# from Mendel to Genom

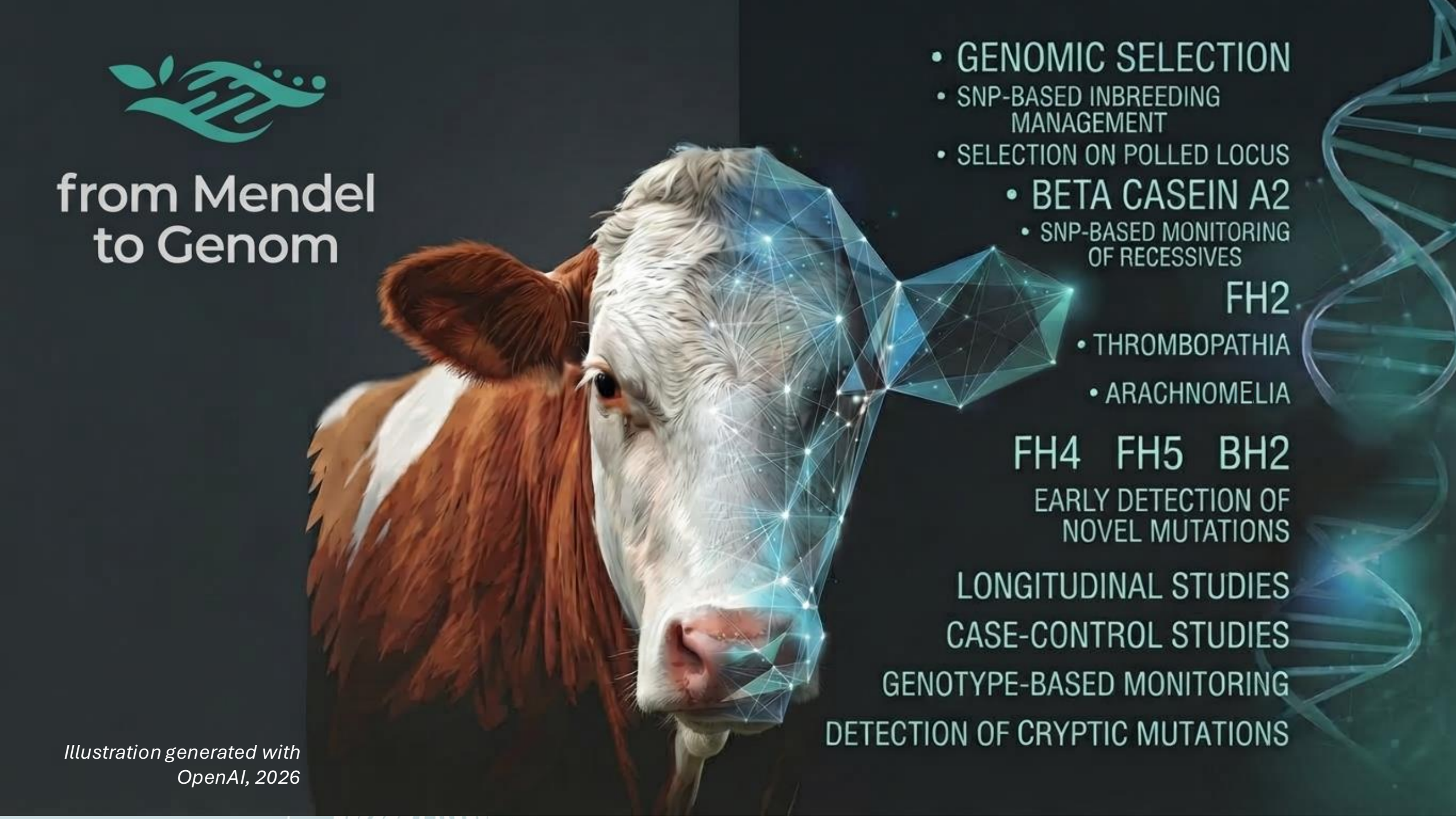



*Illustration generated with OpenAI, 2026*

# SNP-Based Monitoring of Mendelian Traits in Fleckvieh Cattle: From Genetic Defects to Beneficial Variants

*Schwarzenbacher H., Himmelbauer J., Fuerst C.*

ZuchtData EDV-Dienstleistungen GmbH, 1200 Vienna, Austria

World Simmental-Fleckvieh Congress
Brno, Czech Republic, 16th/21st September 2026

# Outline

01 Genomic toolbox to handle Mendelian traits

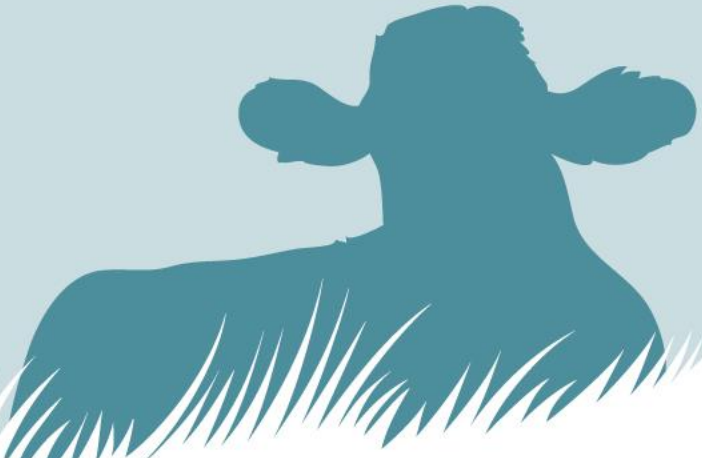
02 Desirable genetic variants

03 Unfavourable variants

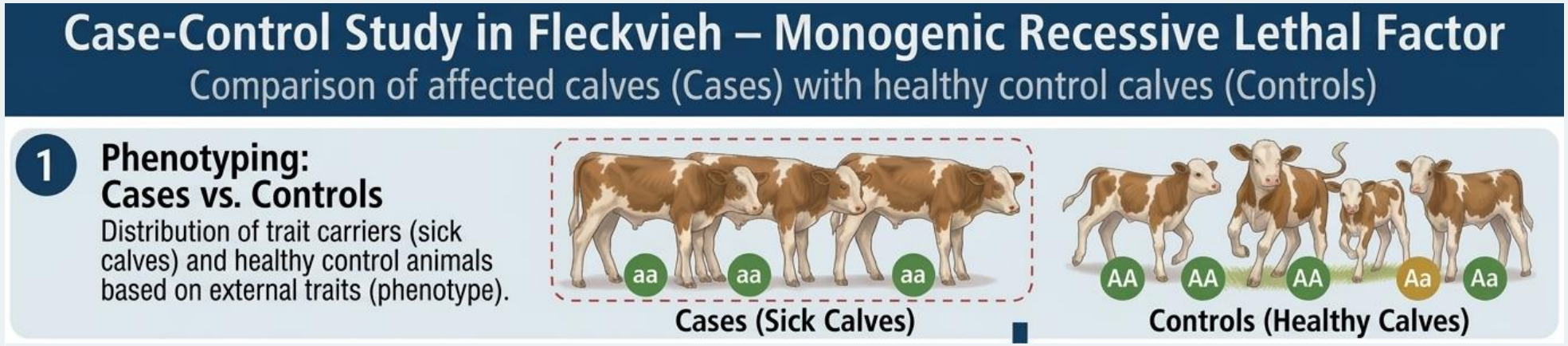
04 SNP-based monitoring

05 Take-home message

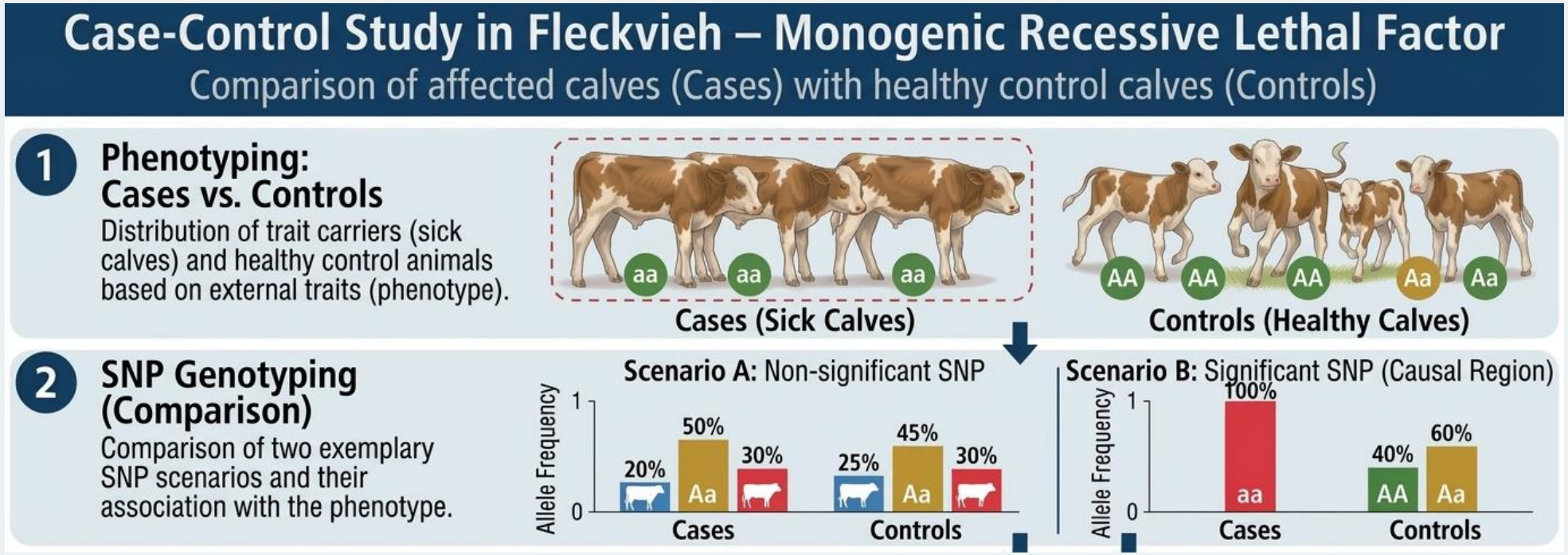
# *The ‚novel' genomic toolbox to manage genetic variants*



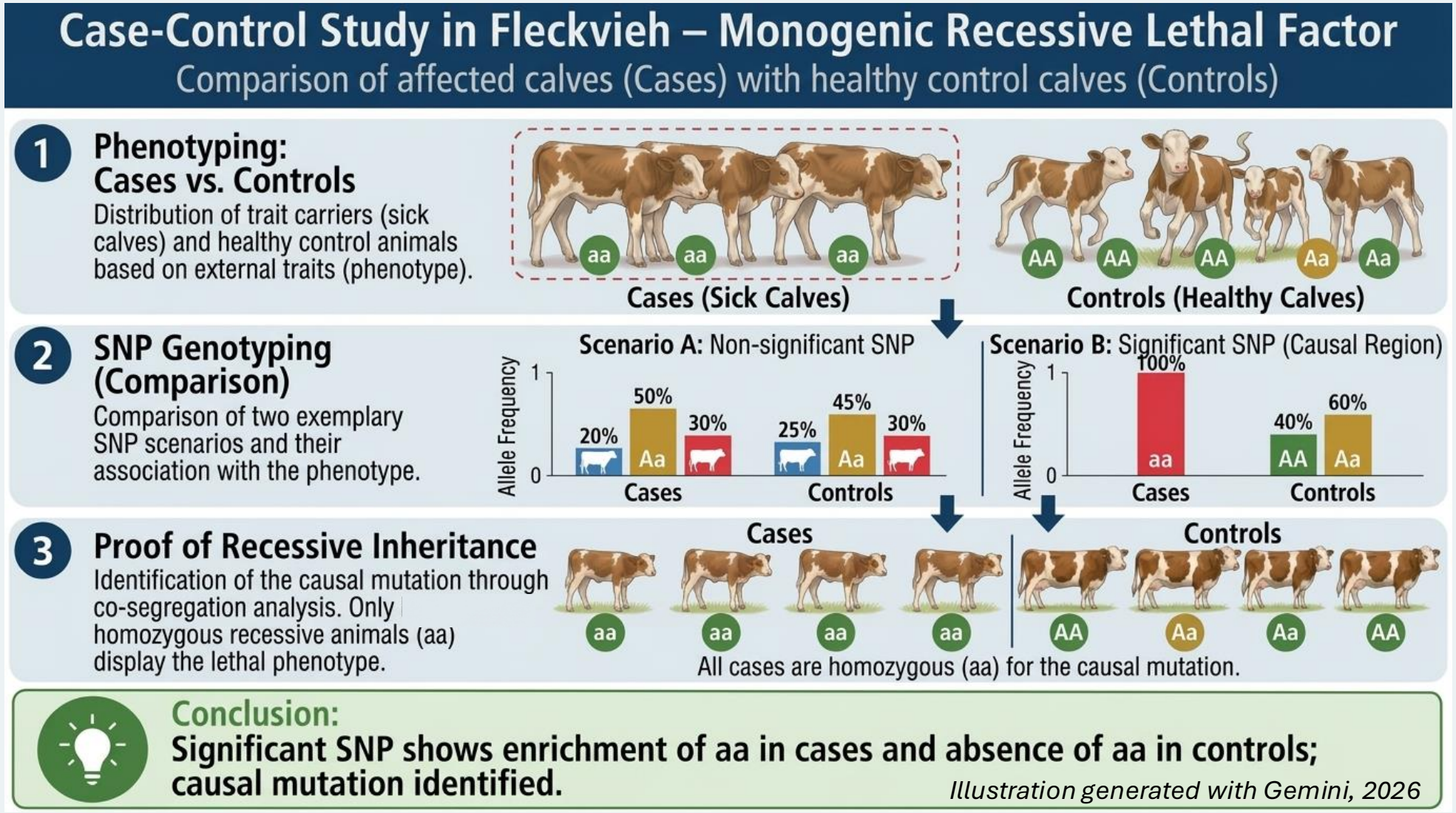
# Case-Control Study



# Case-Control Study

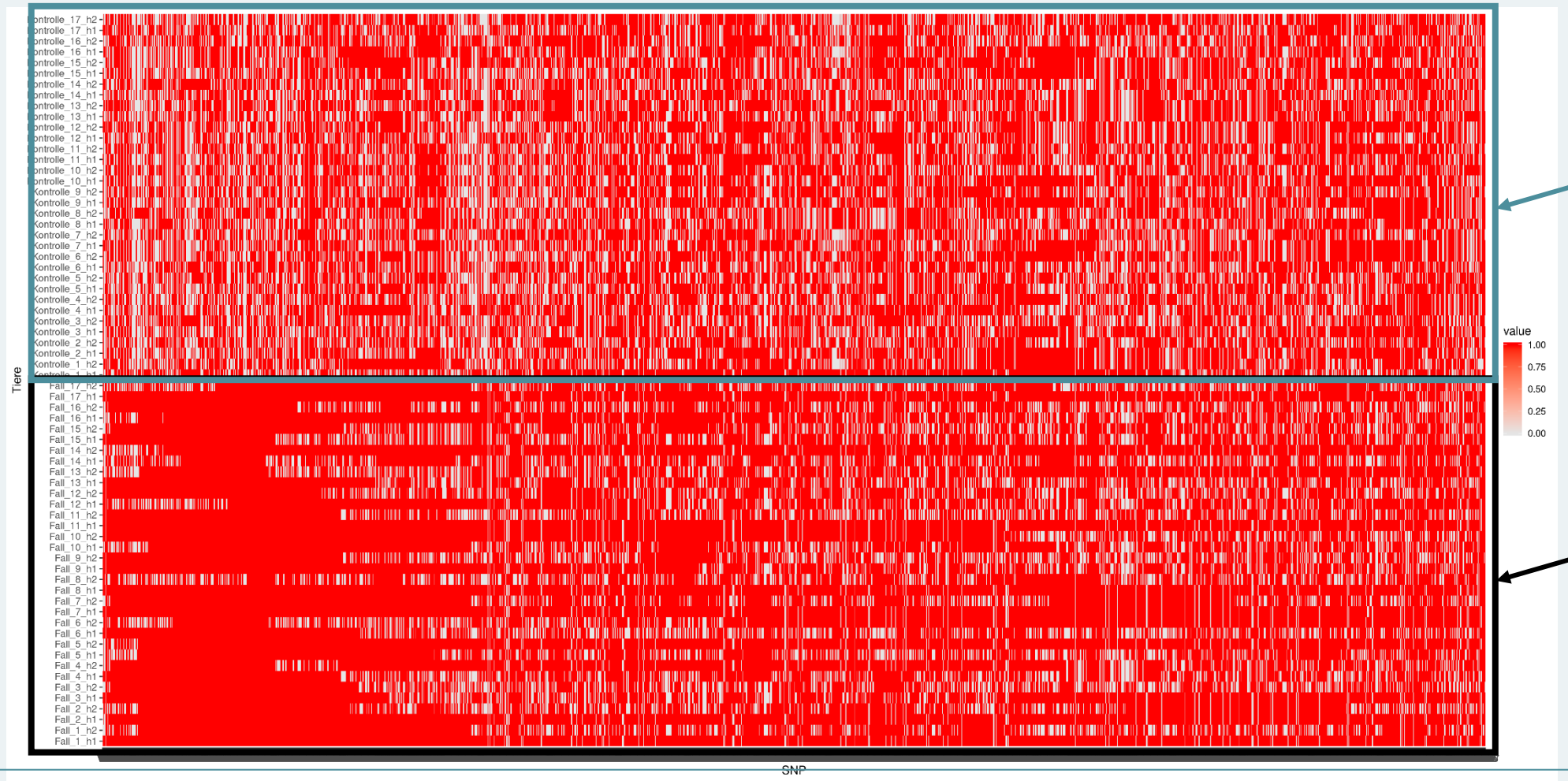


# Case-Control Study



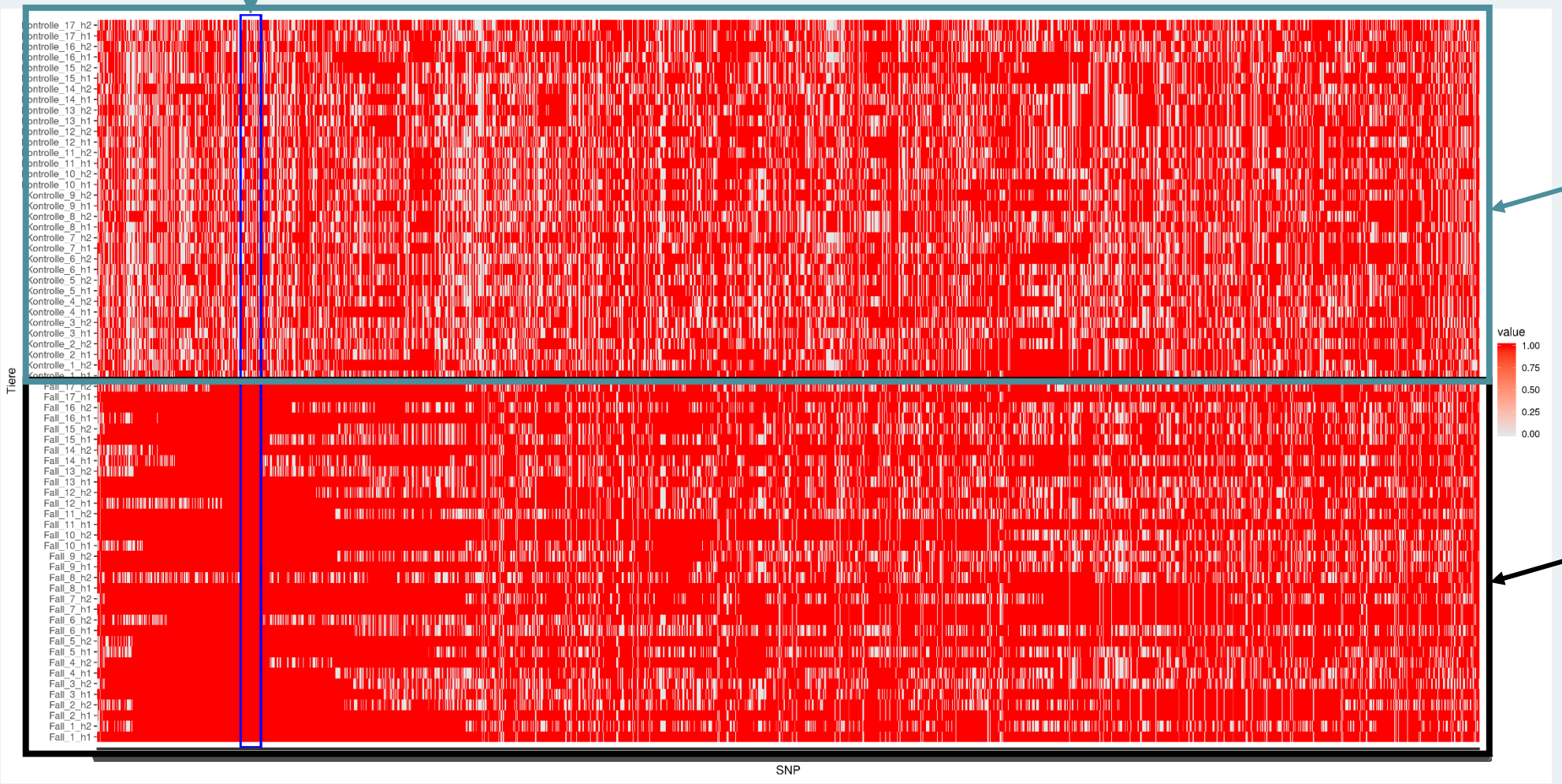
# Dwarfism in Fleckvieh

Chromosome 3, 2,005 SNP, 17 DW-cases, 1,326 controls



# Dwarfism in Fleckvieh

**Haplotype**: ***Homozygous in all cases but never on controls***




17 of 1,326 controls

17 DW cases

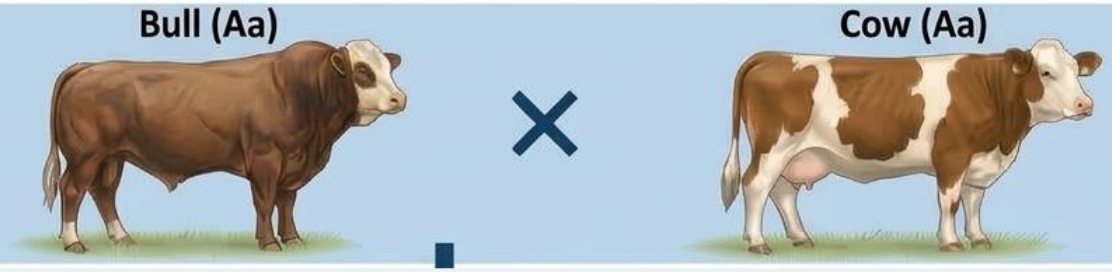
# Genotype-based monitoring

## VanRaden et al. (2008) – Genotype-Based Monitoring

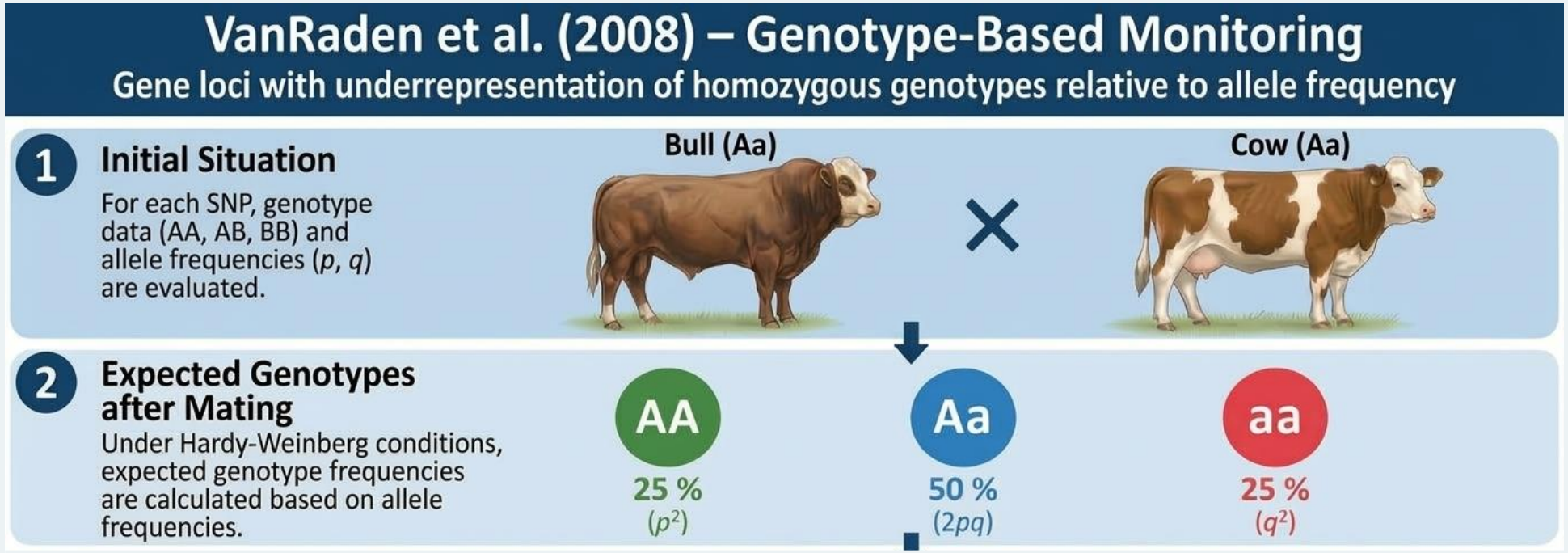
Gene loci with underrepresentation of homozygous genotypes relative to allele frequency

**1 Initial Situation**

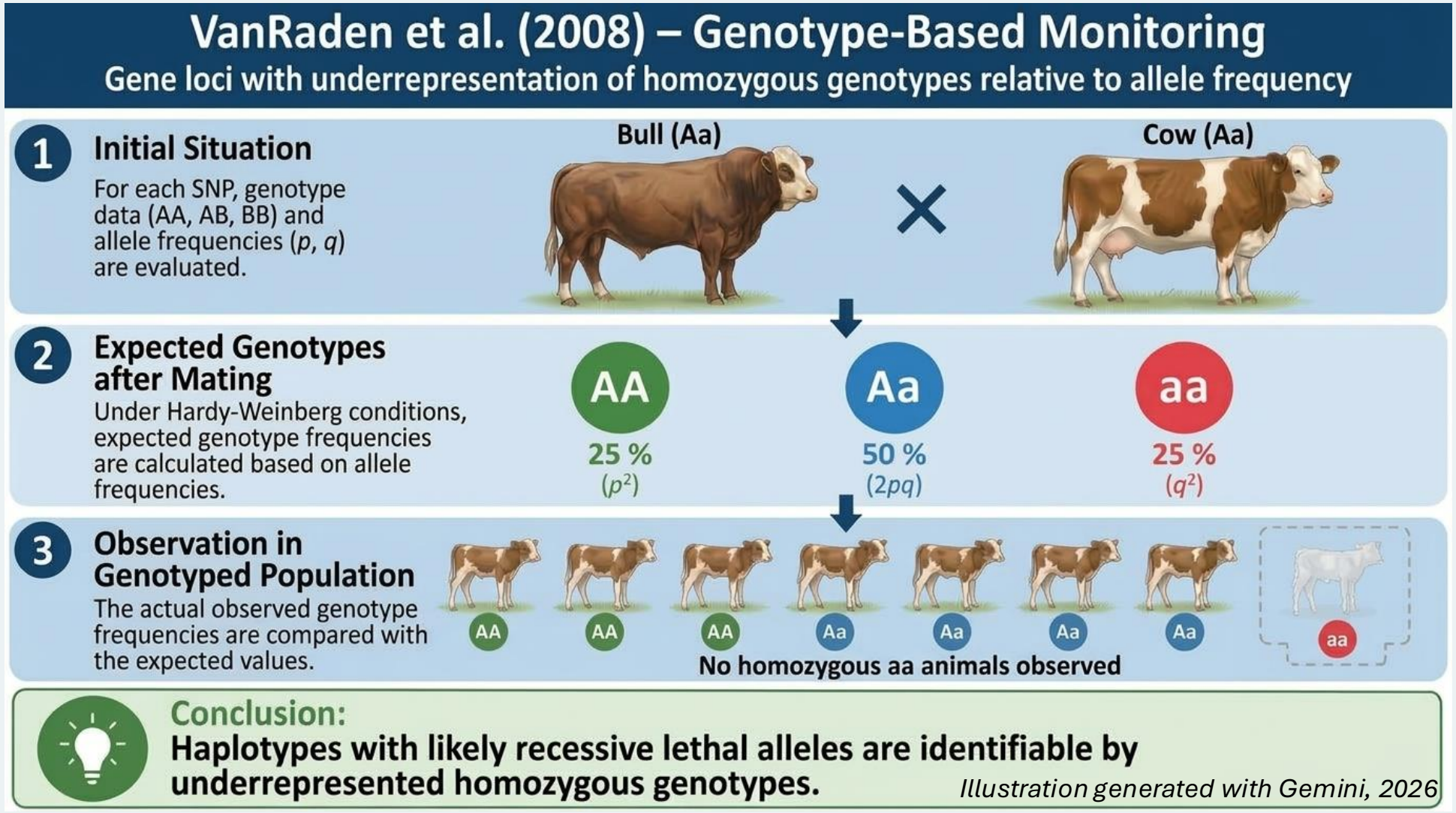
For each SNP, genotype data (AA, AB, BB) and allele frequencies ($p$, $q$) are evaluated.



# Genotype-based monitoring



# Genotype-based monitoring



# Example: Brown Swiss Haplotype 2: (B2)

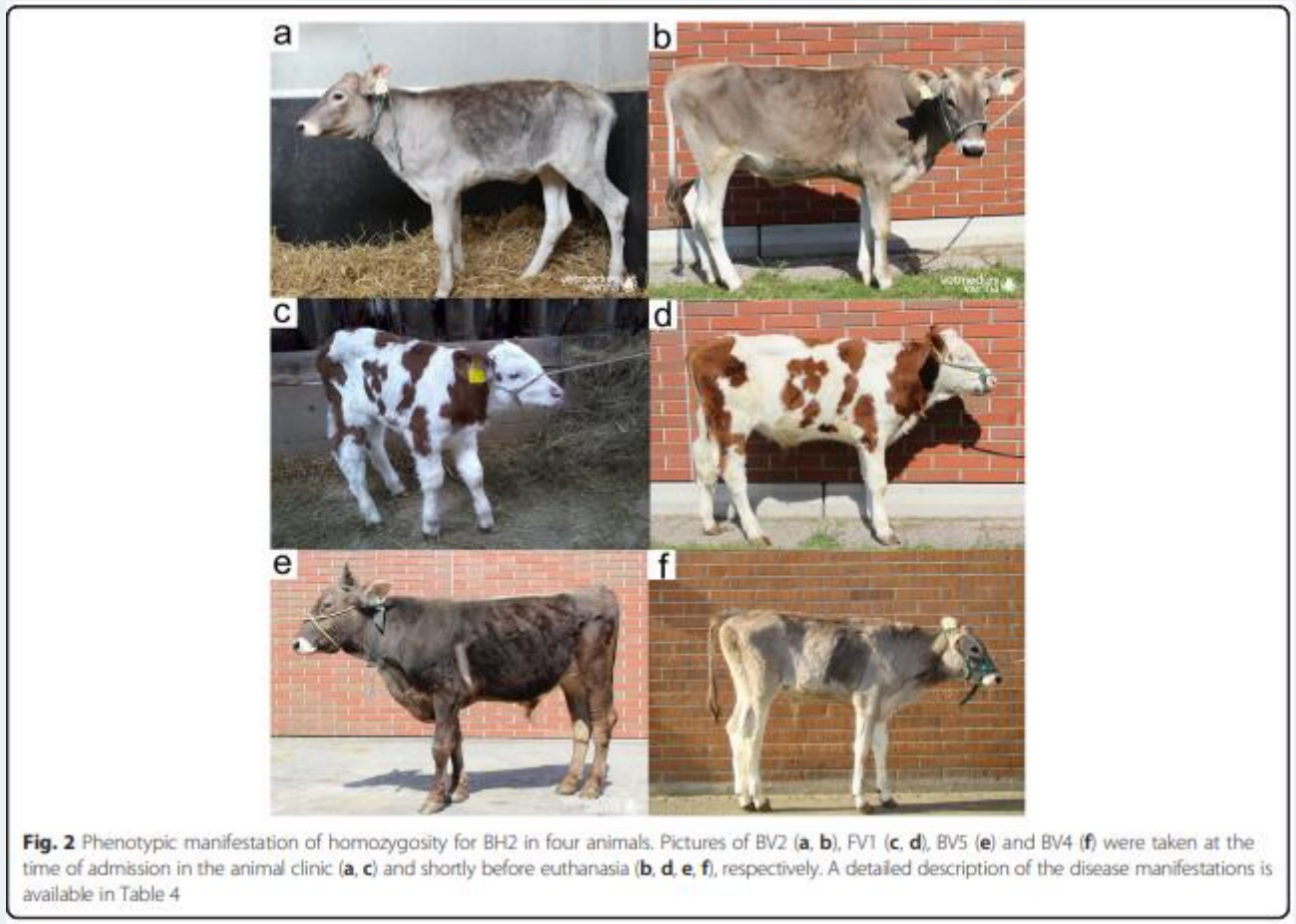



**Fig. 2** Phenotypic manifestation of homozygosity for BH2 in four animals. Pictures of BV2 (**a**, **b**), FV1 (**c**, **d**), BV5 (**e**) and BV4 (**f**) were taken at the time of admission in the animal clinic (**a**, **c**) and shortly before euthanasia (**b**, **d**, **e**, **f**), respectively. A detailed description of the disease manifestations is available in Table 4

Schwarzenbacher *et al. BMC Genomics* (2016) 17:400
DOI 10.1186/s12864-016-2742-y

BMC Genomics

RESEARCH ARTICLE Open Access

A missense mutation in *TUBD1* is associated with high juvenile mortality in Braunvieh and Fleckvieh cattle

Hermann Schwarzenbacher[1], Johann Burgstaller[2], Franz R. Seefried[3], Christine Wurmser[4], Monika Hilbe[5], Simone Jung[4], Christian Fuerst[1], Nora Dinhopl[6], Herbert Weissenböck[6], Birgit Fuerst-Waltl[7], Marlies Dolezal[8], Reinhard Winkler[9], Oskar Grueter[10], Ulrich Bleul[11], Thomas Wittek[2], Ruedi Fries[4] and Hubert Pausch[4*]

segregates also in Fleckvieh …

below-average birth weight

higher stillbirth rate

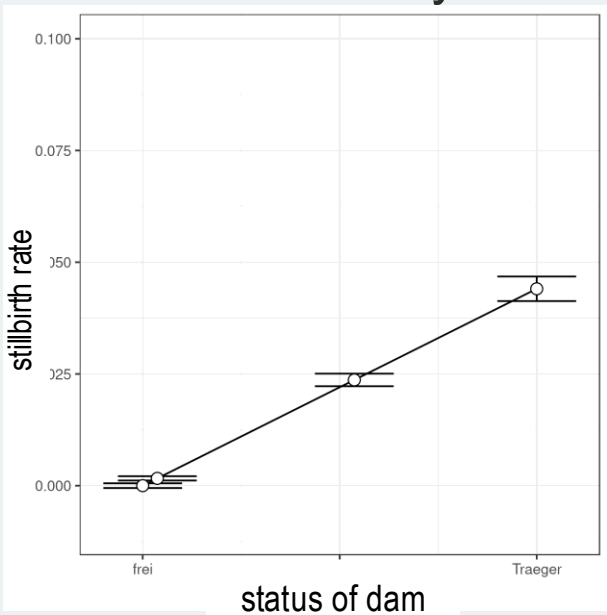
markedly increased mortality during the first seven weeks of life

typical clinical signs are **nasal discharge and pneumonia**

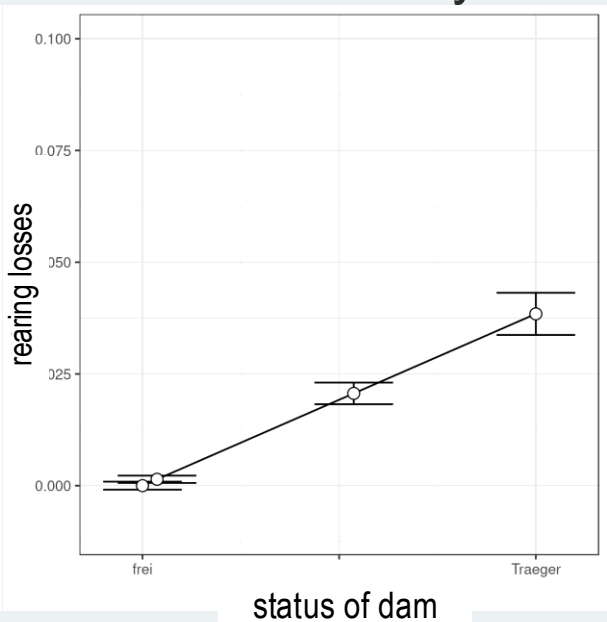
# B2 risk-matings

Risk matings with B2-carrier bulls,
effect of B2-status of dam on still birth and rearing losses

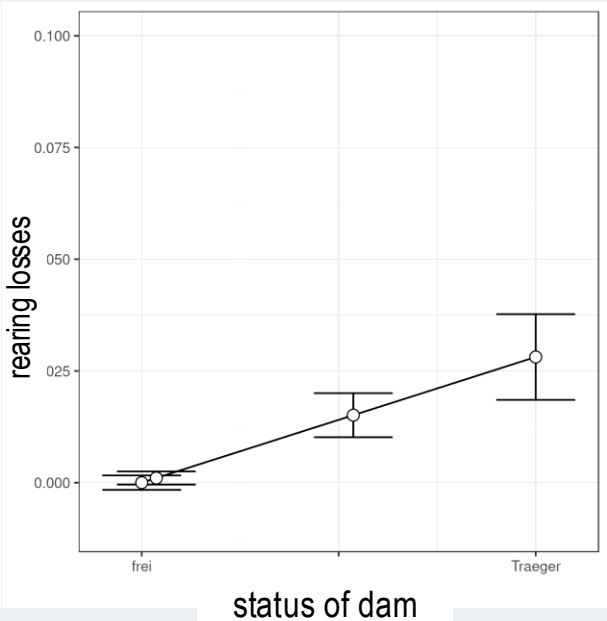
still birth rate
0 to 2nd day



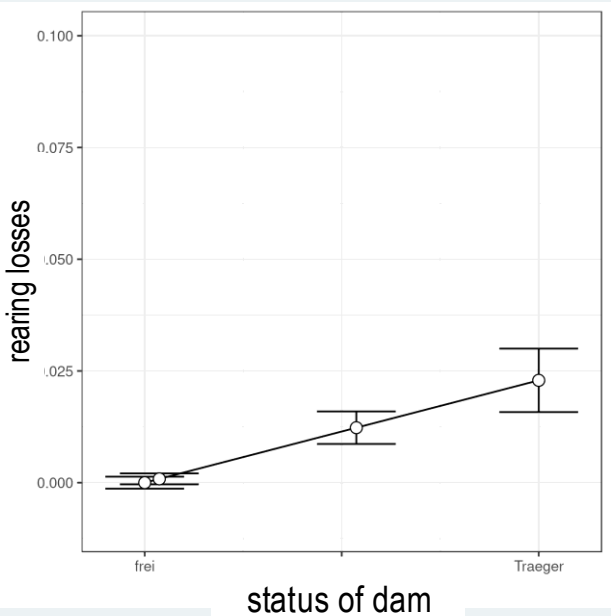

rearing losses
3rd to 30th day




rearing losses
31st d.-10 m. males




rearing losses
31st d.-15 m. females



# Longitudinal Study

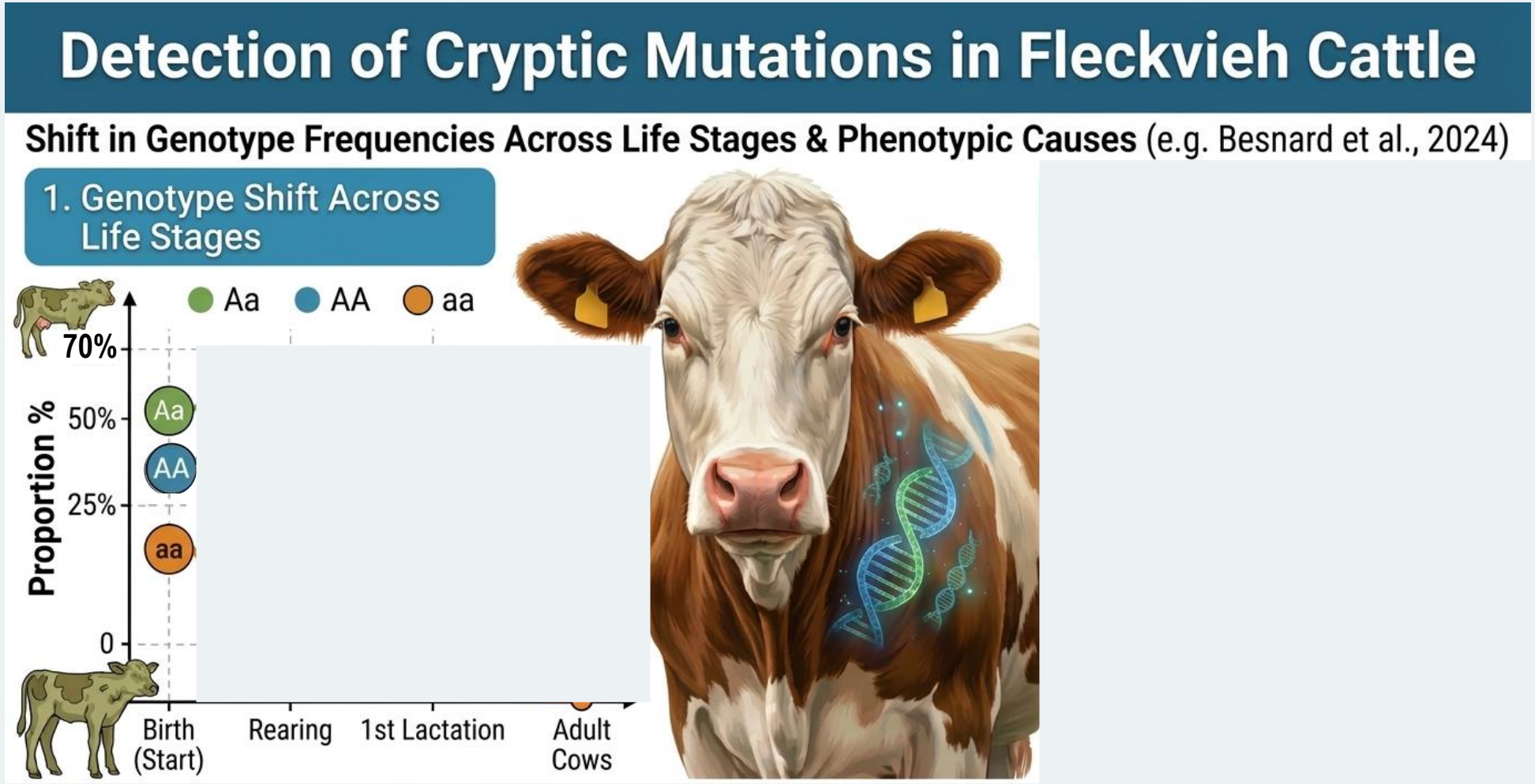
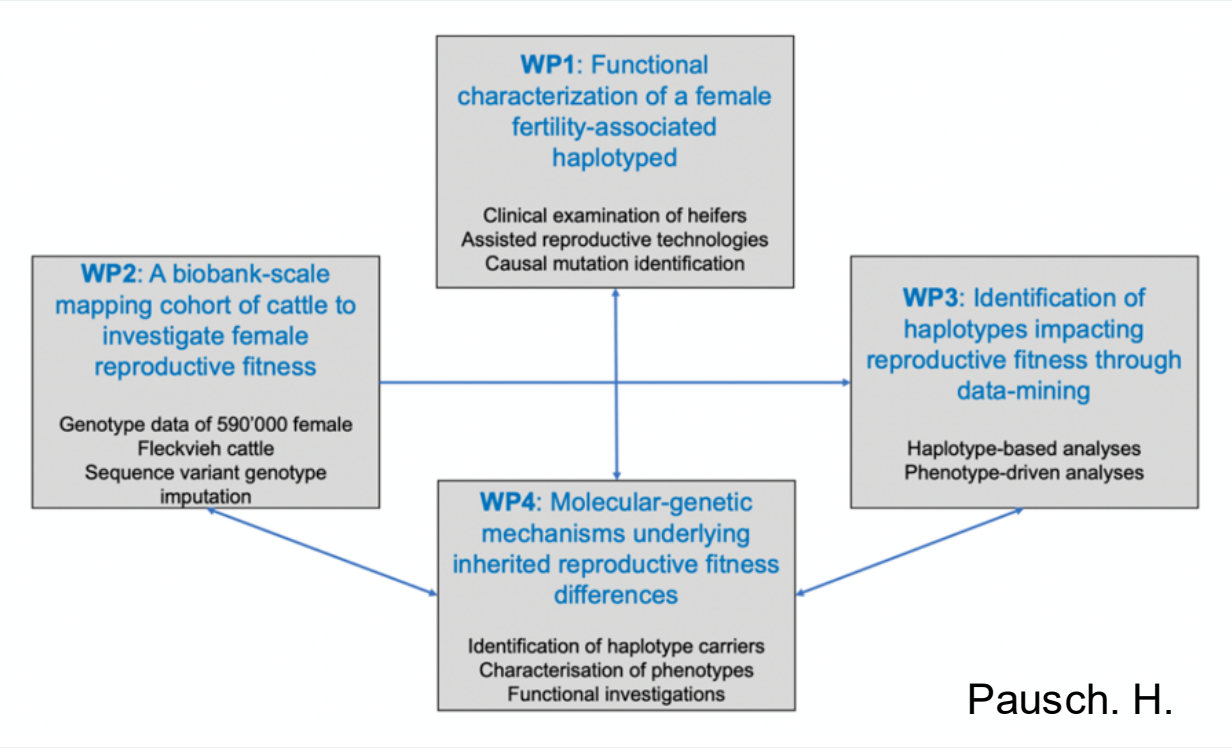



*Illustration generated with Gemini, 2026*

# Longitudinal Study

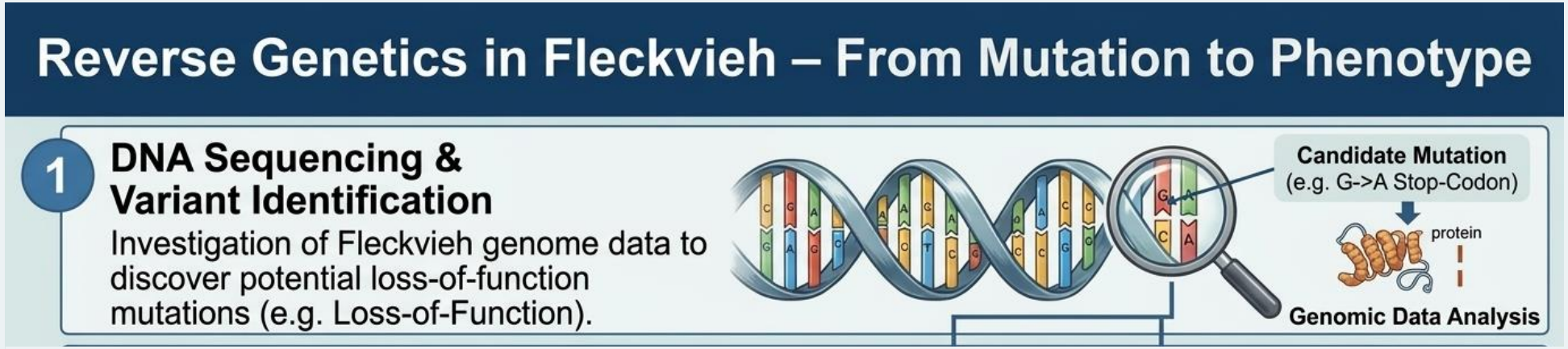
*Research Project, ETH Zürich, Prof. Pausch and colleagues:*

**“A data-mining approach to detect genomic regions impacting female reproductive fitness in cattle.”**

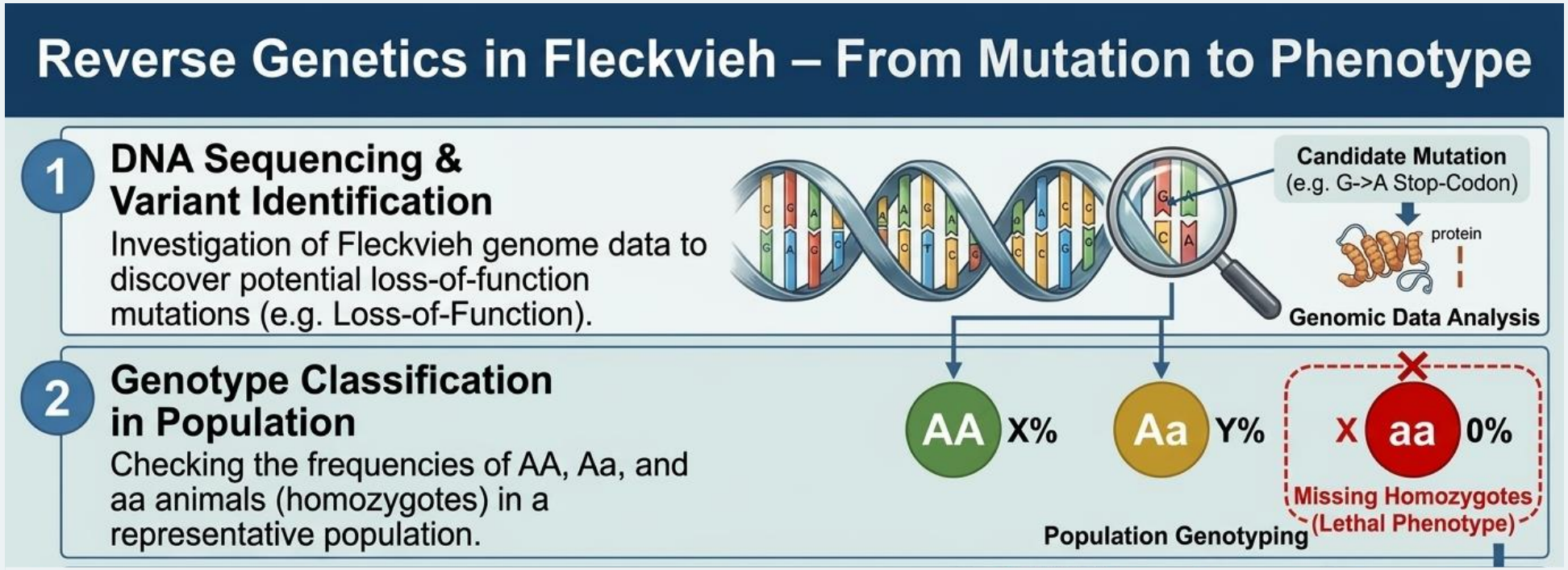



variants that lead to losses in **later life** come with **higher costs!**

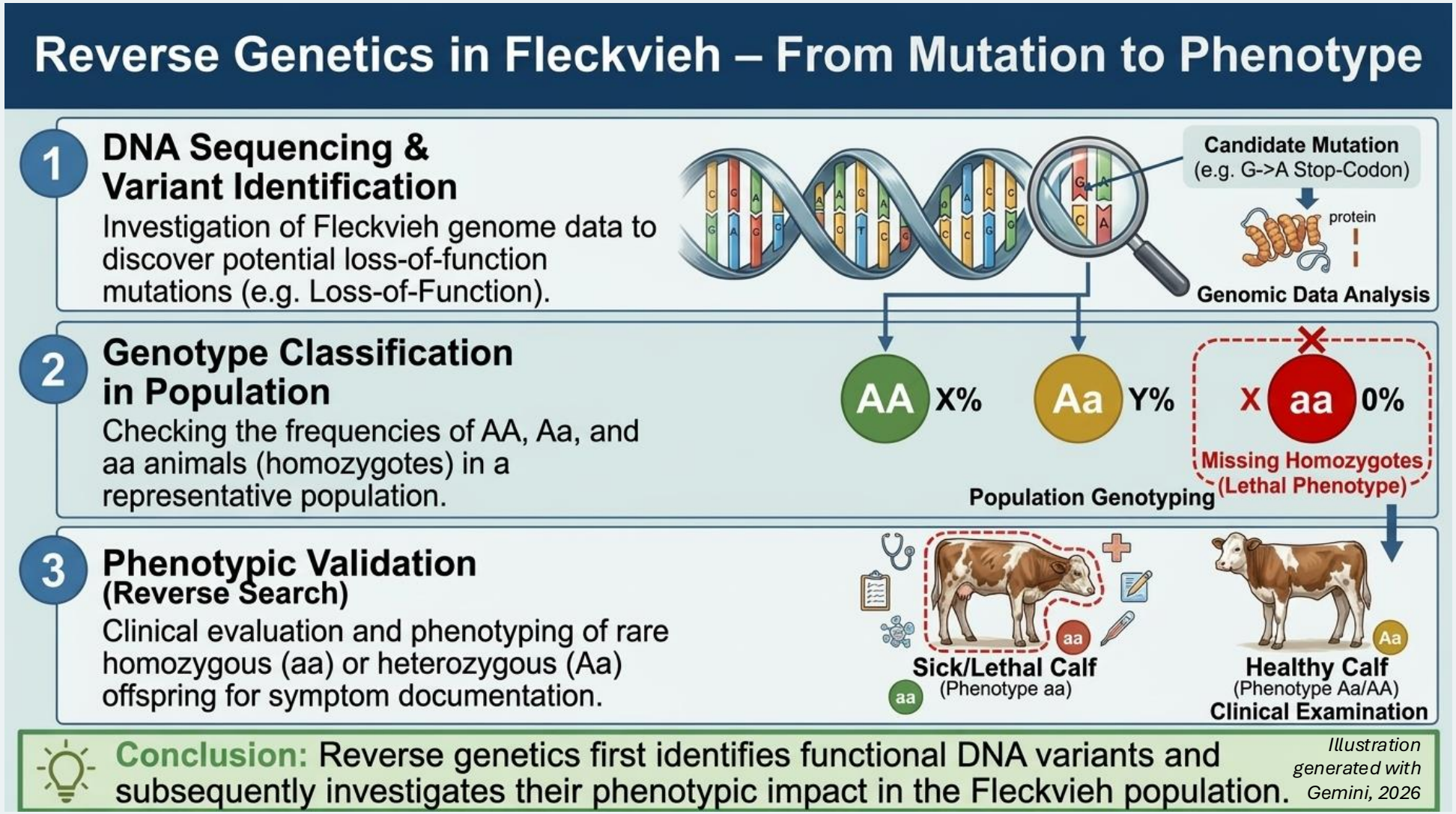
# Reverse Genetics



# Reverse Genetics

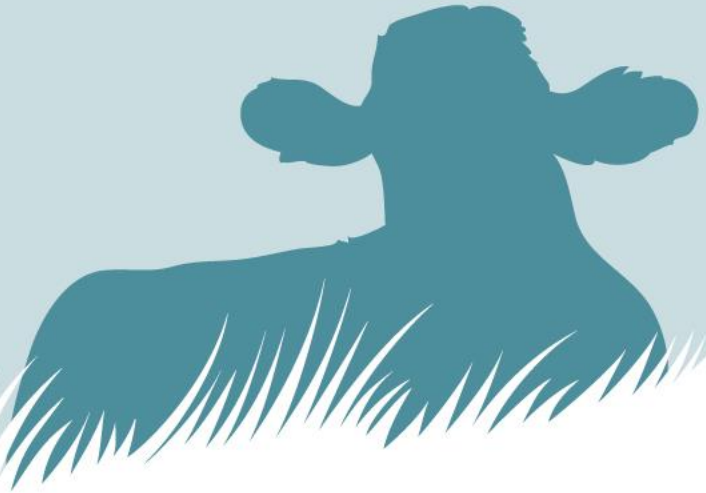


# Reverse Genetics



# *Desirable Genetic Variants*

*Polled*
*κ-Casein*
*β-Casein*



# Inheritance of polled

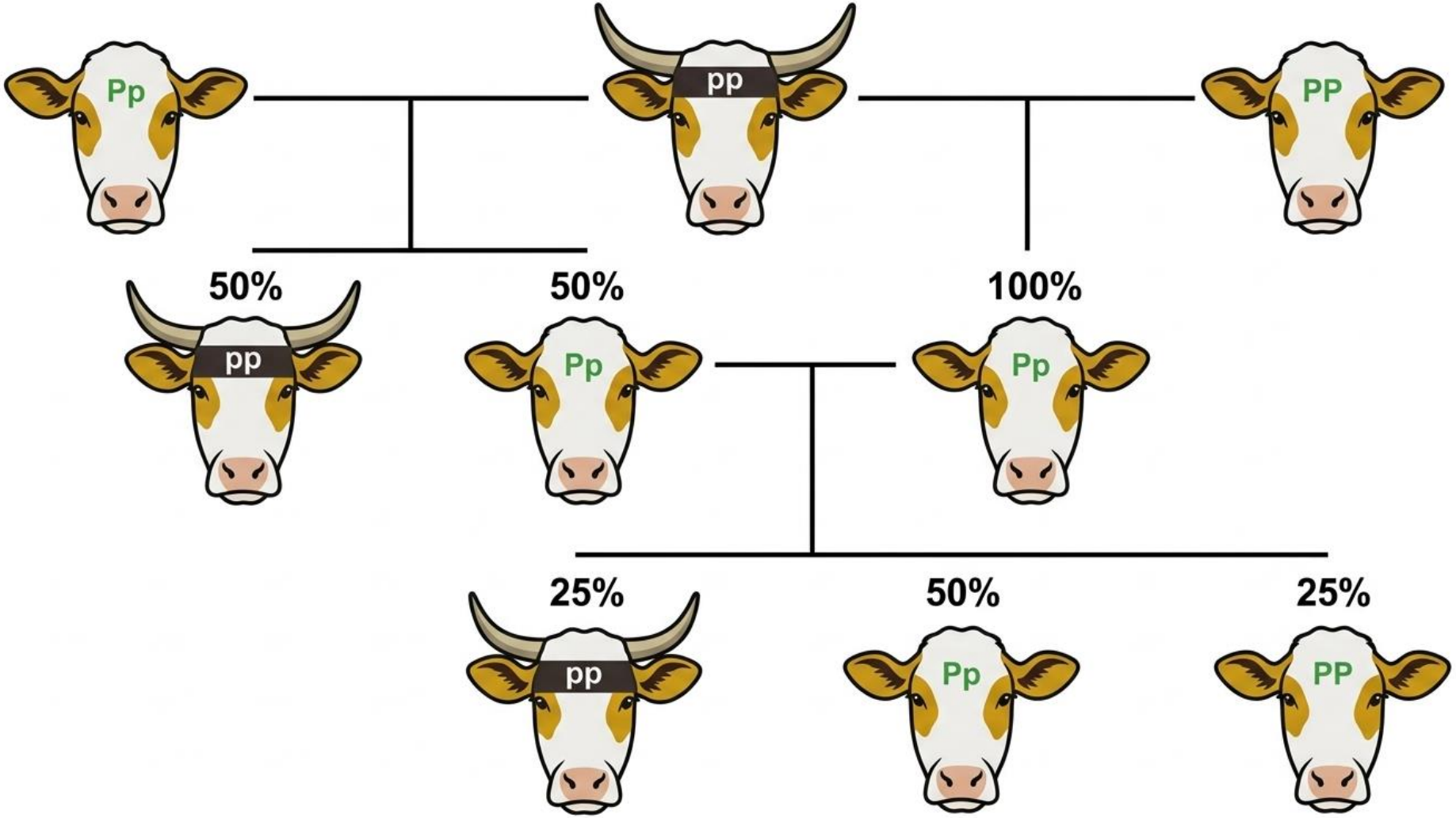


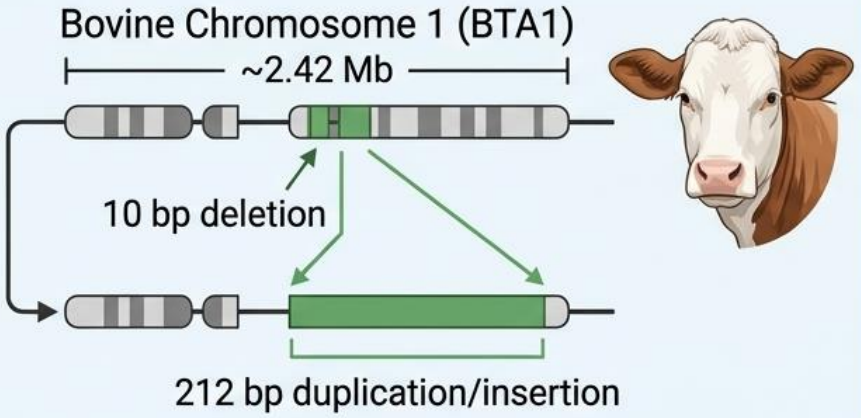

*Illustration generated with Gemini, 2026*

# The genetics of polled

## Genetics of Polledness: Celtic (PC) vs. Friesian (PF) Mutations (BTA1 POLLED Locus)

### Polled Celtic Variant (PC)

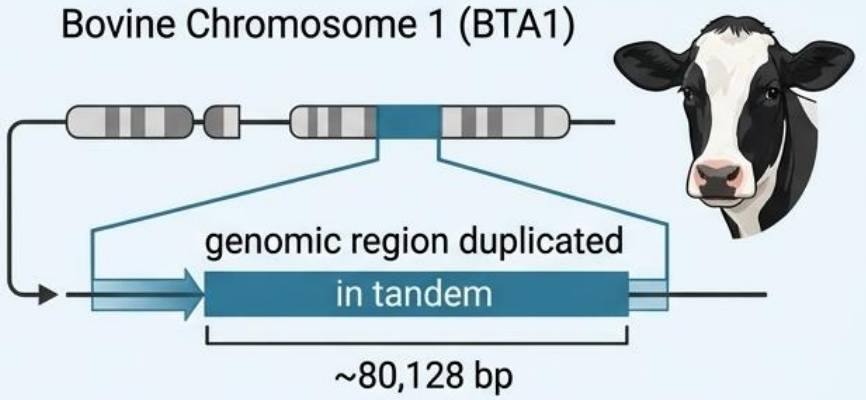
**212 bp Complex Indel** (Insertion-Deletion)




- Commonly found in Fleckvieh, Simmental, Angus, and Hereford.

### Polled Friesian Variant (PF)

**~80 kb Tandem Duplication**



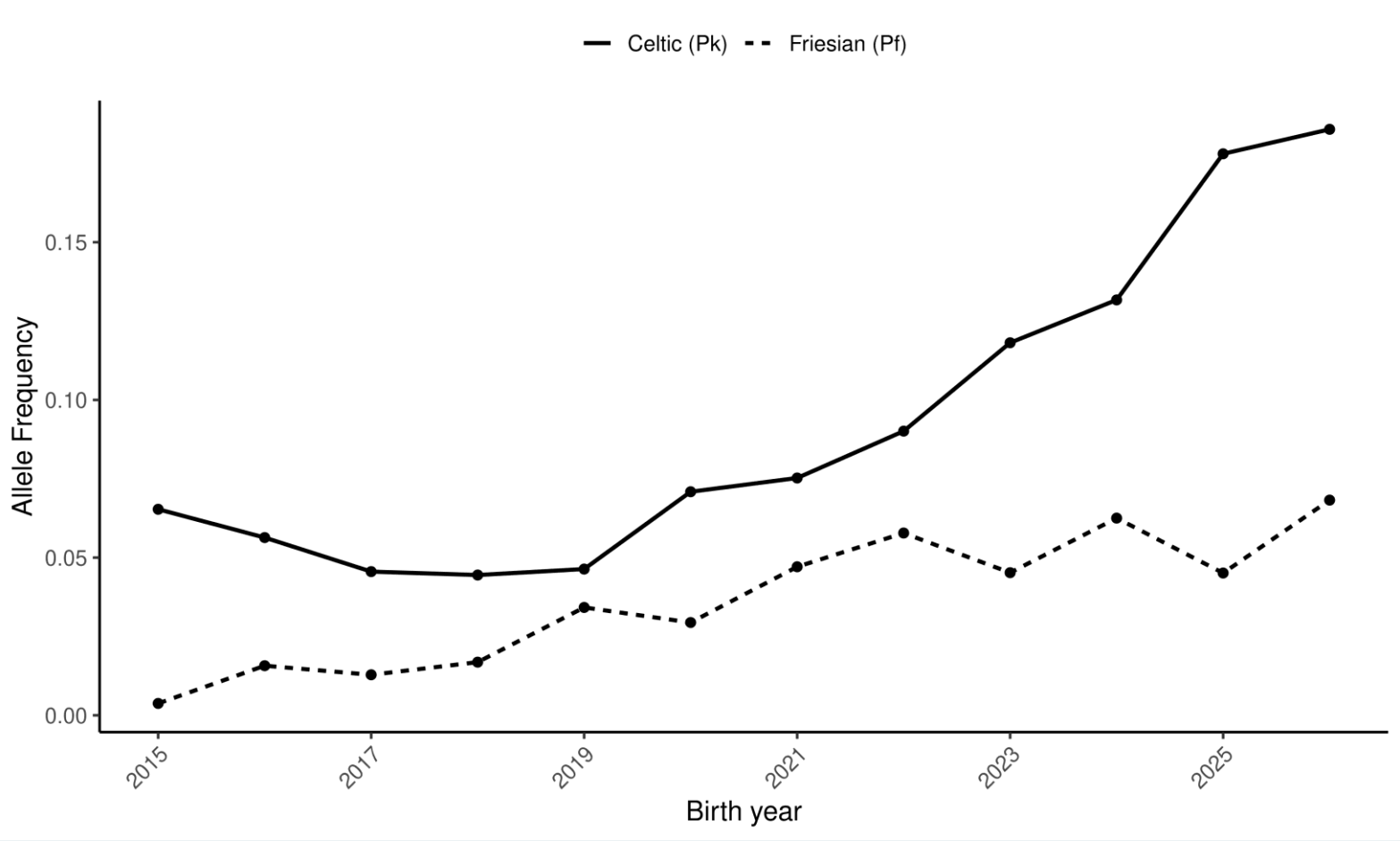

- Commonly found in Holstein-Friesian and Fleckvieh lines.

**Conclusion:** Both PC and PF mutations lie on Chromosome 1 (POLLED locus) and show Autosomal Dominant inheritance, resulting in a naturally hornless phenotype.

*Illustration generated with Gemini, 2026*

# The genetics of polled

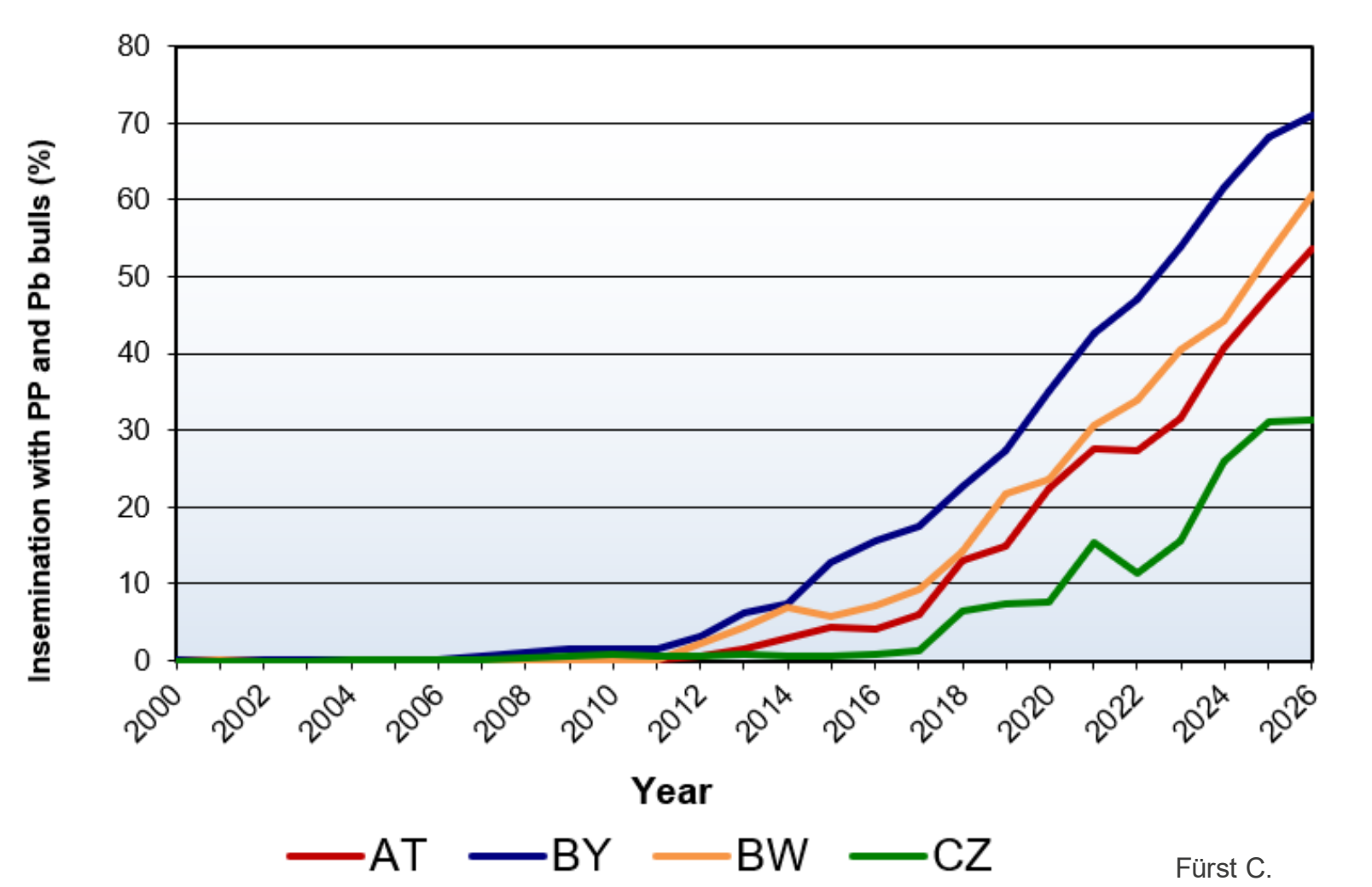
Polled allele frequencies in female genotyped Fleckvieh population




polled is under strong selection

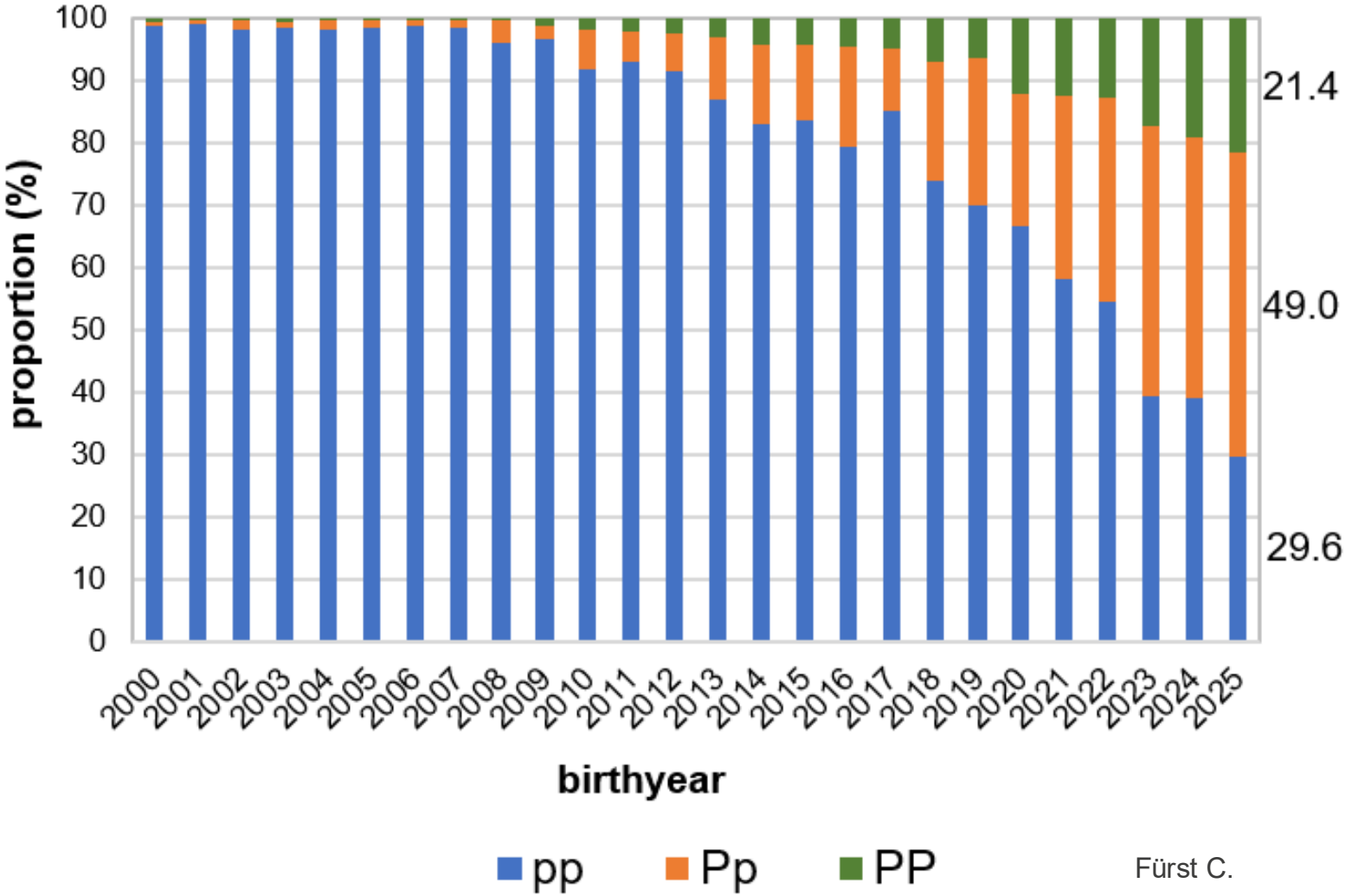
mainly on celtic variant

# Inseminations with polled: Pp+PP



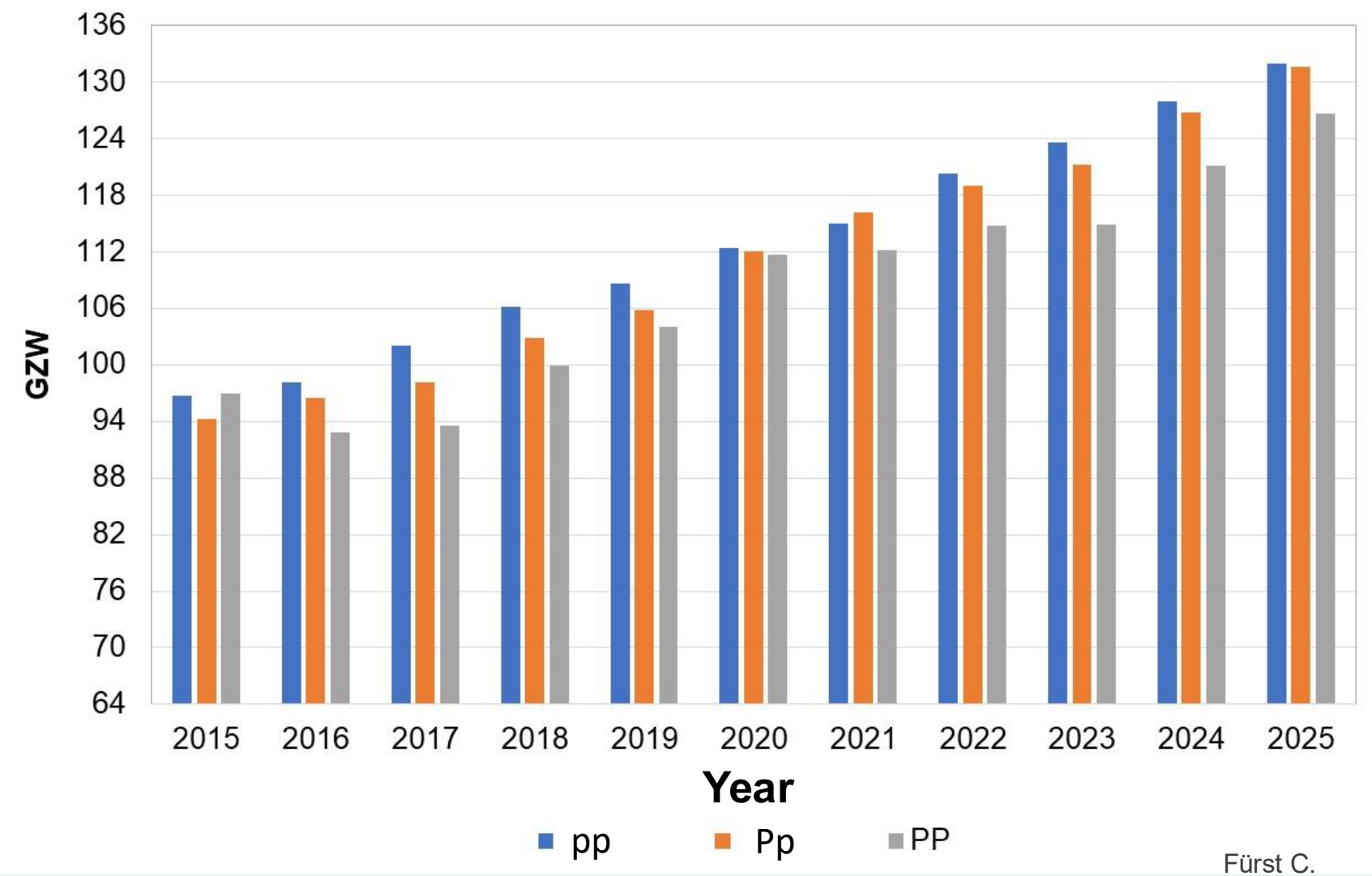

Fürst C.

# Polled status of AI bulls (AT+DE+CZ)




more than
2/3 polled!

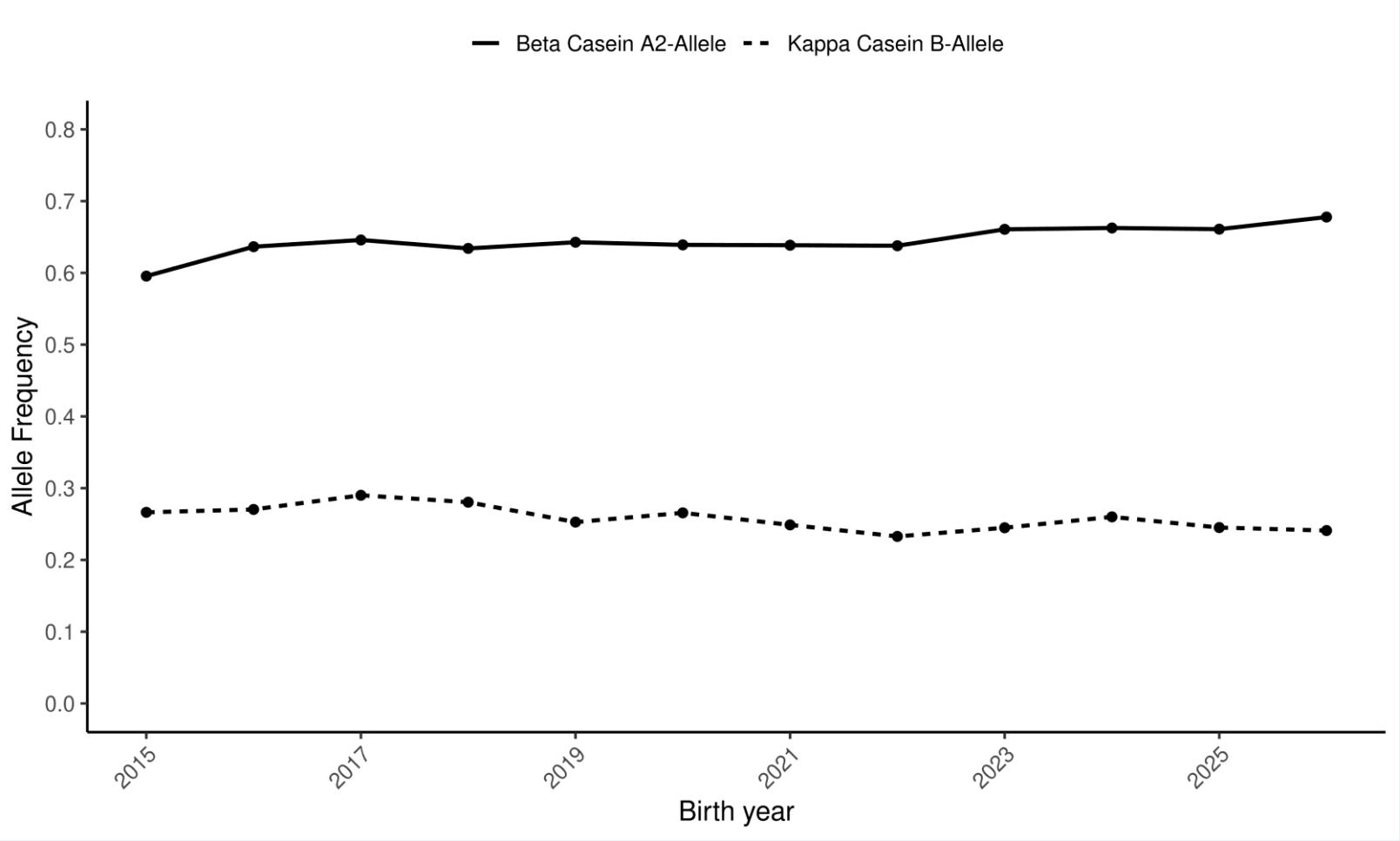
# TMI of used semen by horn status




Fürst C.

# K-Casein and β-Casein

Allele frequencies in female genotyped Fleckvieh population

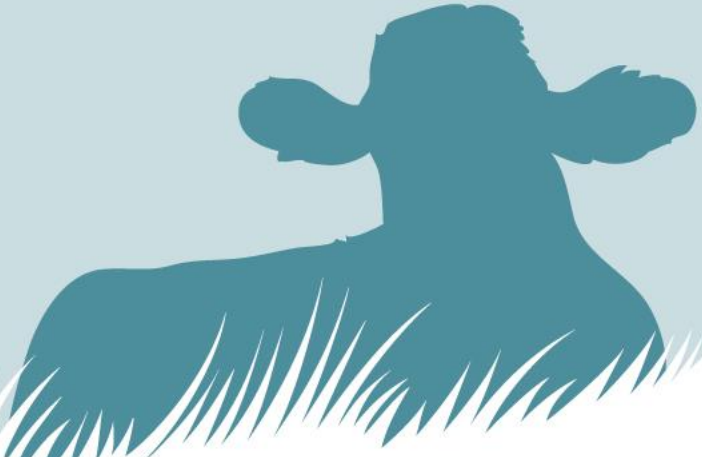



not much selection visible on milk casein composition

**no price premium** for either trait

**scientific evidence still under debate** for β-casein

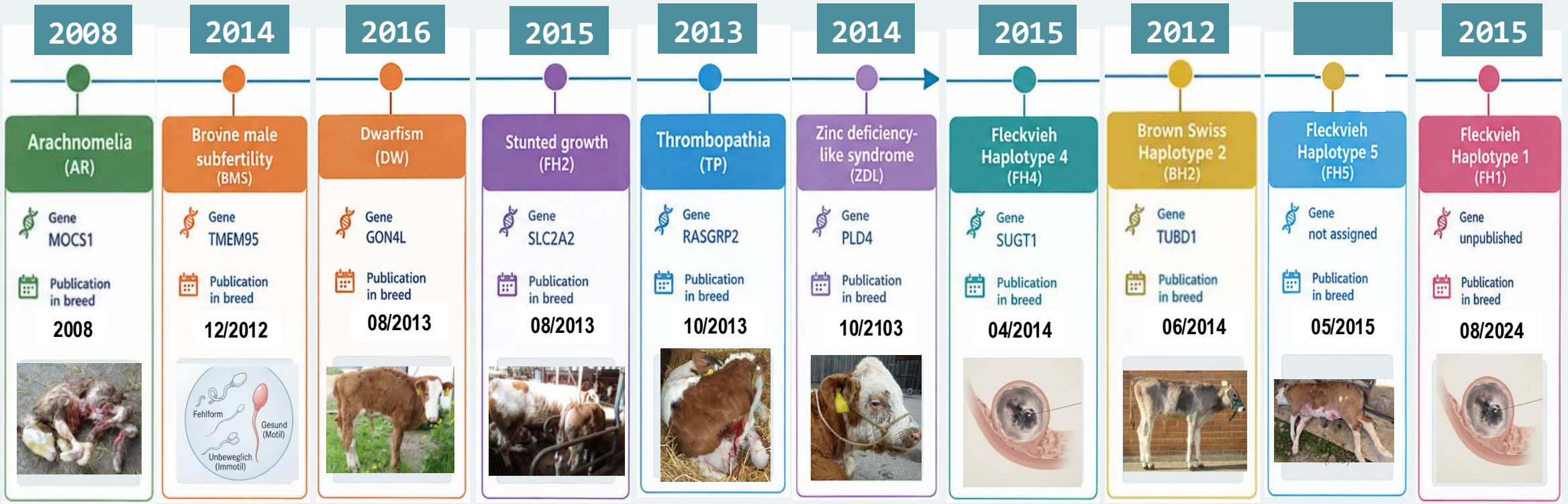
# *Unfavourable Genetic Variants*



# Unfavourable variants in Fleckvieh

Ordered by date of publication in breed

year of scientific publication



# Genetic defect labeling

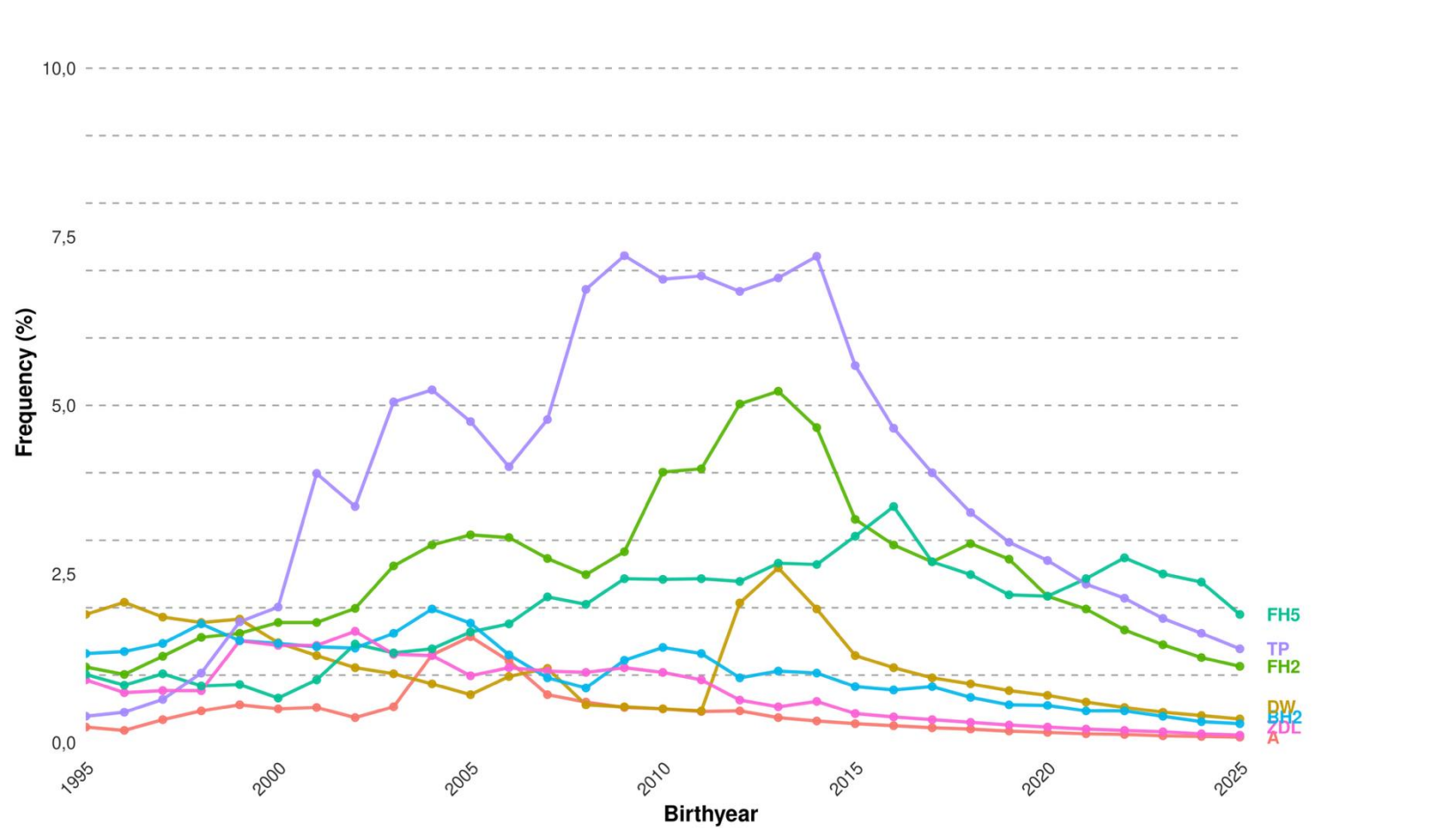
**Pos. 1-2/3:** **Abbreviation for defect**
**Pos. 3/4:** **Carrier status: C = carrier**
**F = free**
**S = homozygous carrier (sure)**

| Defect | Abbrev. |
|---|---|
| Arachnomalia | AR |
| Bovine male Subfertility BMS | MS |
| Brown Swiss-Haplotype 2 BH2 | B2 |
| Fleckvieh-Haplotype 1 FH1 | F1 |
| Fleckvieh-Haplotype 4 FH4 | F4 |
| Fleckvieh-Haplotype 5 FH5 | F5 |
| Stunted growth FH2 | F2 |
| Thrombopathia TP | TP |
| Zinkdefiziency-like Syndrom ZDL | ZL |
| Dwarfism | DW |

**Examples:**
**B2C =**
heterozygous carrier of BH2
**ARF =**
frei von Arachnomalia
**MSS =**
homozygus carrier of BMS

# Defects with visible cases

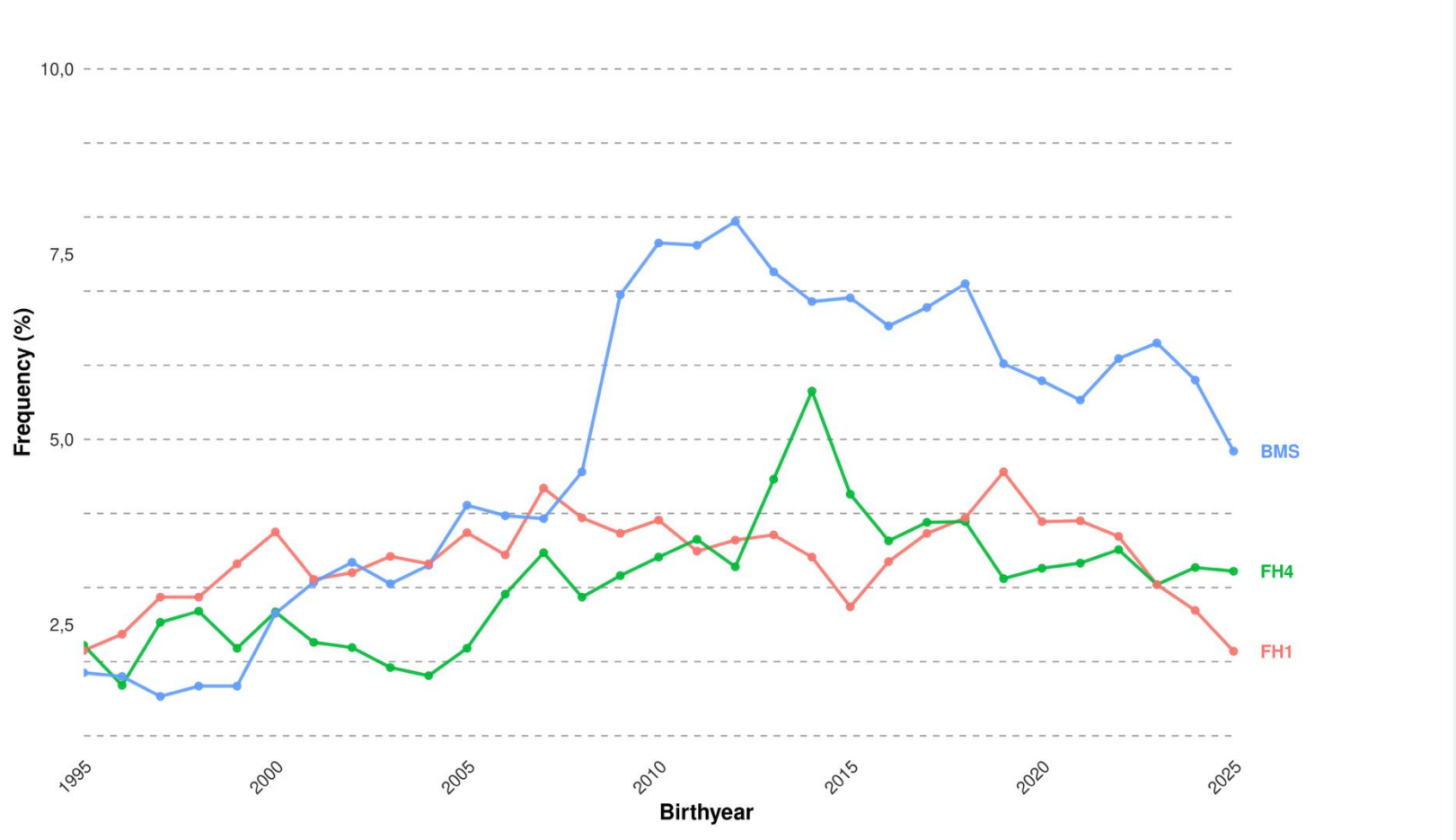
## Frequencies in the female population (AT)




very strong selection against defects with visible cases

# Defects with no visible cases

## Frequencies in the female population (AT)




moderate selection against defects with no visible cases

# *Genomic Monitoring using Custom SNPs*

# Genomic monitoring

Chips consist of different SNP groups

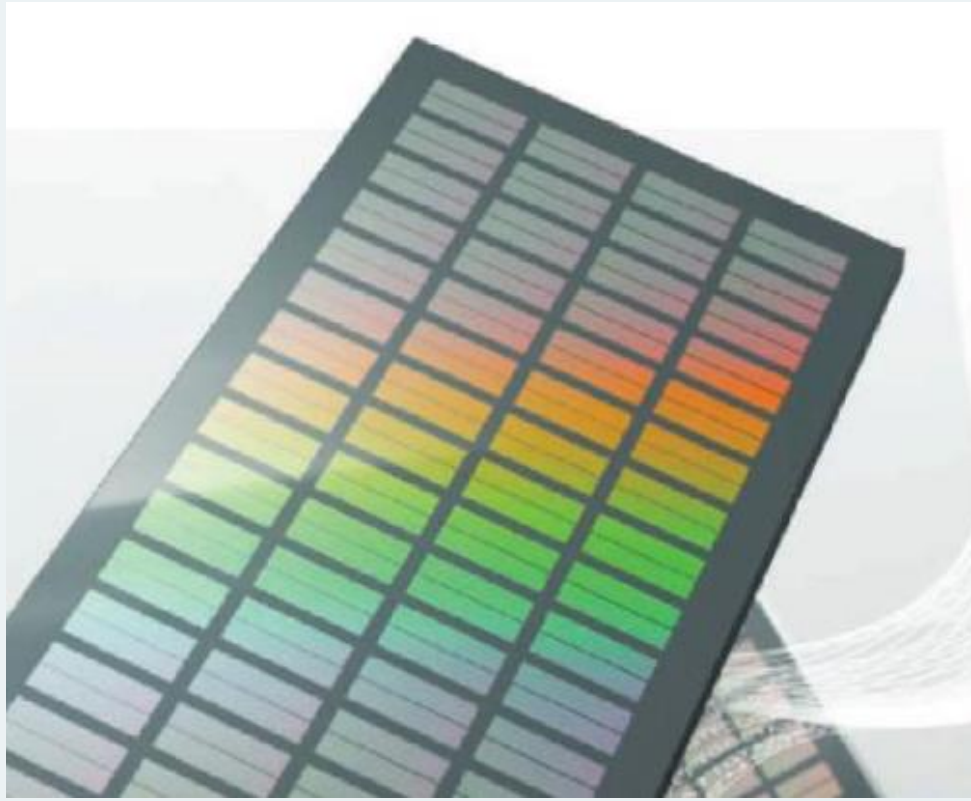
- SNPs for genomic breeding values
  - cover the genome well, no direct effect on the phenotype
- SNPs for parentage verification
  - reliable, occur in many breeds, no genomic prediction possible
- SNPs for direct genetic tests
  - genetic defects and genetic traits
  - usually commissioned and designed by the customer

Design of direct genetic tests based on own research and/or data bases such as OMIA (Online Mendelian Inheritance in Animals)

# SNP genotypes in data base (all breeds)

15/09/2026

| Chip version | No. SNP | No. Custom SNP | No. Animals | No. Genotypes (bn) |
|---|---|---|---|---|
| Illumina K50 V1A | 54 001 | | 6 301 | 0.34 |
| Illumina K50 V1B | 54 001 | | 24 683 | 1.33 |
| Illumina K50 V2 | 54 609 | | 52 457 | 2.86 |
| Illumina K50 V3 | 53 218 | | 1 200 | 0.06 |
| Illumina HD | 777 962 | | 1 254 | 0.98 |
| Illumina K80 GGPHD | 76 879 | | 2 975 | 0.23 |
| Illumina K50 Custom V1 | 56 263 | **1 718** | 15 691 | 0.88 |
| Illumina K50 Custom V2 | 56 715 | **2 181** | 33 091 | 1.88 |
| Illumina K50 Custom V3 | 45 994 | **2 323** | 14 547 | 0.67 |
| Illumina K50 Custom V4 | 46 691 | **2 364** | 100 261 | 4.68 |
| Illumina K50 Custom V5 | 56 460 | **2 508** | 118 977 | 6.72 |
| Illumina K50 Custom V6 | 59 003 | **3 058** | 120 545 | 7.11 |
| Illumina K50 Custom V7 | 59 117 | **3 161** | 284 788 | 16.84 |
| Illumina K50 Custom V8 | 57 805 | **3 773** | 525 686 | 30.38 |
| **Total** | | | **1 302 456** | **75** |



# Genomic monitoring

currently…

- 114 genomic locations
- in 27 cattle breeds
- analysed as part of the 14-day routine

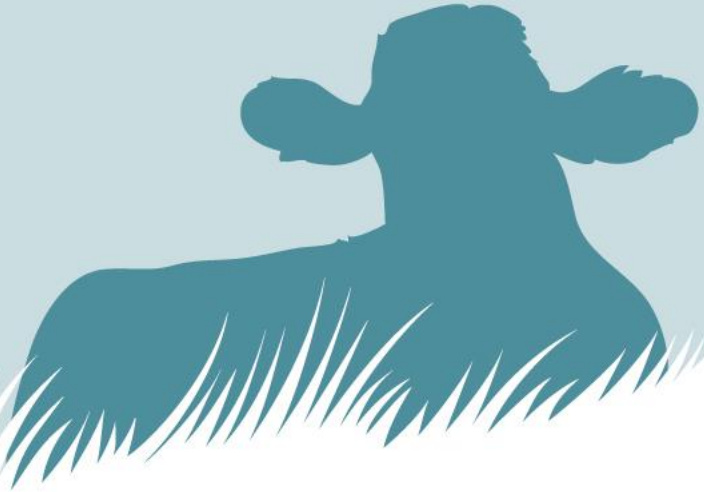
- which variant segregates in which breed
- check allele frequencies over time
- carrier animals

# Many variants are NOT breed-specific

Frequencies of loci found in Fleckvieh that segregate in other breeds

| Locus [OMIA-ID*] | Angus | Blonde Aquitaine | Brown Swiss | Charolais | Ennstaler Bergscheck. | Galloway | Grauvieh | Kaerntner Blondvieh | Limousin | Murbodner | Original Braunvieh | Original Pinzgauer | Pustertaler Sprinzen | Schot. Hochlandr. | Tuxer | Wagyu | Waldviertler Blondvieh | Weiß-Blaue Belgier |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| A (FV) [001541] | | | | | 8.1 | | | | | | | | | | | | | |
| BH2 [001939] | | | 1.9 | | | | | | | | | | | | | | 18.6 | |
| FH2 [000366] | | | | | | | | | | | 1.5 | | | | | | | |
| FH5 [002960] | | | | | | | | | | | | | | | 0.8 | | | |
| TP [002433] | | | | | 0.9 | | | | | 0.6 | | | 5.4 | | | | | |

# *Take-home message*



# Conclusions

01 Genomic toolbox enable efficient management of Mendelian traits.
Especially in large populations under genomic selection.

02 Increased awareness among breeders and decision makers and less ‚fear‘.
Rezessives with visible cases are under rigorous selection.
Genetic defects are still a disruption of the breeding business!

03 Higher speed needs clear vision and good breakes.

from Mendel
to Genom

# THANK YOU!

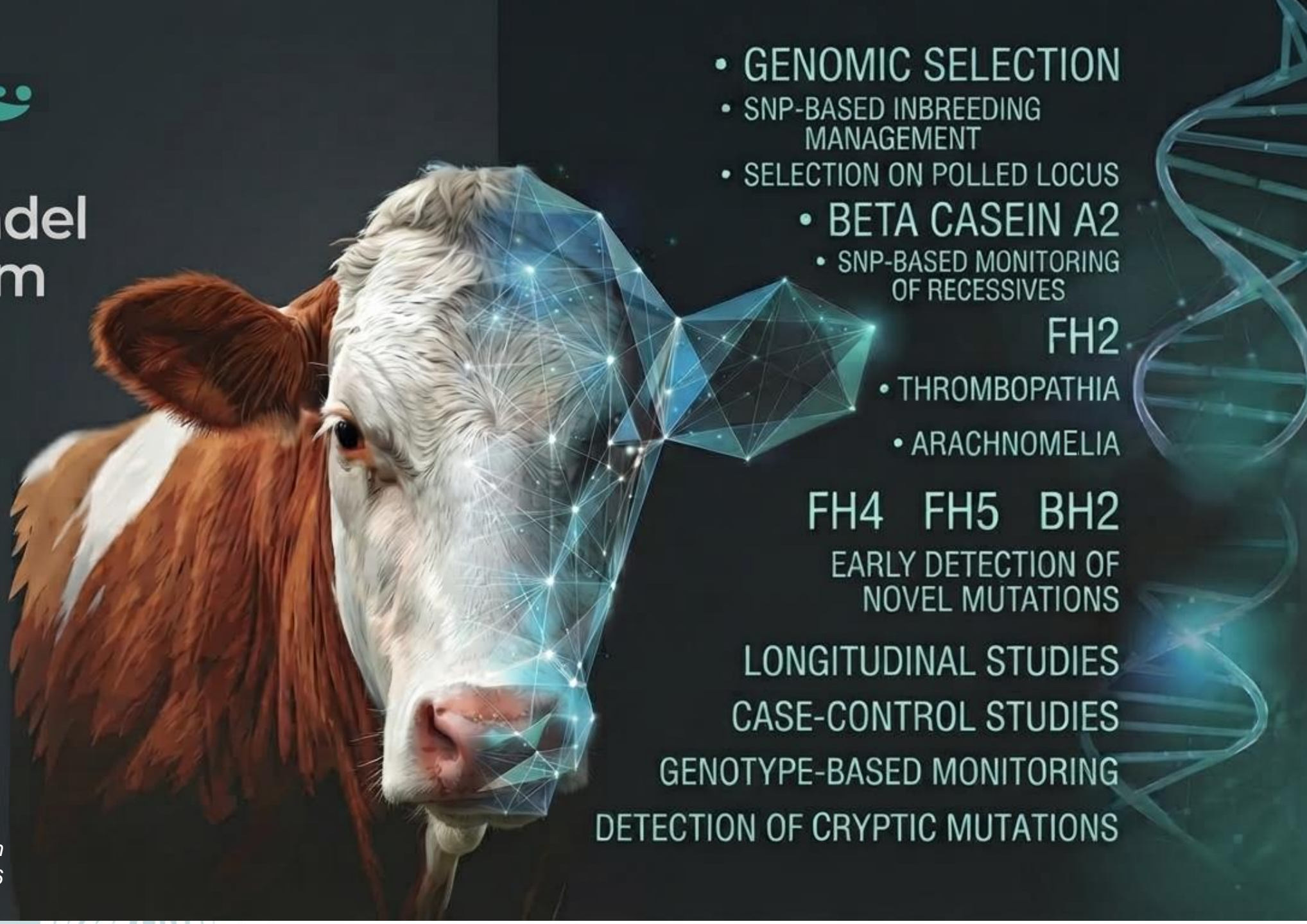



*Illustration generated with OpenAI, 2026*