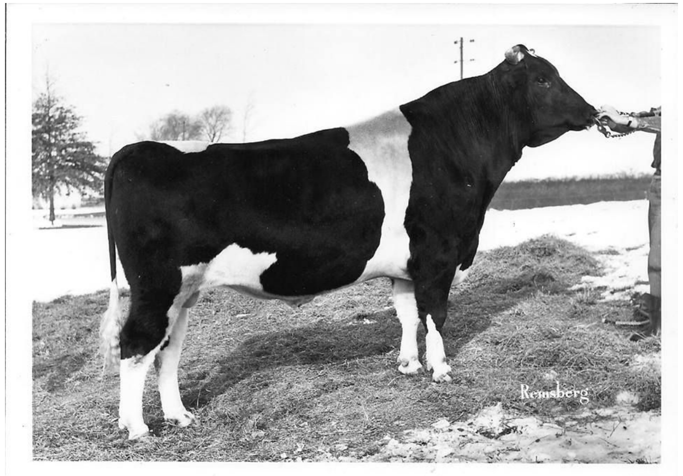
51% of sire born in ≤ 2010 trace back to Elevation


Holstein-Friesian WORLD

THE NEWSPAPER OF THE BREED

OCTOBER 25, 1966

49% of sire born in ≤ 2010 trace back to Chief

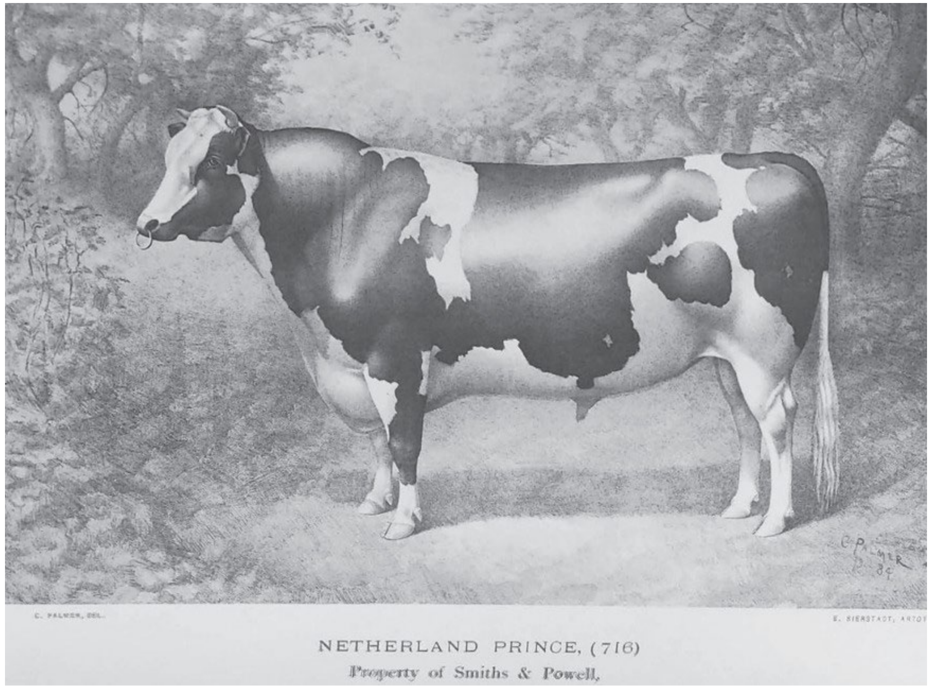


<1% of sire born in ≤ 2010 trace back to Ivanhoe (carrier of BLAD & CVM)

Recreating the lines of 2 additional sires in existence in the 1960's

Netherland Prince
1880

Netherland Jacob
1880



NETHERLAND PRINCE, (716)
Property of Smiths & Powell,

Recovering lost genetic diversity in Holsteins is focus of professors' research

https://www.psu.edu/news/research/story/recovering-lost-genetic-diversity-holsteins-focus-professors-research/

Holsteins born at Penn State to improve genetic diversity are 'udderly' amazing

https://www.psu.edu/news/research/story/holsteins-born-penn-state-improve-genetic-diversity-are-udderly-amazing/

Preserving diversity

Assessment of genetic diversity, inbreeding and collection completeness of Jersey bulls in the US National Animal Germplasm Program

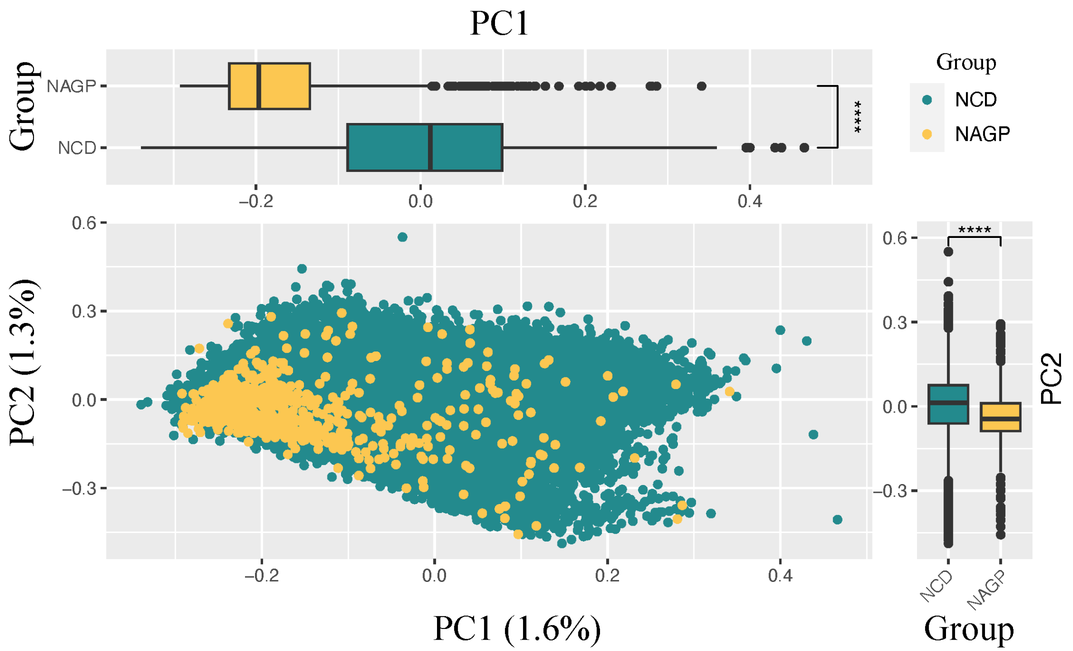
USDA

National Animal Germplasm Program
Established in 1999

“Provide genetic security and increase genetic understanding of U.S. livestock”

Voluntary contribution of samples by AI companies, individual breeders, and university research collections

USJersey

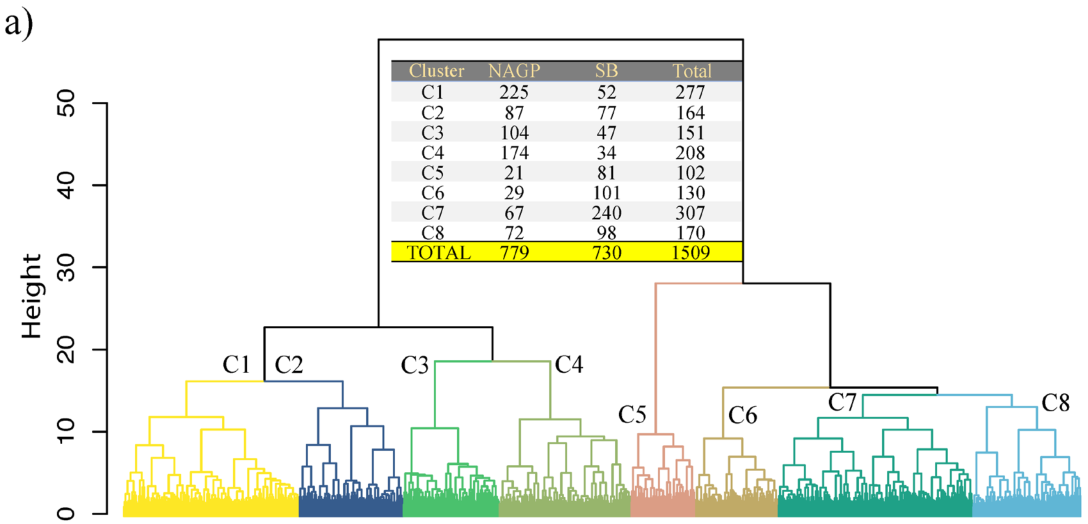


	NCD	NAGP	Total
1950-1989	362	146	508
1990-1999	1452	243	1695
2000-2009	2416	213	2629
2010-2019	29380	136	29516
2020-	7150	41	7191
Total	**40,760**	**779**	**41,539**

Objectives

1. Compare genomic data of Jersey bulls in the NAGP collection with Jersey bulls in the CDCB database, to ascertain their inbreeding status, relatedness, sample origin and genetic diversity.
2. Assess collection completeness and identify collection gaps.
3. Ascertain the usefulness of using Ward’s clustering for identifying bulls for collection.

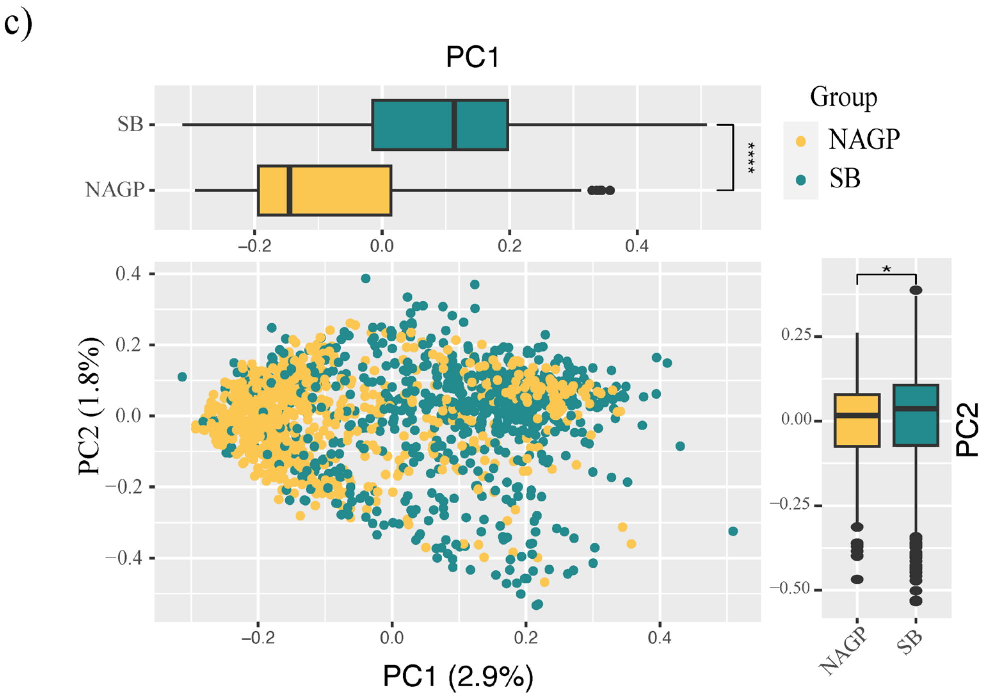
1000 Popular Sires of Bulls

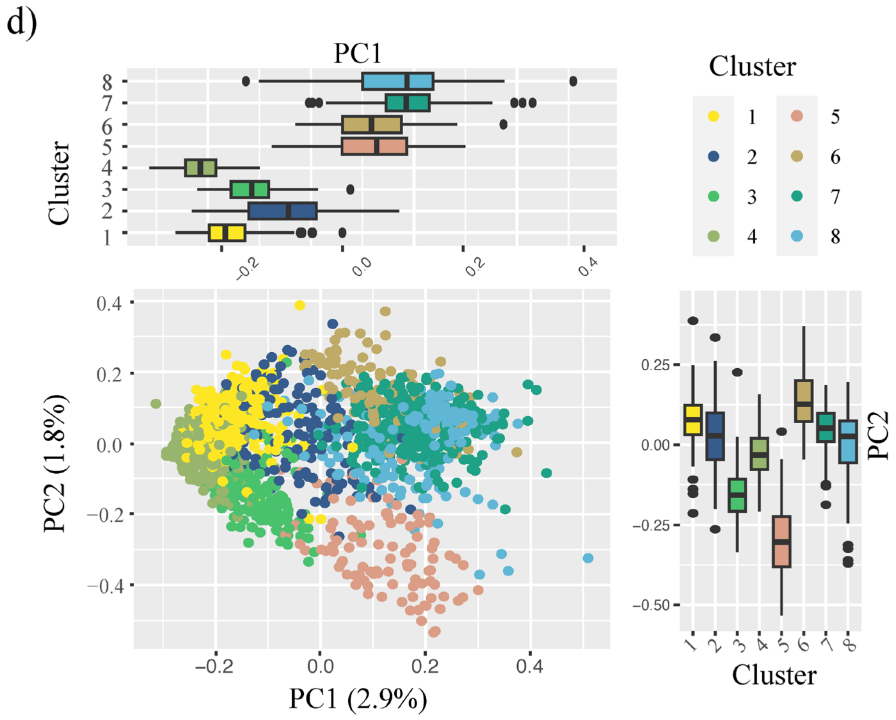


Cluster	NAGP	SB	Total
C1	225	52	277
C2	87	77	164
C3	104	47	151
C4	174	34	208
C5	21	81	102
C6	29	101	130
C7	67	240	307
C8	72	98	170
TOTAL	779	730	1509

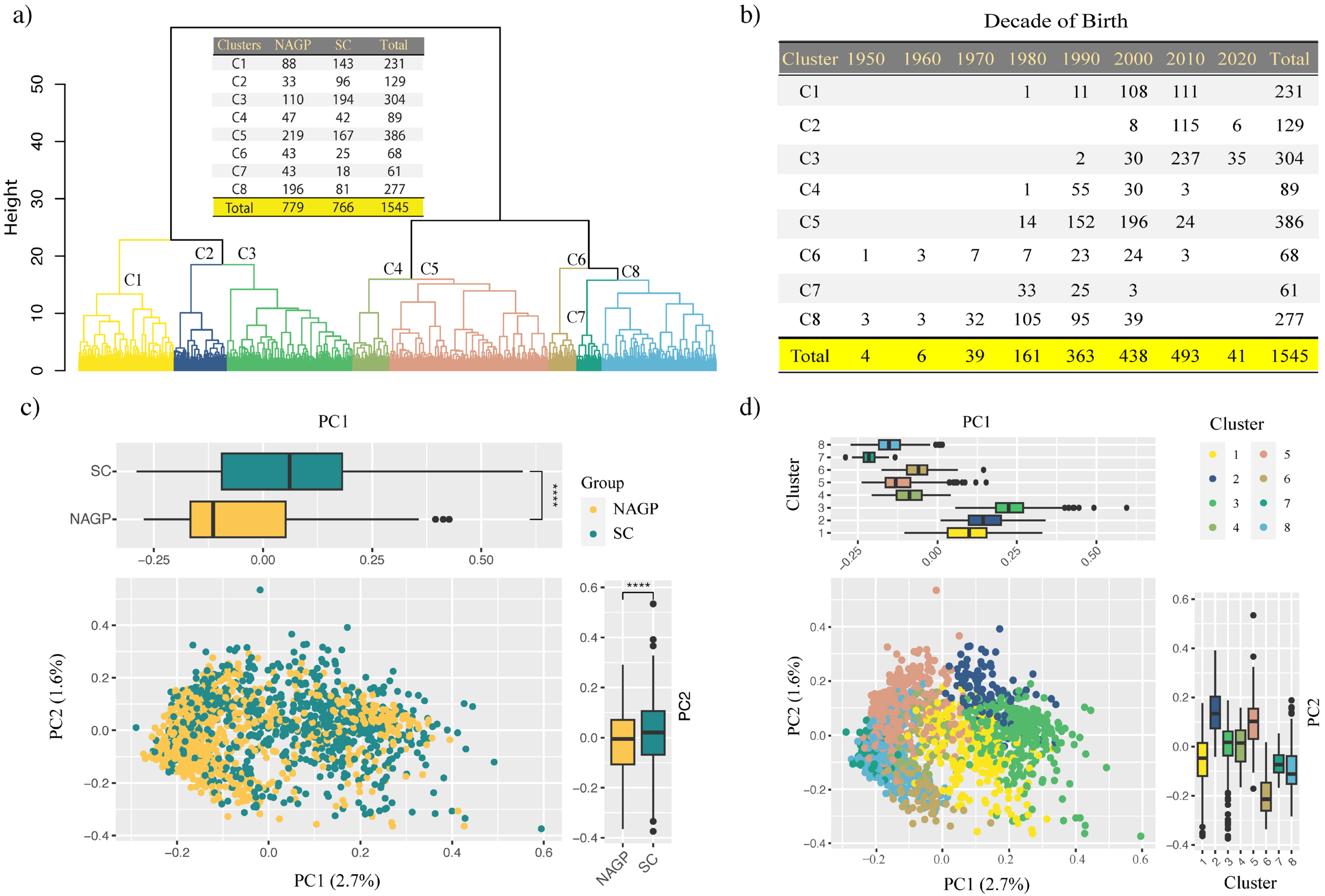
b)

	Decade of Birth								
Cluster	1950	1960	1970	1980	1990	2000	2010	2020	Total
C1				14	133	126	4		277
C2				1	46	78	39		164
C3	4	6	29	49	28	27	8		151
C4			6	75	100	27			208
C5				8	27	36	31		102
C6						5	125		130
C7						4	290	13	307
C8					1	25	115	29	170
Total	**4**	**6**	**35**	**147**	**335**	**328**	**612**	**42**	**1509**





1000 Popular Sires of Cows



a)

Clusters	NAGP	SC	Total
C1	88	143	231
C2	33	96	129
C3	110	194	304
C4	47	42	89
C5	219	167	386
C6	43	25	68
C7	43	18	61
C8	196	81	277
Total	779	766	1545

b)

Decade of Birth

Cluster	1950	1960	1970	1980	1990	2000	2010	2020	Total
C1				1	11	108	111		231
C2						8	115	6	129
C3					2	30	237	35	304
C4				1	55	30	3		89
C5				14	152	196	24		386
C6	1	3	7	7	23	24	3		68
C7				33	25	3			61
C8	3	3	32	105	95	39			277
Total	4	6	39	161	363	438	493	41	1545

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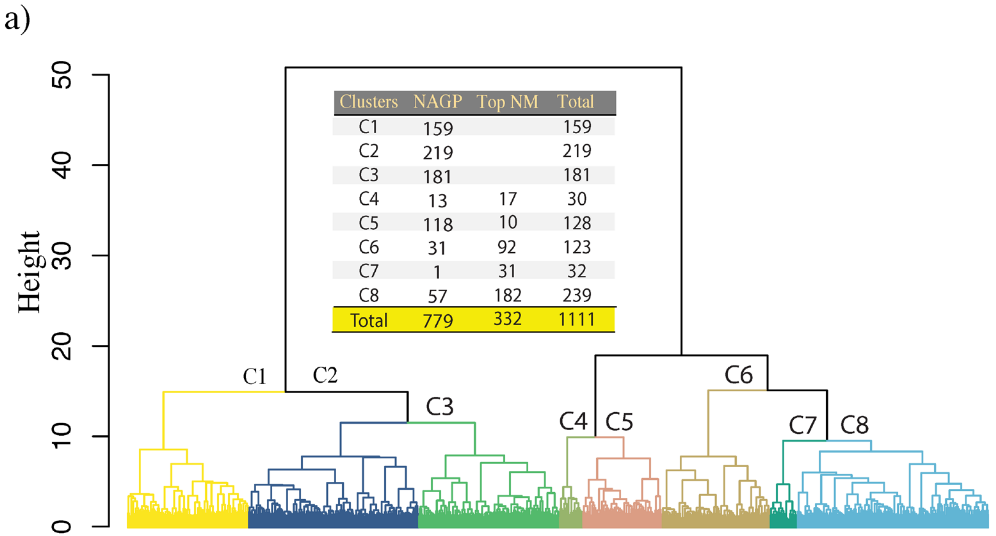
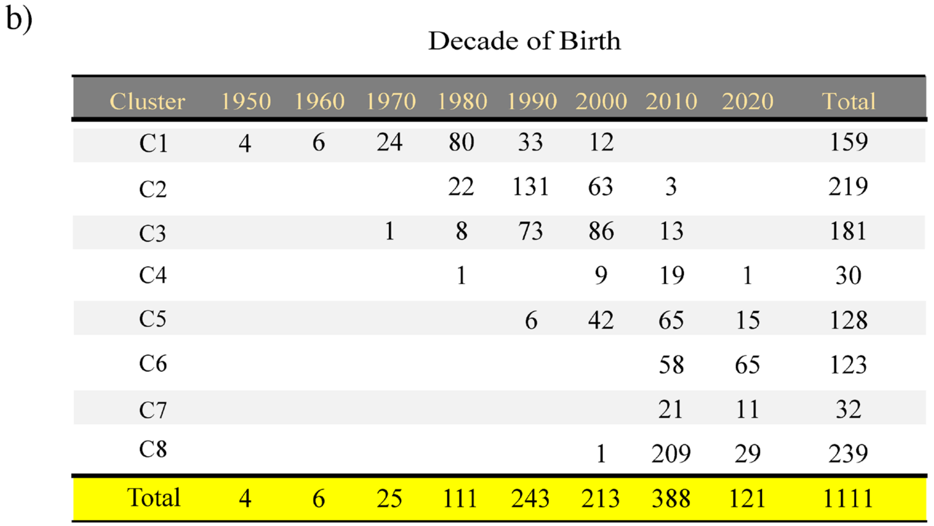
Top 1% of Net Merit

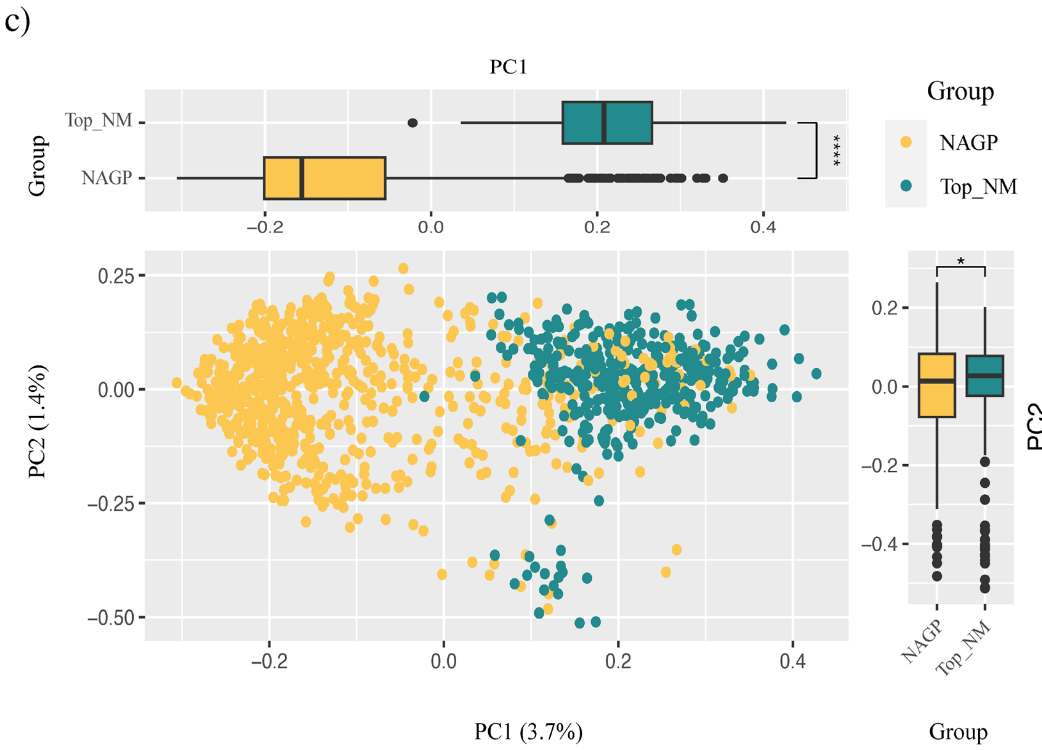


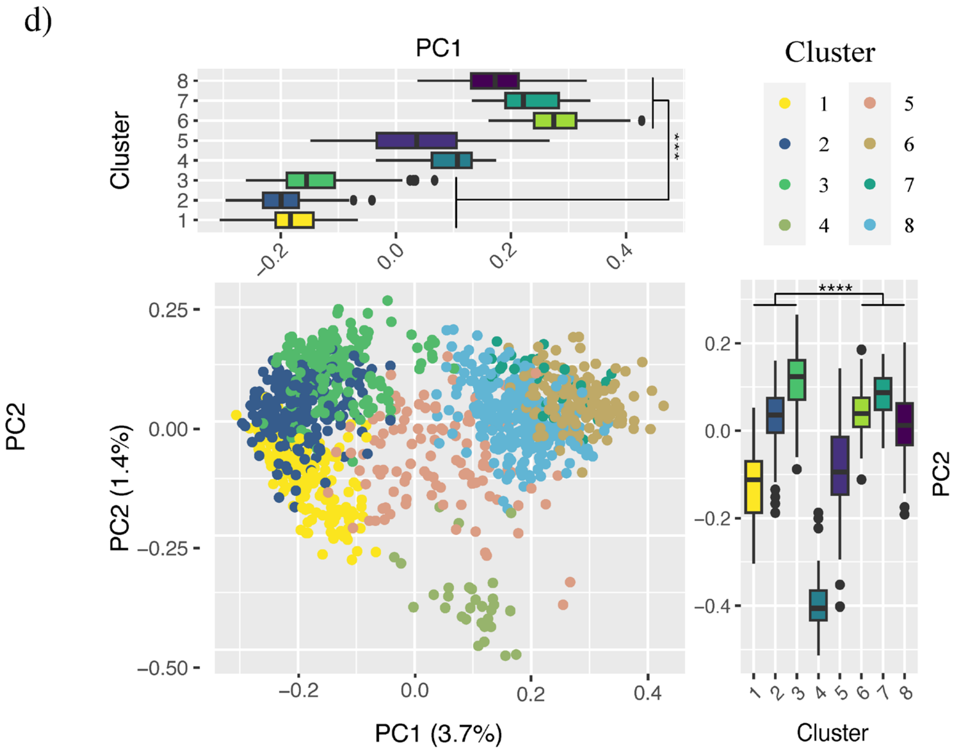
Clusters	NAGP	Top NM	Total
C1	159		159
C2	219		219
C3	181		181
C4	13	17	30
C5	118	10	128
C6	31	92	123
C7	1	31	32
C8	57	182	239
Total	779	332	1111



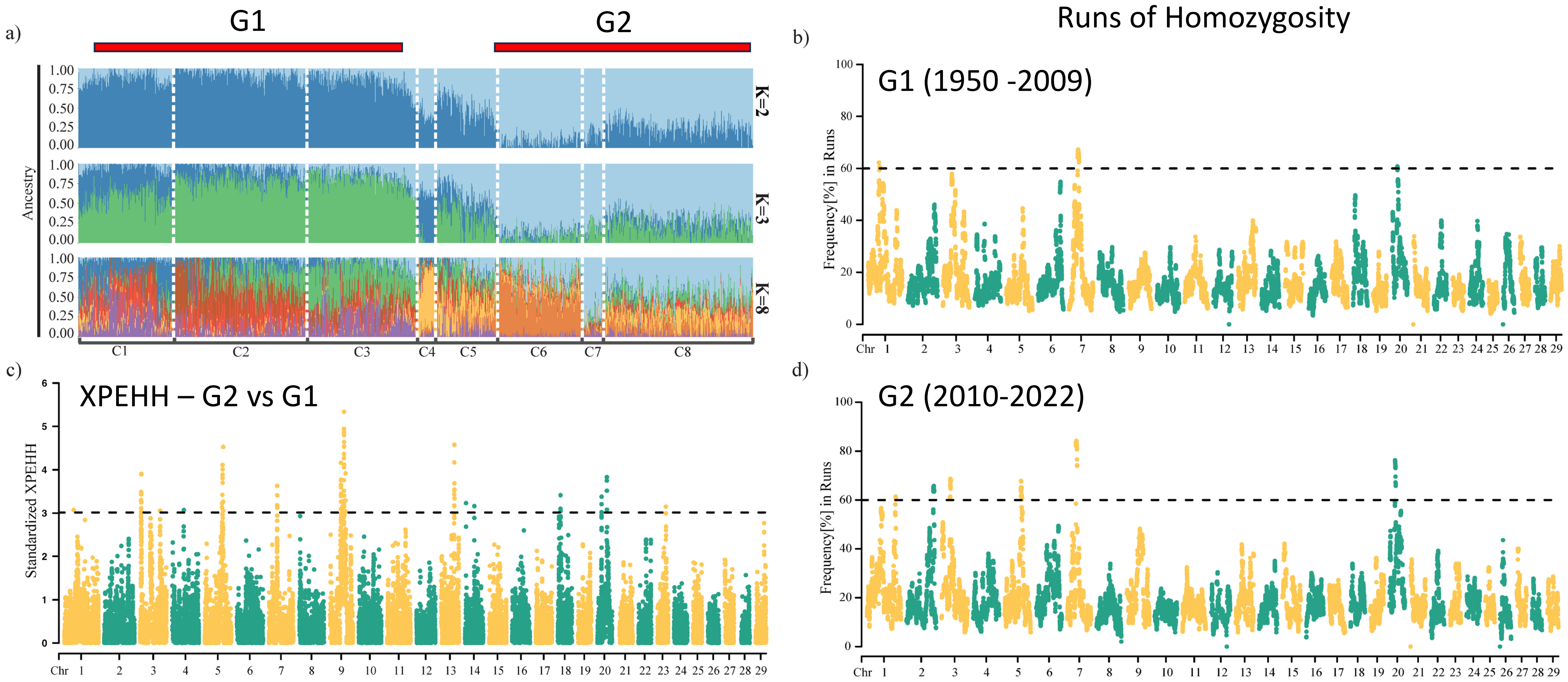
Decade of Birth

Cluster	1950	1960	1970	1980	1990	2000	2010	2020	Total
C1	4	6	24	80	33	12			159
C2				22	131	63	3		219
C3			1	8	73	86	13		181
C4				1		9	19	1	30
C5					6	42	65	15	128
C6							58	65	123
C7							21	11	32
C8						1	209	29	239
Total	4	6	25	111	243	213	388	121	1111

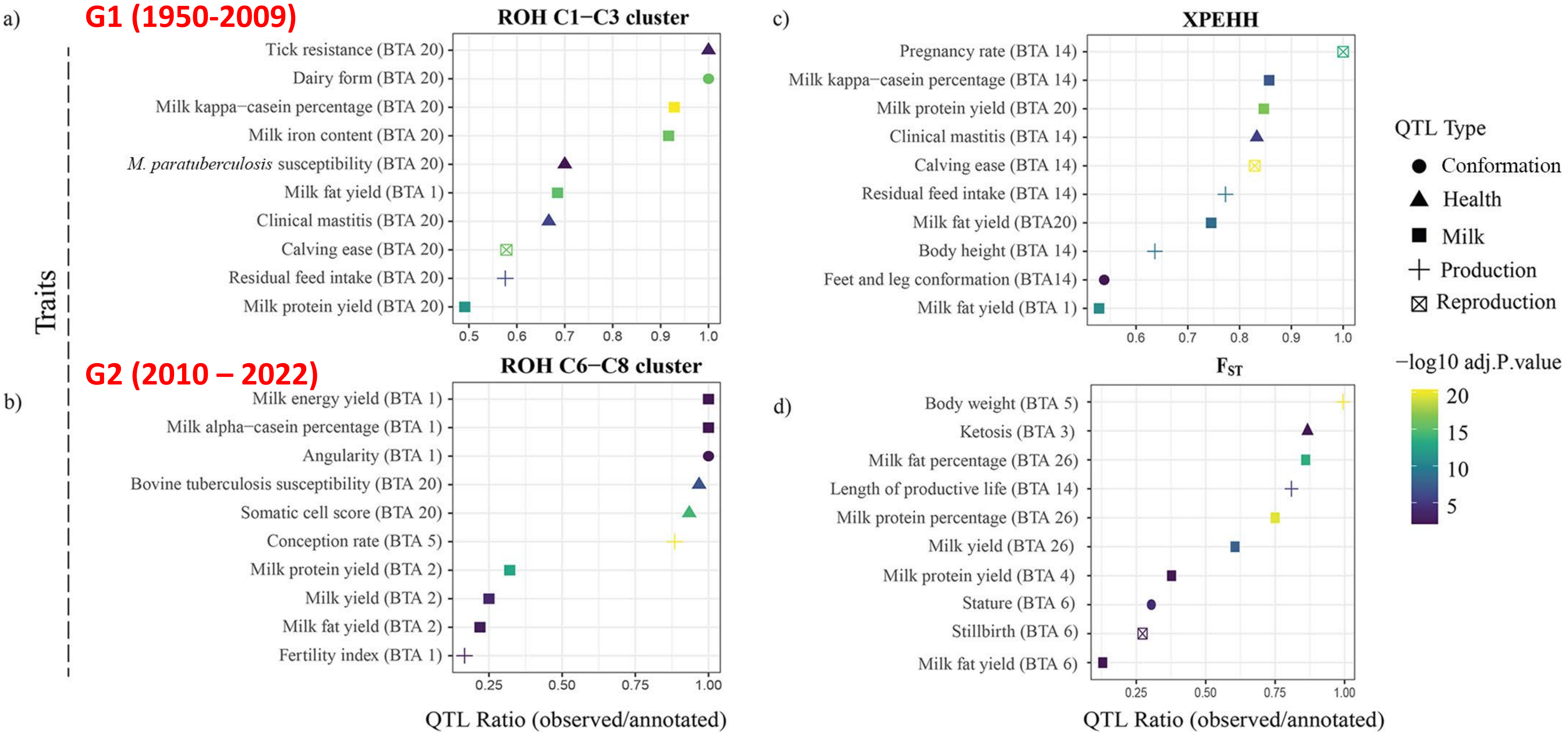




Population Structure and Selection Signatures



Functional Importance - QTL Enrichment Analysis


G1 (1950-2009)

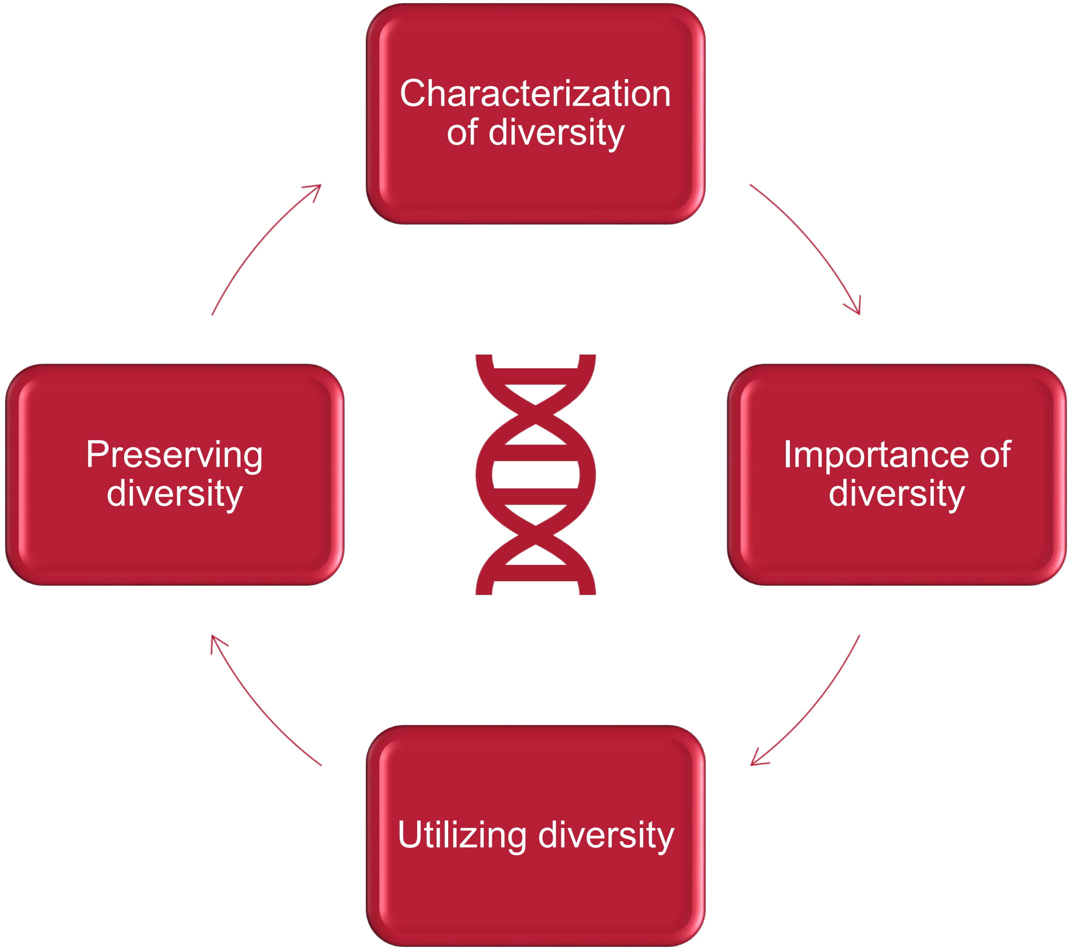
G2 (2010 – 2022)

Research Conclusions

- NAGP broadly captures the genetic variation in the Jersey population.
- Marginally higher genetic diversity in the collection compared to NCD.
- More recent population moving away from collection, augment collection with increased emphasis on collecting more current generation bulls.
- Continuous sampling and Ward’s method (Ped / Genomic) is effective.
- Selection for production, fertility, health and conformation.
- Encourage more AI companies / Individual breeders to contribute germplasm.

Summary

- Characterization of diversity
    - Critically evaluate your data, approach, and analyses → result interpretation
    - Foundation for understanding importance, utilization, and preservation of diversity
- Importance of diversity
    - There is value in both indigenous and highly commercialized breeds
    - Diversity provides biological plasticity
    - Compromise between genetic gain and inbreeding
- Utilizing diversity
    - Continuous opportunities to improve efficiency and accuracy of breeding programs and genetic selection to utilize the immense genetic diversity within cattle
- Preserving diversity
    - Identify, plan, and take action!



Discussion

- Do we need and how would we pursue strategies to integrate historic genetic diversity into current population? (ie. Holstein lineages)
- How do we support the initiation of new programs and augmenting of existing germplasm preservation programs?
- Should we be more active in evaluating and implement strategies to keep, maintain, increase diversity in our selection programs? What is currently being done to address this?
- What is currently occurring and how do we support organized and intentional selection of local cattle whether indigenous or crosses to add value? Hence, not relying on the crossing with commercialized breeds as the only avenue for genetic improvement.
- Actively plan for reassessing programs (germplasm, selection algorithms, breeding programs) and adapting strategies over time. Evaluate progress with the utilization and preservation of diversity.

Thank you!

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