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INTERNATIONAL SOCIETY FOR ANIMAL GENETICS

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Parentage & genetic testing – *the role of ISAG and future perspectives*

WBFSH webinar, Tuesday, 7 Dec, 2021

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1

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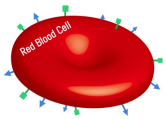
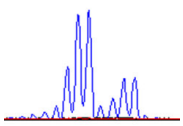
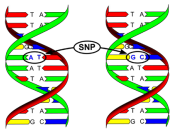
The International Society and the International Foundation for Animal Genetics (ISAG and IFAG) evolved from a series of annual workshops for comparing methods for detecting red cell antigens and variants of proteins to an organization focusing on basic and applied research on molecular genetics. The Society and the Foundation support exchange of research ideas, results and applications by organizing conferences and workshops, comparison tests and publishing Animal Genetics







2

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- ISAG's involvement is to establish rules for conducting Comparison Tests (CTs). ISAG does not endorse the performance of laboratories beyond their participation in these CTs.
- The aim of the CT is to enable laboratories to maintain high and comparable standards, and to have international agreement on nomenclature and rules for kinship testing.

3

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ISAG Comparison Test

- Determine concordance for genetic testing and analyses among laboratories
- Participating laboratories take turns to be "Duty lab" which is the laboratory responsible for preparing and shipping samples for testing
- Laboratories report results to ISAG
- ISAG use a Genotype Rating System to calculate an Absolute Ranking Score

4

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Rank	Absolute Genotype Accuracy (%)
1	100-98
2	97.9-95
3	94.9-90
4	89.9-80
5	79.9-

- Demanded by the International Studbook Committee (ISBC)
- Many Breeding Organizations demand laboratories to be in Rank 1

5

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The 64 horse chromosomes = the genome

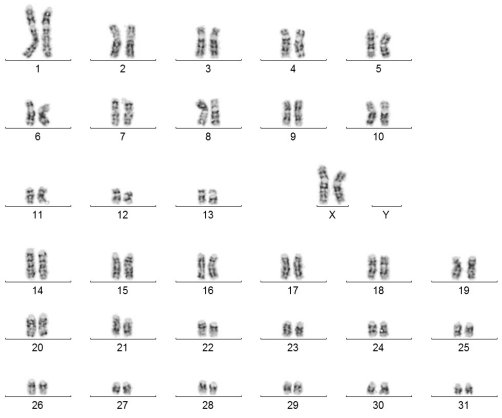
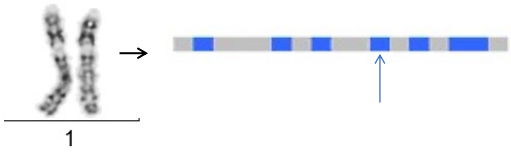


Photo: Terje Raudsepp

6

Genomic markers

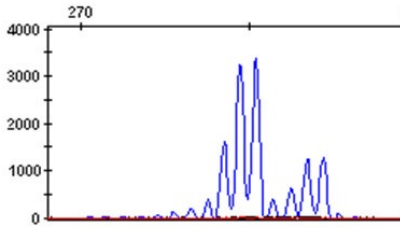


Twilight – the first horse to be sequenced, Photo: NHGRI

Microsatellites/Short Tandem Repeats (STRs)

7

Microsatellites = Short Tandem Repeats (STRs)



CTCTCTCTCTCTCTCTCTCTCTCT
CTCTCTCTCTCTCTCTCTCTCT
CTCTCTCTCTCTCTCTCTCT
CTCTCTCTCTCTCTCTCT

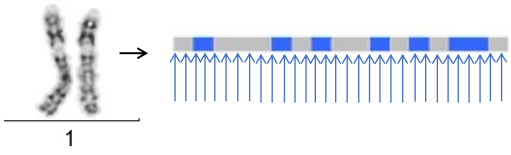
Microsatellite = Short Tandem Repeat (STR) => Genetic Length Variation

10-20 alleles / marker

12 markers in few chromosomes

8

Genomic markers

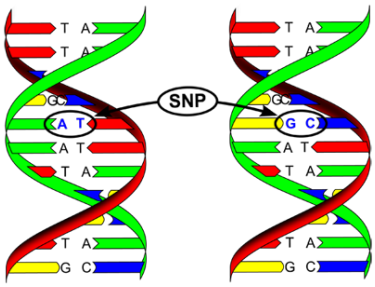


Twilight – the first horse to be sequenced, Photo: NHGRI

Single Nucleotide Polymorphisms (SNPs)

9

Single Nucleotide Polymorphism (SNPs)

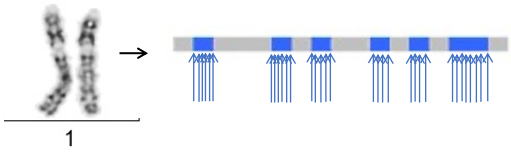


TTGTTCAAATCAGTGTCCAAAT
⇣
TTGTTCAAATCAGGGTCCAAAT

- Mutations => Genetic Variation
- Single Nucleotide Variation (SNV)
- Single Nucleotide Polymorphism (SNP)

10

Genomic markers



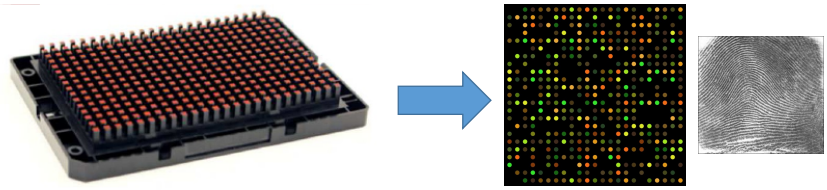
Twilight – the first horse to be sequenced,
Photo: NHGRI

1. Single Nucleotide Polymorphisms (SNPs) detected on SNP-chips/arrays
2. SNPs detected by Genotyping By Sequencing (GBS)

11

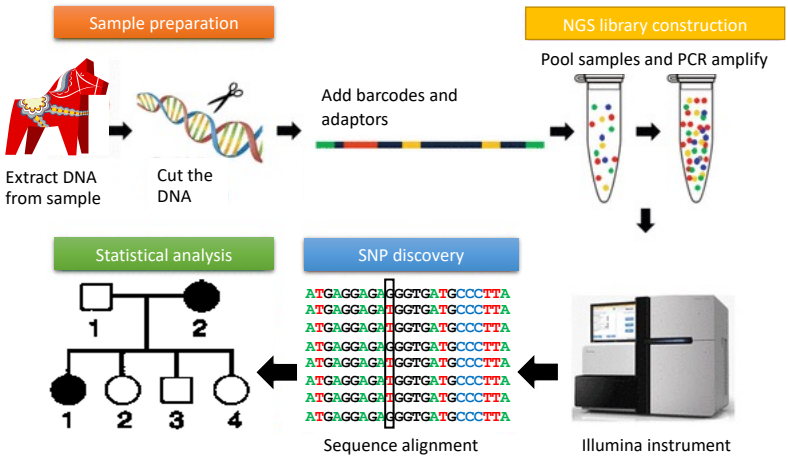
SNP-chips

- Show genetic variation between individuals
- 50,000-670,000 markers per chip
- That is, one marker every 4,000 to 40,000 basepair
- => ”Genetic fingerprint"



12

Genotyping by Sequencing (GBS)



13

From bloodtyping to genotyping

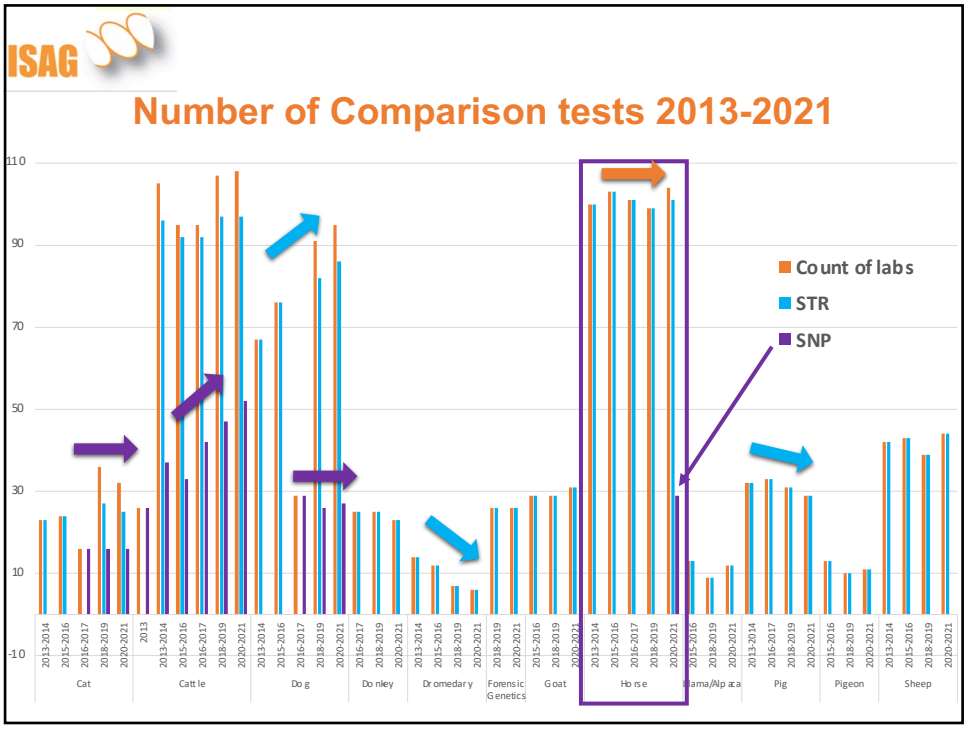
Blood Typing
- Probability of exclusion <90-95%
- Since 2008, bloodtyping of horses is no longer considered an ISAG standard for individual identification or parentage verification of horses

DNA Typing Microsatellites
- Probability of exclusion >99.99999%
- Since 2001, the ISAG standard for individual identification or parentage verification of horses (9 STRs)
- Since 2011, the ISAG standard for individual identification or parentage verification of horses (12 STRs)

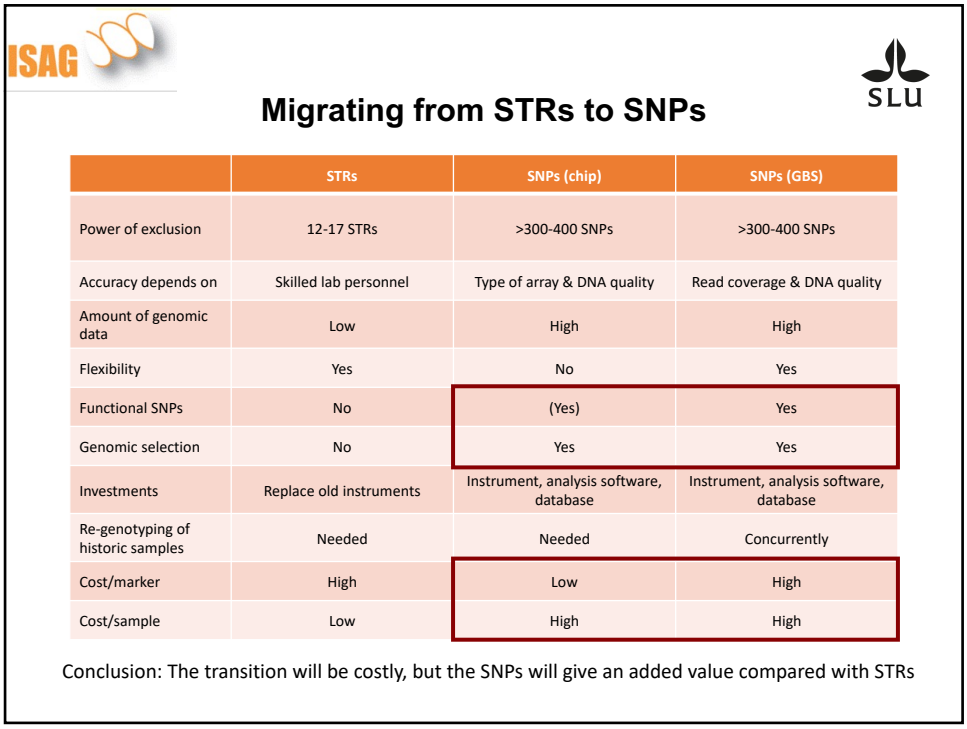
DNA Typing SNP-chip
- At least 300-400 SNPs, probably many more, needed to obtain a probability of exclusion similar to 12 STRs
- Genotyping methods discussed among typing laboratories, trial CT started in 2017
- Parent animals need to be re-genotyped

DNA Typing GBS
- Genotyping By Sequencing (GBS) also detects SNPs, but is more flexible in marker composition
- Microsatellites can be sequenced at the same time, providing an opportunity for switch from STRs to SNPs
- Data is exchangeable with SNP-chips

14



15



	STRs	SNPs (chip)	SNPs (GBS)
Power of exclusion	12-17 STRs	>300-400 SNPs	>300-400 SNPs
Accuracy depends on	Skilled lab personnel	Type of array & DNA quality	Read coverage & DNA quality
Amount of genomic data	Low	High	High
Flexibility	Yes	No	Yes
Functional SNPs	No	(Yes)	Yes
Genomic selection	No	Yes	Yes
Investments	Replace old instruments	Instrument, analysis software, database	Instrument, analysis software, database
Re-genotyping of historic samples	Needed	Needed	Concurrently
Cost/marker	High	Low	High
Cost/sample	Low	High	High

Conclusion: The transition will be costly, but the SNPs will give an added value compared with STRs

16

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Thank You for listening!

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Coreograph (SWB) e. Balougraph - Looping

Photo: Sofie Benneborg

17