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Marker based estimation of effective population size for the Swiss Franches Montagnes horse breed

C. Flury[1], B. Haase[2], C. Drögemüller[2], P.-A. Poncet[3], T. Leeb[2] and S. Rieder[1]

[1]Swiss College for Agriculture, Zollikofen, Switzerland
[2]Institute of Genetics, Vetsuisse Faculty, University of Berne, Berne, Switzerland
[3]Swiss National Stud Farm, Avenches, Switzerland

christine.flury@shl.bfh.ch

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Introduction

The Franches Montagnes horse breed (FM) is the only indigenous horse breed of Switzerland.

At least three epochs of introgression are documented for the 20st century (Poncet et al., 2006).

Pedigree based effective population size (period from 1910 – 1995) was found to be 114.5 (Poncet et al., 2006).

LD-based estimation of effective population size was subject to recently published articles (i.e. Hayes et al., 2003; Tenesa et al., 2007; Gautier et al., 2007)

➔ **Application of LD-based estimation of effective population size for the Franches Montagnes breed and comparison with estimates based on pedigree information.**

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Methods -1-

Genotypings

- 91 Franches Montagnes horses
- genotyped for 20 microsatellites, chromosome 3
- positions were derived based on horse genome assembly
- Arlequin 3.0 (Excoffier et al. 2005) for HWE-testing

LD-measures

- PowerMarker V3.25 (Liu and Muse, 2005) for derivation of D‘ and r^2

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Methods -2-

Estimation of Ne

- According Hill (1981): $E(r^2) = \frac{1}{(1+4N_e c)} + \frac{1}{n}$

Pedigree Analysis

- Pedigree: 3893 individuals (first entry: 1871)
- CFC 1.0 (Sargolzaei et al., 2006) estimation of individual F
- Slope of regression $\log_e$ (1-F) on t to estimate Ne (Woolliams, 2007)

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Results

Genotypings

locus	position	distance	N	alleles	obs.het	exp.het	p-value	std
ECA03_003	71465548		91	7	0.6264	0.6847	0.0287	0.0004
ECA03_004	73341046	0.0188	91	6	0.6264	0.7392	0.0511	0.0006
ECA03_005	75672813	0.0233	91	6	0.7033	0.7650	0.0684	0.0006
ECA03_021	77033520	0.0136	91	4	0.4835	0.4238	0.3695	0.0015
MS_Kit_Intron	77754626	0.0072	91	4	0.2857	0.3075	0.1760	0.0009
ASB23	79279361	0.0152	91	4	0.6813	0.6720	0.4966	0.0015
ECA03_006	81330672	0.0205	91	7	0.7363	0.7082	0.2851	0.0013
ECA03_007	83478009	0.0215	90	3	0.5889	0.5831	0.9723	0.0005
ECA03_009	87055874	0.0358	91	6	0.7802	0.7009	0.6861	0.0012
ECA03_010	88932363	0.0188	90	7	0.6000	0.7715	0.0004	0.0001
ECA03_011	91235922	0.0230	86	5	0.4884	0.7463	0.0000	0.0000
ECA03_012	93130623	0.0189	91	4	0.5824	0.5573	0.3918	0.0014
ECA03_013	95290881	0.0216	91	3	0.0550	0.0540	1.0000	0.0000
ECA03_014	97251837	0.0196	91	7	0.4725	0.4501	0.8804	0.0007
AHT097	99036601	0.0178	88	5	0.5341	0.5232	0.4757	0.0015
ECA03_015	100808371	0.0177	91	3	0.4286	0.4580	0.8529	0.0011
ECA03_016	102921911	0.0211	88	8	0.4205	0.8223	0.0000	0.0000
ECA03_017	104888793	0.0197	91	5	0.3846	0.4142	0.4230	0.0011
ECA03_018	106841002	0.0195	91	5	0.6374	0.6204	0.5774	0.0013
ECA03_019	108571784	0.0173	89	8	0.4719	0.7463	0.0000	0.0000
Mean		**0.0195**		**5.35**	**0.5294**	**0.5874**		

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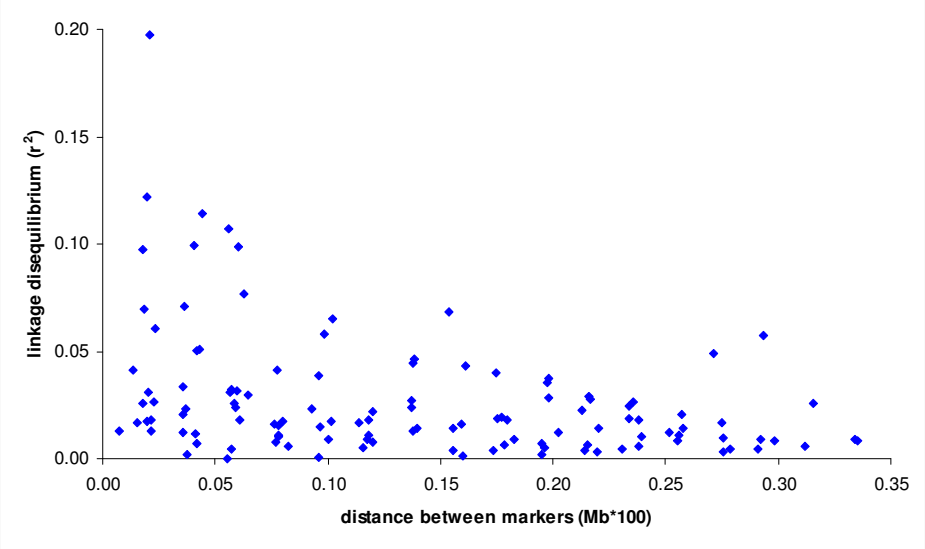
Results – marker information

LD – measures

N	8	12	14	11	6	9	7
Bin	**0 – 2**	**2 – 4**	**4 – 6**	**6 – 8**	**8 – 10**	**10 – 12**	**12 – 14**
Distance (Mb)	1.62	2.90	5.09	7.21	9.37	11.18	13.54
D‘	0.4735	0.4354	0.3539	0.3160	0.3352	0.3491	0.3543
r^2	0.0503	0.0424	0.0421	0.0311	0.0237	0.0193	0.0252

Results – marker information

LD – measures: r^2



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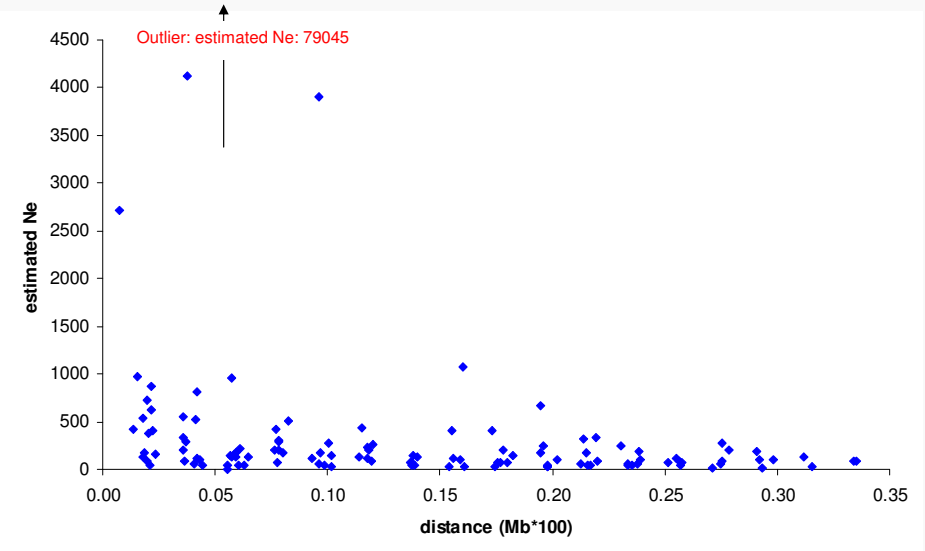
Results – marker information

Estimation of Ne



Results – marker information

Estimation of Ne

Epochs of import:

1890 – 1920 Arabian, Warmblood, Draft Horses

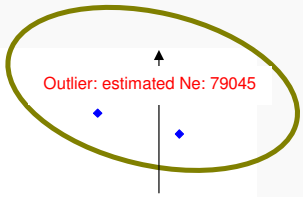
10 – 14 Gen.

1950 – 1965 Arabian, Warmblood

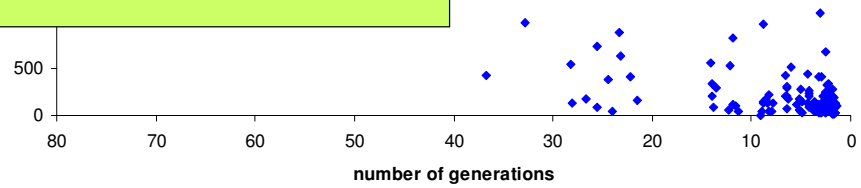
5 - 7 Gen.

1985 – 1990 Warmblood

2 Gen.



INTROGRESSION ?



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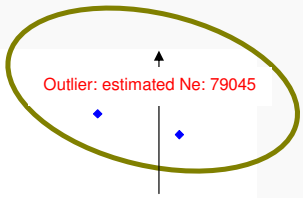
Results – marker information

Estimation of Ne



INTROGRESSION ?

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Results – pedigree information

Pedigree Analysis

	#	# of inbreds	average F	max / min F	# generation equivalents
ancestors	3802	2270	0.019	0.261 / 5e-7	11.677
sample	91	91	0.049	0.111 / 0.021	12.607

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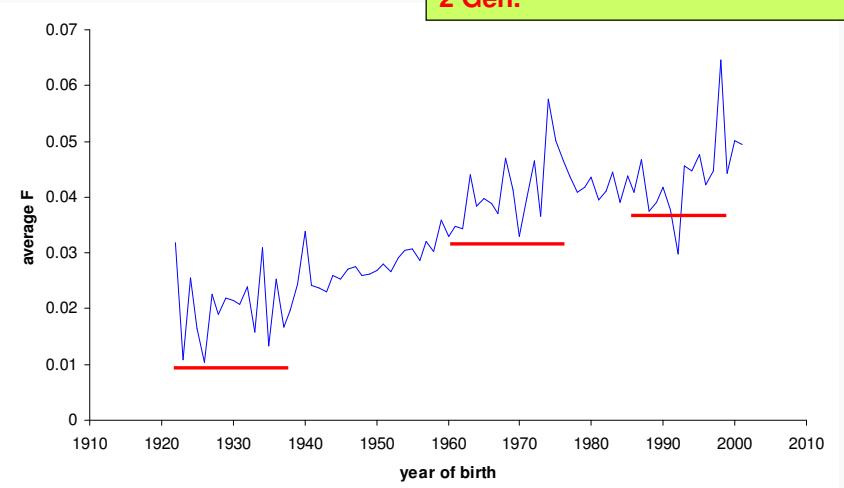

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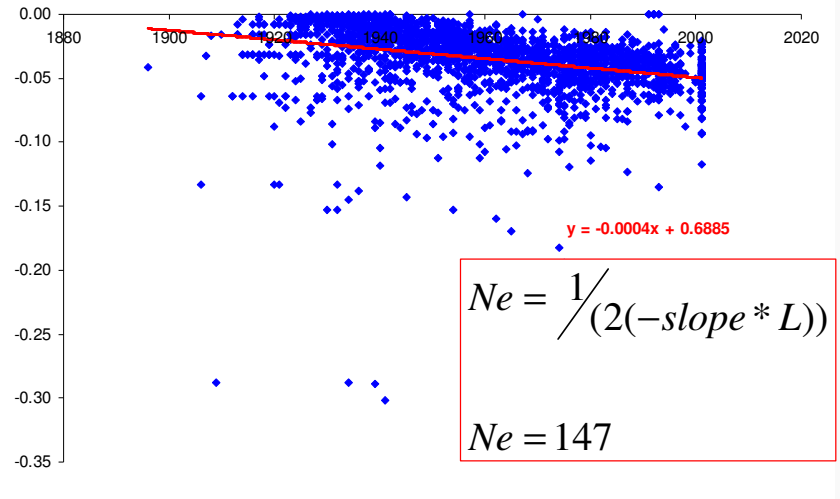
Results – pedigree information



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Conclusions

In total 120 marker pairs were informative. This leads to a restricted number of pairs per distance bin.

➔ **Genotyping of additional markers is necessary.**

Estimated Ne showed a decreasing trend with increasing distance over the last 70 generations, however, with several outliers. These fall into the „epochs of import“.

➔ **Introgression led to an increase in Ne.**

➔**Pedigree analysis confirmed the effect of introgression.**

➔ **Estimates for Ne** are in the range of **114.5** (Poncet et al. 2006) **to 126** (marker based estimates, 91 ind) **to 147** (pedigree based estimates, 91 ind)**.**

The three epochs of import led to an increase of genetic variability within population. For the future, optimum contribution mating is highly recommended to maintain actual genetic variability.

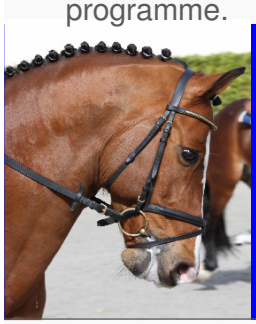
Outlook

Complete retrospective analysis by:

- Genotyping additional markers (other regions, chip data).
- Haplotype derivation and estimation of Ne based on CSH-method.

Prospective:

- Development of optimum contribution method for the FM-breeding programme.



Thank you!