

# SNPs associated with osteochondrosis in horses on different stage of training

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# Aim

- Looking for the genetic background of osteochondrosis
- Looking for the best scale and moment for OCD evaluation



# Definition of osteochondrosis

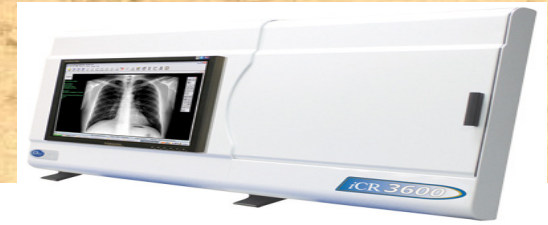
- Polish definition
- other definitions

**OC** lesions, disturbances, flattening

**OC** - **D**issecans

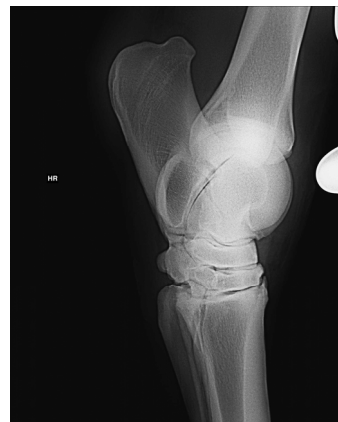


# Material and methods



All stallions (87) and mares (114) tested during two successive years on performance test stations were examined for OC(D) before and after performance tests.

Horses were x-rayed using digital equipment in fetlocks (1 image), stifles (2 images) and hocks (1 image) from right and left side.



## Material and methods

- **Scale used:**

- <b>OCD</b> (+)	3	<b>1</b>
- no signs of <b>OC(D)</b> (-)	0	<b>0</b>
- some marks of <b>OC(D)</b> (+/-)	1-2	<b>0</b>

**All joints were taken into account, one single note was given for every horse.**

# Material and methods

## Genotyping

Genomic DNA was isolated from the blood samples by the MasterPure Genomic Purification Kit (Epicentre)

DNA was then used to genotype each horse using the Illumina Neogen Equine Community Array, which consists of 65 157 SNP markers evenly distributed across 31 autosomes in average spacing of 43,2 kb.

Total call rate achieved the value of 99.75%

The quality of SNP clusters was analyzed by using GenomeStudio (Illumina).

63 946 SNPs were used for statistical analysis

# Material and methods

## Statistical analysis - SNP

Statistical analysis including Cochran-Armitage test and logistic regression assuming an additive model of inheritance (except SNP effect, also training centre, sex, age, breeder and pedigree information ) were used.

Testing SNP effects:

- Cochran-Armitage test

$$T = (N_{ABsick}N_{healthy} - N_{ABhealthy}N_{sick}) + 2(N_{BBsick}N_{healthy} - N_{BBhealthy}N_{sick})$$

- Logistic regression

$$\text{logit}(P_{sick}) = \beta_1 X_{SNP} + \beta_2 X_{\text{training centre}} + \beta_3 X_{\text{sex}} + \beta_4 X_{\text{breed}} + \beta_5 X_{\text{age}} + \beta_6 X_{\text{breeder}} + \beta_7 X_{\text{sire}} + \beta_8 X_{\text{dam}}$$

# Material and methods

## Statistical analysis - $h^2$

Additionally heritability was calculated at two different stages of training. The animal model with sick/healthy as dependent variable was used with the fixed effect – training centre, breed, sex, breeder, age was fitted as a fixed effects and animal as random effect. The pedigree file was used. The Monte Carlo Markov Chain (MCMC) method was applied. Heritability was counted as the following ratio:

$$\frac{VAR(A)}{VAR(A) + VAR(R) + \pi^{2/3}}$$

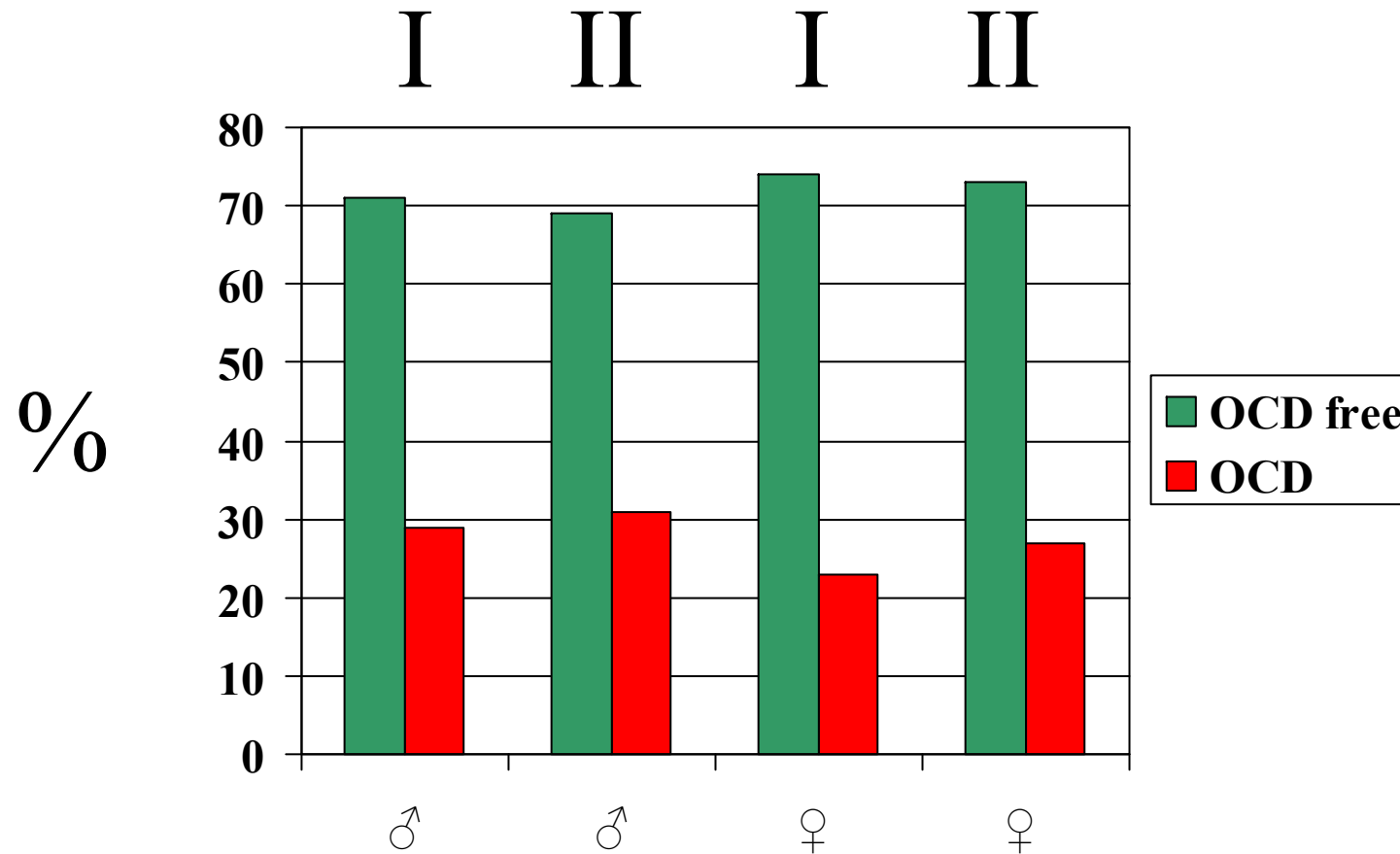
Heritability estimated on the observed scale 0-1 were transformed to continuous scale using the equation:

$$h^2 = h_{0,1}^2 \left[ \frac{(1-p)}{i^2 p} \right]$$



# Results

## OCD evaluation



# Results

## I

## II

### SNP evaluation

CUHNSNP001510	.000051490	BIEC2_1074231	.000018822
UKUL1185	.000051490	BIEC2_677587	.000047403
BIEC2_998451	.000105411	BIEC2_1061013	.000115934
BIEC2_138865	.000106818	BIEC2_164636	.000136555
BIEC2_907143	.000110218	BIEC2_1075002	.000173901
BIEC2_55129	.000158218	BIEC2_1005365	.000212600
BIEC2_827168	.000190845	BIEC2_644797	.000238888
BIEC2_1061013	.000191701	BIEC2_55129	.000255875
BIEC2_66407	.000225441	BIEC2_952637	.000286266
BIEC2_769816	.000254224	BIEC2_66407	.000291625
BIEC2_960044	.000269643	TBIEC2_320567	.000292482
BIEC2_1062127	.000277862	UKUL613	.000294517
BIEC2-353266	.000283181	BIEC2_873544	.000348315
UKUL1686	.000313887	BIEC2_103998	.000373418
BIEC2_353080	.000347359	BIEC2_306489	.000383204
BIEC2_658643	.000356241	BIEC2_503078	.000391158
BIEC2_391945	.000445754	BIEC2_481125	.000462345
BIEC2_522473	.000463300	BIEC2_481127	.000462345
BIEC2_1006070	.000549447	BIEC2_755946	.000482501
BIEC2_755946	.000607778	BIEC2_645189	.000531445

The collection of significant SNP was not the same in the both investigations.

# Results

## SNP evaluation

### I and II

BIEC2_1061013	.000191701	.000115934
BIEC2_55129	.000158218	.000255875
BIEC2_66407	.000225441	.000291625
BIEC2_755946	.000607778	.000482501

Only four SNP (out of 20 observed as important for each term) were involved in OCD for both stage of training. Multiple testing lowered the significance of SNP effects.

# Results

## $h^2$ evaluation

I - 0.303

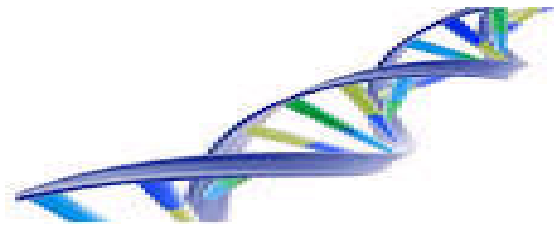
II - 0.265

Heritability estimations are not exactly the same on both stages of the training, however standard errors are not known.

## Conclusions

1. Not the same SNPs are associated with the OCD status of horse on different stages of training. 4 SNP are significant for both stages of training.
2. Heritability estimation for the horse OCD status does not reach the same level on different stages of training.

That might bias different evaluation of the horse OCD health status between times and places by using BVE and genotyping. Comparison of different research need exactly information on the training status of evaluated horses. It seems useful to evaluate the horse before any kind of the training or conditioning.



**Thank you for your attention!**