



IT-Solutions for
Animal Production

WBFSH webinar on
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Parentage testing based on SNPs - experiences from Germany

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¹IT solutions for animal production (vit), Verden, Germany

Outline

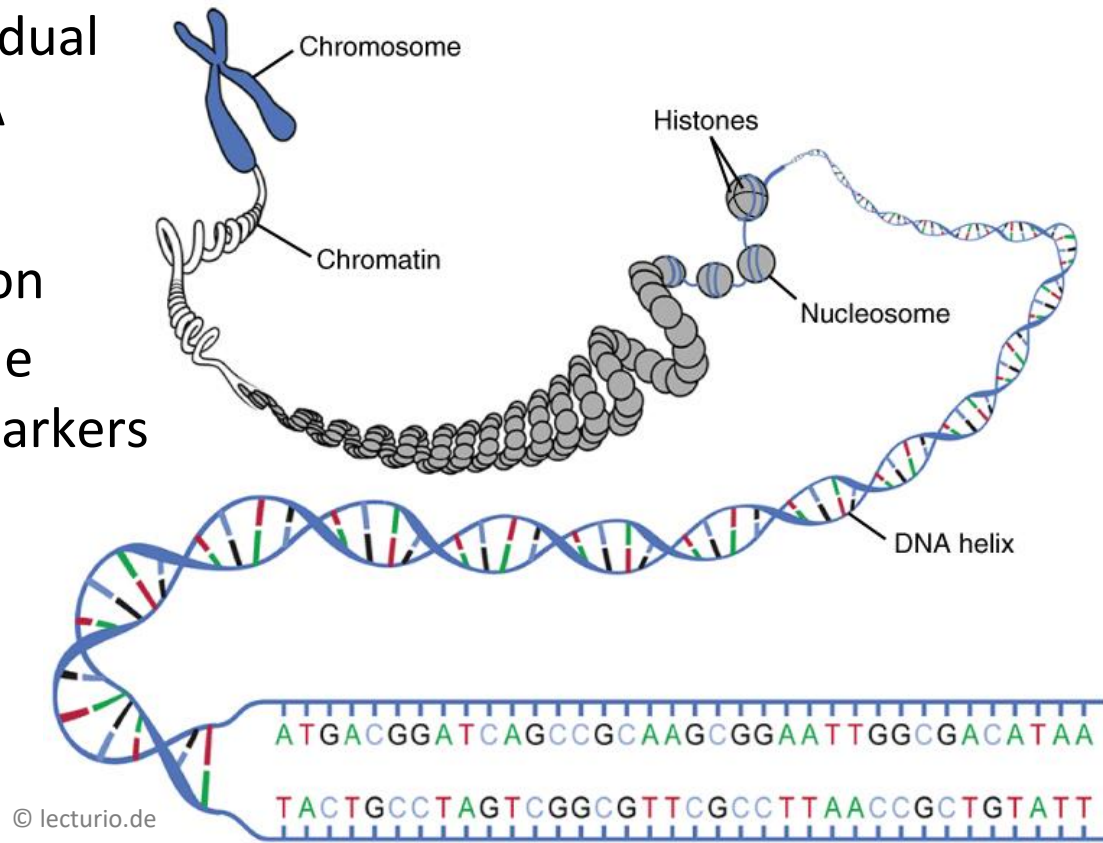
- ❖ background types of markers
 - microsatellites (MS) = short tandem repeats (STRs)
 - single nucleotid polymorphisms (SNPs)
- ❖ parentage testing
 - transfer from MS to SNP
- ❖ first experiences from routine (breeding season 2021)
- ❖ prospects



Quelle: St.Georg / ©Kiki Beelitz

Background (I)

- ❖ much of DNA is non-coding
 - much + highly individual variation in the DNA (genetic fingerprint)
- ❖ different types of variation
 - certain types suitable for use as genetic markers

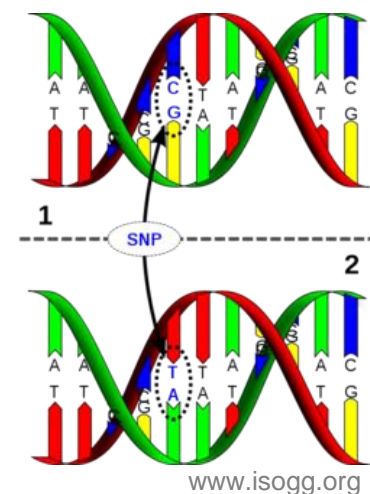


Background (II)

...GCCTAAGCGTAGTAGTAGTAGTA
 CCTAAGCGTAGTAGTAGTAGTA

- ❖ microsatellite (MS) = short tandem repeat (STR)
 - certain DNA motifs (2-6 basepairs) are repeated
 - typically 5-50 times repeated
 - many different expressions → few STRs are enough for individual characterization
 - traditionally used for parentage testing

- ❖ single nucleotide polymorphism (SNP)
 - single basepair difference within DNA sequence
 - spread over the whole genome (markers)
 - little information value per marker → need of more SNPs for a specific pattern
 - essential ‘tool’ of genomic analyses (SNP arrays)



SNP vs. MS

❖ MS

- world standard for parentage testing so far (ISAG)
- approx. 20 or less MS enough for identification
- no additional application possibilities (only parentage testing)

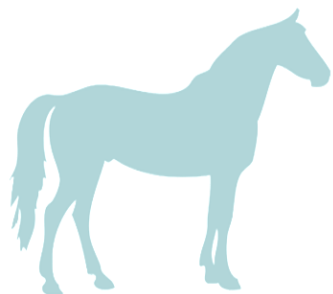
❖ SNP

- reduced costs per genotype
- faster throughput & laboratory automation capability
- variety of genomic application possibilities
 - parentage testing as only one aspect

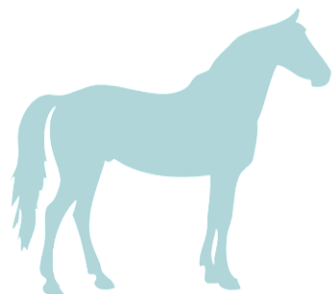


Transfer from MS to SNP (I)

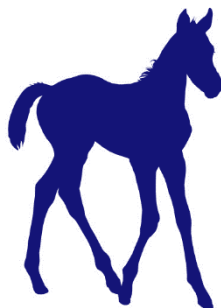
- ❖ basis of parentage testing: same type of markers across generations
- ❖ starting with SNP genotyping of foals:



MS



MS



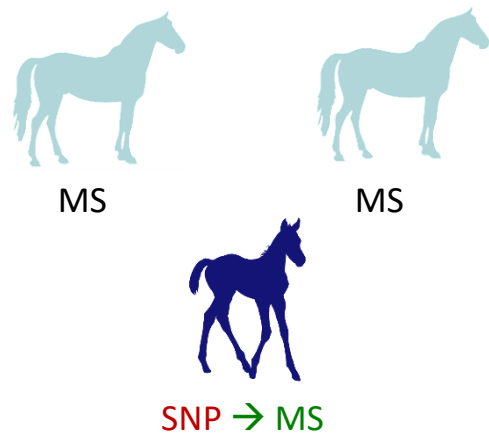
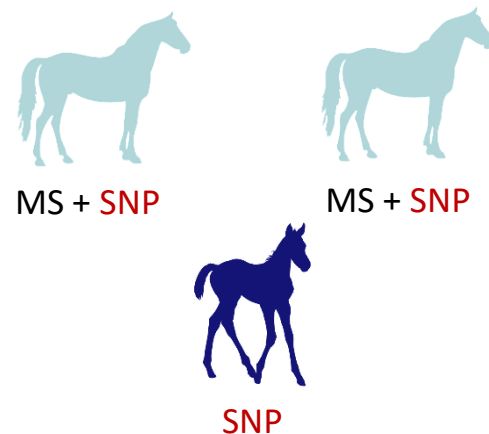
SNP

challenge: different types of genetic markers in (potential) parents and offspring

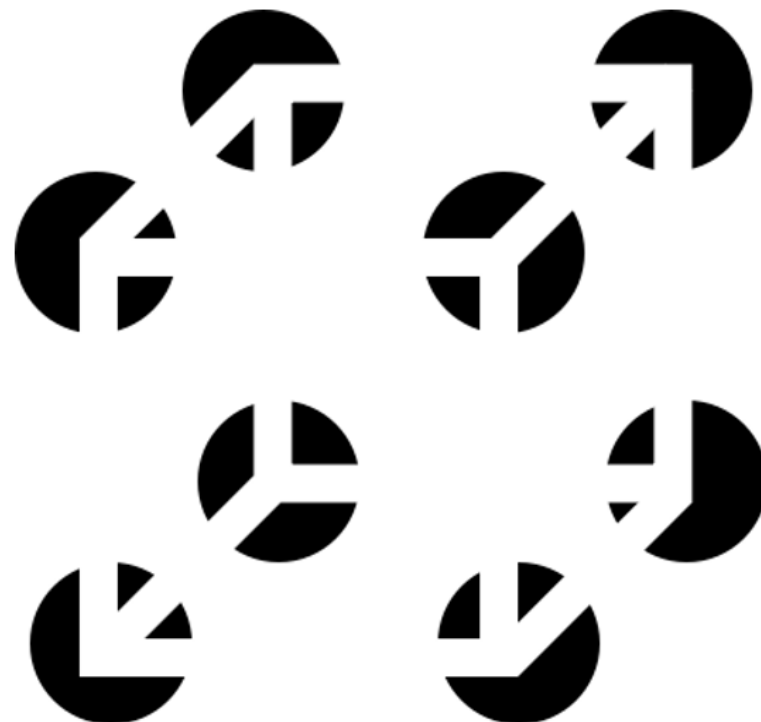


Transfer from MS to SNP (II)

- ❖ challenge of two different types of genetic markers
- ❖ two possible solutions:
 - direct transfer to SNP parentage testing
 - need of extra genotyping of parents (€€€)
 - **bridging the gap via imputation**
 - foal: MS-imputation based on SNPs

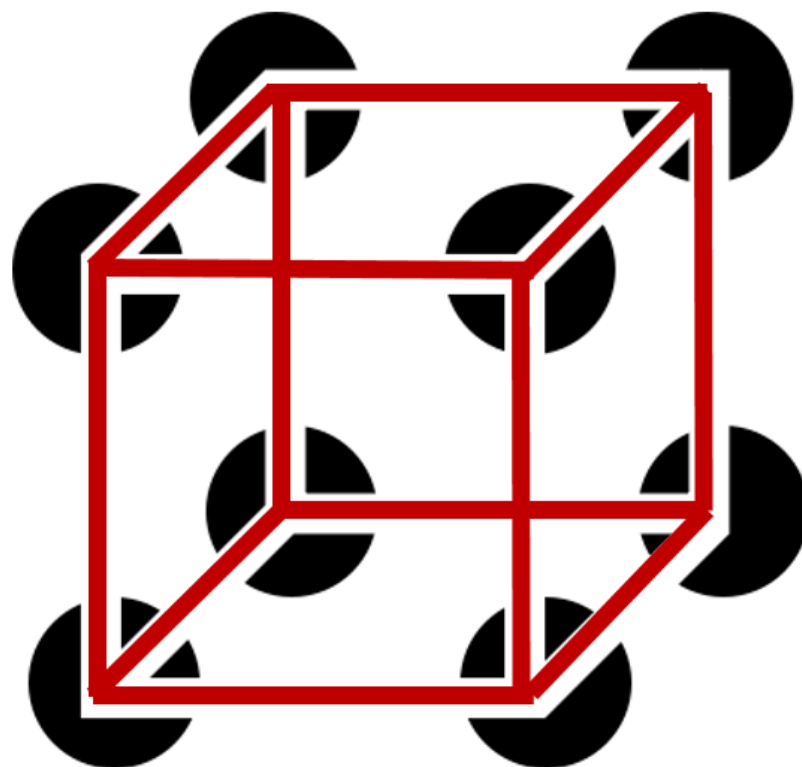


Imputation principle



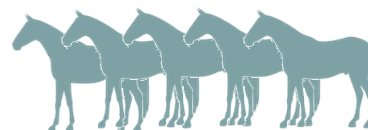
Imputation principle

- ❖ completion of information based on previous knowledge
- ❖ training set: dataset of horses genotyped for SNPs and MS



Imputation principle

- ❖ 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



...ATCGT **A** CGTA **23** GGTAA **C** TGATT...
 ...ATCGT **T** CGTA **18** GGTAA **A** TGATT...
 ...ATCGT **T** CGTA **12** GGTAA **C** TGATT...

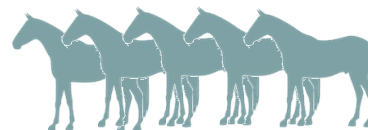


...ATCGT **T** CGTA **??** GGTAA **C** TGATT...



Imputation principle

- ❖ 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
- development of a system for imputation of MS based on SNP information
(FBN Dummerstorf, Nolte et. al)



...ATCGT **A** CGTA **23** GGTA**A** **C** TGATT...

...ATCGT **T** CGTA **18** GGTA**A** **A** TGATT...

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...ATCGT **T** CGTA **12** GGTA**A** **C** TGATT...




Routine SNP-based MS imputation

- ❖ 13 MS imputed for the routine parentage testing
 - 11 from 12 of the ISAG core panel
 - 2 optional marker
- ❖ high accuracies achieved by imputation for these MS
 - around 98-99%
 - using all SNPs on the chromosome of the MS
- ❖ comment: 14 MS included in research & development
 - one marker was 'lost': AHT5
 - positioning at the beginning of chromosome 8
 - only few SNPs available in front of this MS
 - decided not to use in routine



First experiences from routine

- ❖ transfer of the developed imputation system to routine (vit)
 - routine work for breeding season 2021 all IAFH member studbooks (TRAK, HOL, OL, OS and WESTF)
- ❖ proof of principle 
 - until 01.12.2021 N=11,047 horses passed the new system:

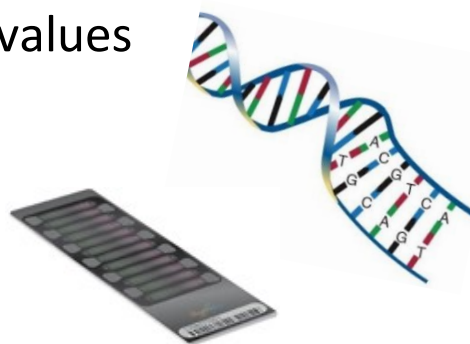
	TRAK	HOL	OL	OS	WESTF	Total
Number of samples without indications of Mendelian conflicts*	1,040	2,553	3,729	3,128	425	10,875
Number (%) of samples with indications of Mendelian conflicts	20 (1.9%)	29 (1.1%)	63 (1.7%)	54 (1.7%)	6 (1.4%)	172 (1.6%)
Total number of horses per studbook	1,040	2,582	3,792	3,182	431	11,047

*handling of Mendelian conflicts as ISAG (single discrepancies between imputed and lab-generated MS do exist)



Prospects

- ❖ cost and labor efficient way to the new system
 - bridging the gap for transfer from MS to SNP parentage control
- ❖ so far only developed for Warmblood horses
(possible for other breeds: training set needed)
- ❖ new genomic applications becoming feasible
 - SNP genotyping of all foals with optimized SNP array
commercially available Equine80select
 - genetic characteristics
 - genomic breeding values
 - etc.





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Thank you !

