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Extended vs. new imputation systems for supporting transfer to SNP-based parentage control in horses of different breeds

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Session 51
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Outline



- ❖ background
 - ◆ German Riding Pony (GRP) – some facts about the breed
 - ◆ successful transition from microsatellite- to SNP-based parentage control in Warmblood horses (WB)
- ❖ SNP-based microsatellite imputation
 - ◆ GRP reference data
 - ◆ test runs to evaluate options for a routine system
- ❖ prospects



photo: private

SNP = single nucleotide polymorphism

The breed: German Riding Pony



- around 1970s in Europe: increasing demand for riding horses for young people and children
 - formation of a new pony breed: German Riding Pony (GRP)
- breed origin:
crosses of English riding ponies with Arabian and English Thoroughbred
- breed characteristics
 - withers height 138 - 148 cm
 - all colors allowed
 - suitable for riding and sporting purposes of all kinds, especially for children
- breeding regulations for GRP
 - crosses with many breeds allowed incl. Warmblood horses

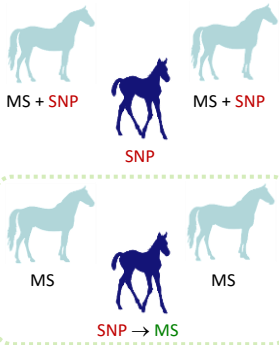


photo: private

Transition from microsatellites (MS) to SNPs



- basis of parentage control: same type of genetic markers across generations
- challenge: introduction of SNP genotyping (foals) implying two different types of markers
- possible solutions for parentage control:
 - direct transition to SNP parentage testing
 - need of extra genotyping of parents (€€€)
 - bridging the gap via imputation
 - foal: MS-imputation based on SNPs
- SNP-based MS imputation routinely used for parentage control of Warmblood horses in Germany since 2021 (about 110,000 horses until 25th August 2025)

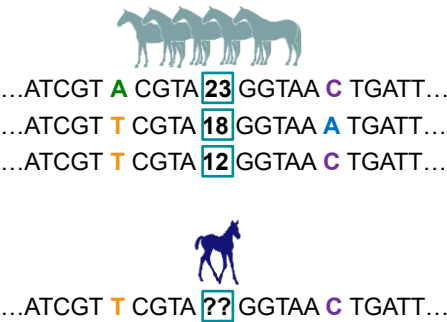


SNP = single nucleotide polymorphism, MS = microsatellite or short tandem repeat (STR)

SNP-based MS imputation I



- imputation = completion of information based on previous knowledge
- training set: dataset of horses genotyped for SNPs and MS
- certain SNP combinations occur with a certain form of a MS

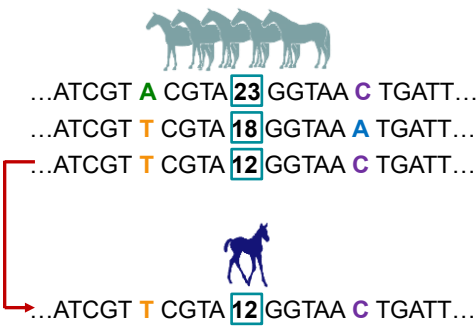


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SNP-based MS imputation II



- imputation = completion of information based on previous knowledge
- training set: dataset of horses genotyped for SNPs and MS
- certain SNP combinations occur with a certain form of a MS
- developed SNP-based MS imputation system (Nolte et al., FBN Dummerstorf) implemented as routine in 2021 (vit)



- meanwhile used by nearly all German Warmblood breeding organizations
- ✓ proof of principle: over 100,000 horses having passed the system (95 % of WB foals; 25th Aug. 2025)

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Integration of German Riding Pony data



- by the end of 2024: N = 2,110 reference GRP available (with SNP + MS)
- setup of three test scenarios with different reference populations
 - a) GRP reference (N = 1,695 horses)
 - b) WB reference (N = 12,417 horses)
 - c) GRP + WB reference (N = 14,112 horses)
- validation animals:
 - 20% randomly chosen GRP (N = 415) in (a) and (c) → 10x
 - all GRP in (b)
- calculation of the mean imputation accuracies for all analyzed MS in all scenarios
 - 12 MS-marker from ISAG core panel + 2 optional marker (in total N = 14)



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Comparison of test runs with GRP data



MS marker	scenario (a) – GRP	scenario (b) – WB	scenario (c) – GRP + WB
ASB2	0,990	0,950	0,990
HTG4	0,996	0,942	0,993
HMS2	0,968	0,886	0,981
HTG6	0,998	0,967	0,998
HMS3	0,981	0,890	0,986
HTG7	0,996	0,965	0,996
HTG10	0,992	0,959	0,996
HMS6	0,998	0,976	0,998
HMS7	0,988	0,899	0,989
VHL20	0,994	0,855	0,995
AHT4	0,992	0,891	0,993
AHT5	0,924	0,659	0,935
ASB17	0,977	0,907	0,983
ASB23	0,991	0,940	0,995

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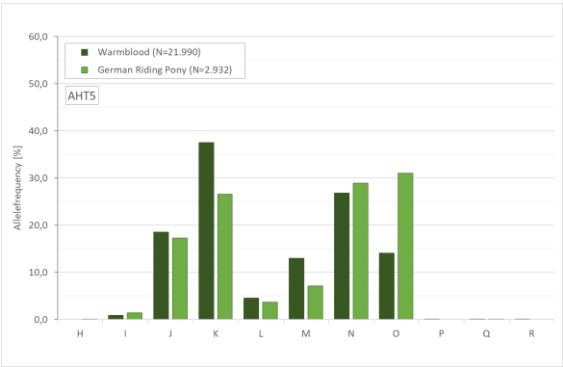
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Test runs with GRP data – results



- mean imputation accuracy: WB reference < GRP reference < GRP + WB reference
 - best results with a joint reference population of WB and GRP for N = 13 marker
- MS marker AHT5
 - lower imputation accuracy in each scenario
 - location at the beginning of equine chromosome 8 → few SNPs around
 - lowest accuracy with WB reference (0.66)
 - shift in MS allele frequencies between reference and target population
- for GRP + WB reference additional test run with WB validation animals
 - no detrimental effects on MS imputation accuracy for WB horses

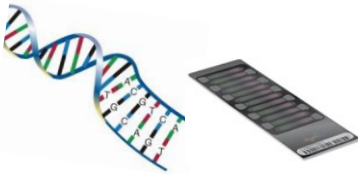


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Prospects



- integration of GRP reference data (MS + SNPs) into a joint reference population with WB
 - extended system for SNP-based MS imputation
 - basis of routine parentage control for GRP and WB since breeding season 2025
- increase of routine genome-wide SNP genotyping for parentage control → facilitating that pony breeding benefits from genomic developments
 - e.g. genetic characteristics and defects, populations structure (genetic diversity), etc.
- other (pony) breeds intend to follow and already started working on their reference populations



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Thank you!



Thanks to IAFH for providing the Warmblood data