Extent of genome-wide linkage disequilibrium in Pinzgau cattle

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Aim of our study

„Evaluation of genetic diversity for management of Slovak Pinzgau cattle"

- Genetic diversity parameters
  - **Inbreeding coefficient**
  - **Effective size of population**
  - Genetic distances
    - Similarity **within** populations
    - Similarity **among** individuals / breeds

- Pedigree analyses
- Molecular analyses – SNPs
                – microsatellites
                – SNP chip arrays

- import – 1894

- alpine breed

- dual-purpose

Slovakian
Pinzgau
cow
Slovakia

- unique traits

- cultural heritage

- endangered - in 1990 – **90,000** cows
              - in 2012 – **3,101** herdbook cows
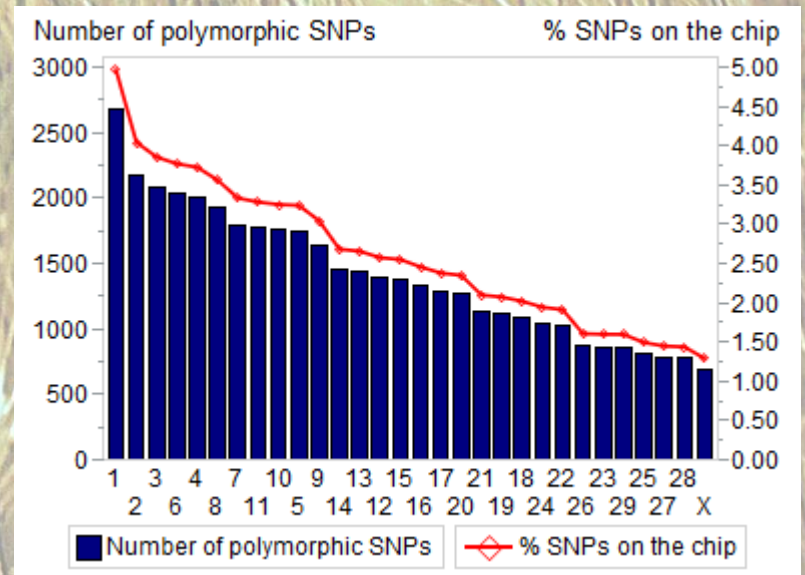
LD – linkage disequilibrium

- **high-density SNP arrays** – genomic selection evaluation

- **GWAS** – to identify the genomic regions (affecting the variation of phenotypic traits) and detecting the causal mutation

- **LD** – rate of decline with genetic distance between loci within a population
  - to determine the no. of markers identifying **QTL**
  - the chromosomal regions influenced by **selection**
  - managing genetic resources and **diversity**
  - non-random association of alleles at 2 or more loci – evolutionary history (**ancestral EPS**)
  - affected by:
    - genetic linkage
    - the rate of recombination and mutation
    - genetic drift
    - non-random mating
    - selection

Methods

- **19 bulls** – purebred Pinzgau

- Illumina Bovine**SNP50** v2 BeadChip – 54,609 SNPs

- **Quality cont**rol
  – sex chromosomes
  – missing genotypes
  – low call rate

- **MAF**
- LD – pairs of **adjacent** SNPs
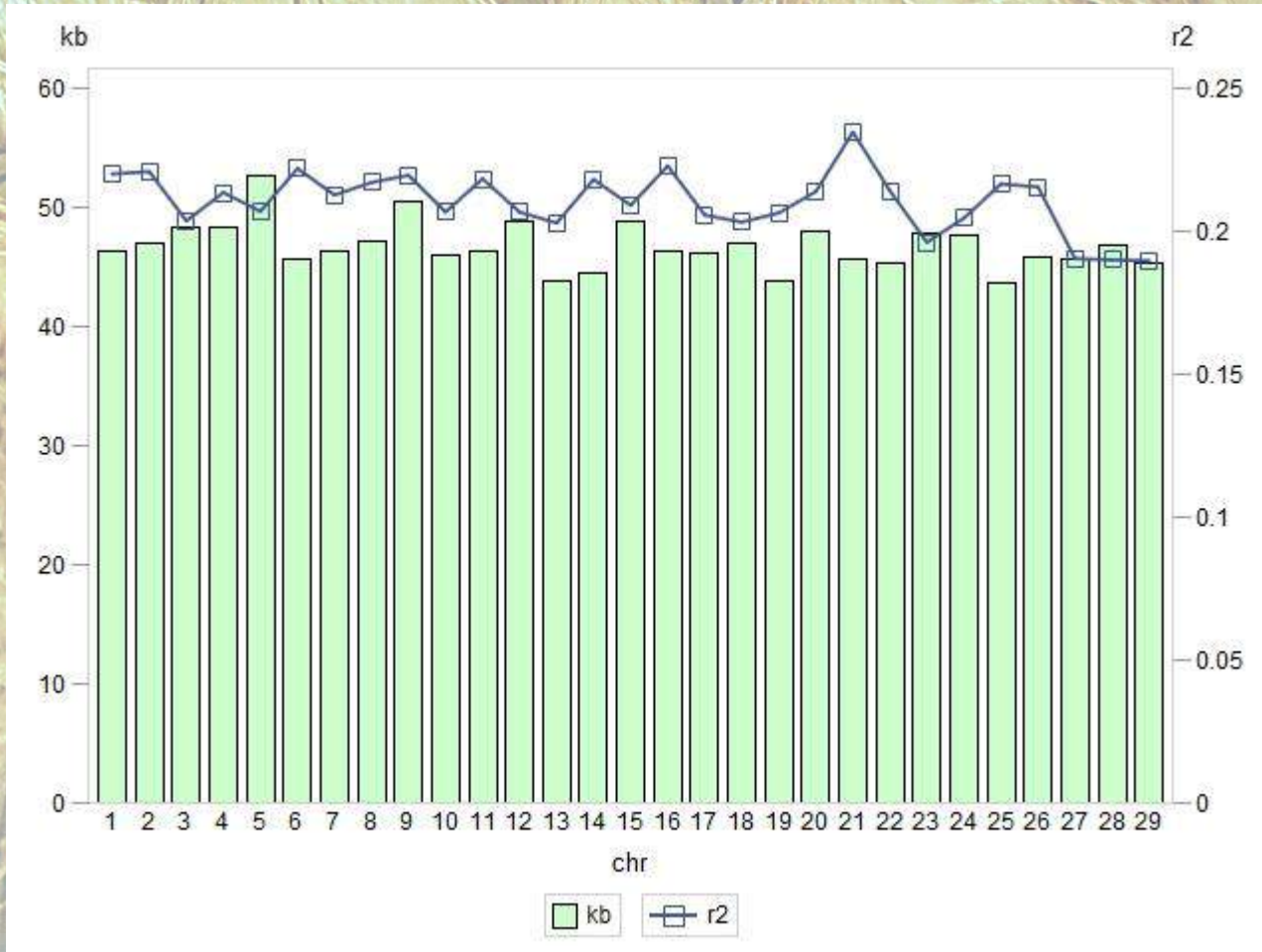 – r^2 = 0.01-0.99

MAF – minor allele frequency

- refers to the frequency at which the least common allele occurs in a given population

- Using treshold MAF are excluding monomorphic SNPs

- 42,248 polymorphic SNPs

Results

46.89 ± 47.48 kb

BTA 25
BTA 5

0.21 ± 0.218

BTA 29
BTA 21

Distribution of average gaps and *r²*-values between adjacent markers across autosomes

Results

Pairwise linkage disequilibrium (r^2) for adjacent SNPs at various distances pooled over all autosomes

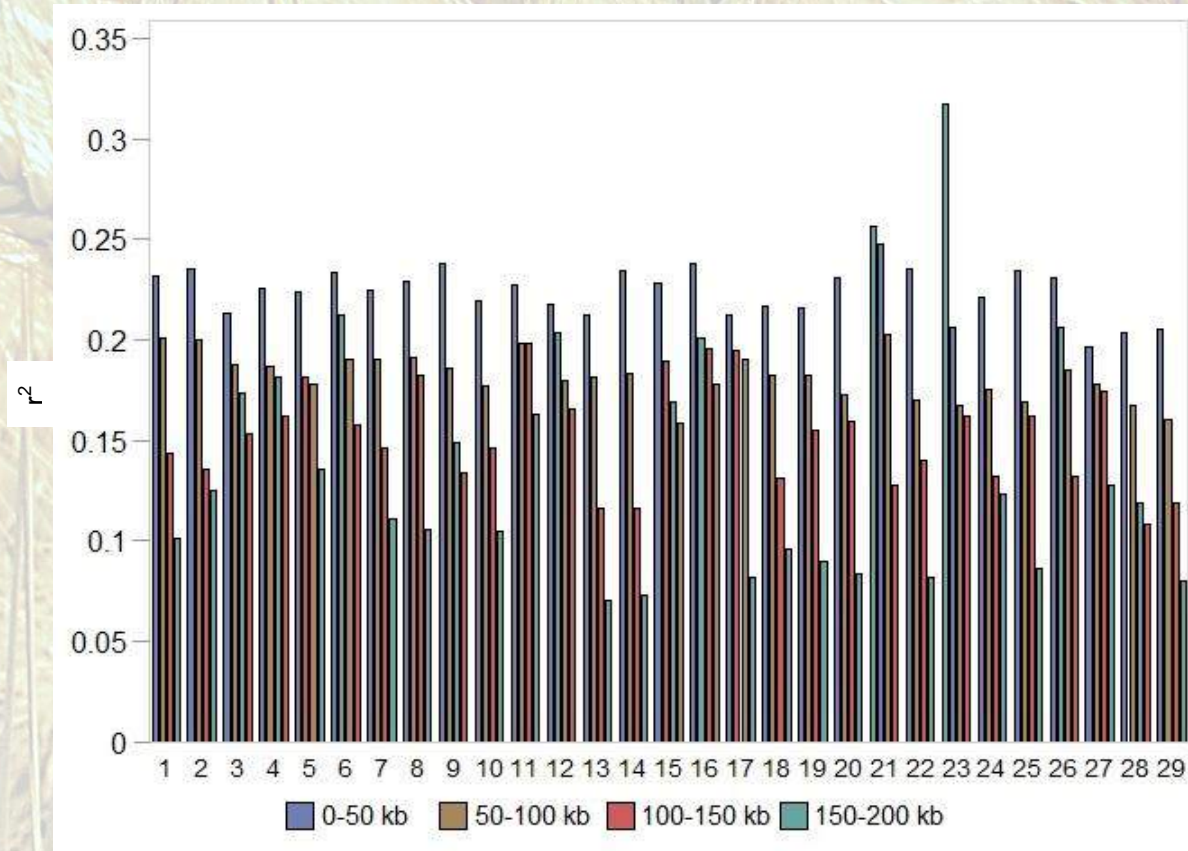| Bin (kb)	Pairs (n)	$r^2 \pm$ SD	Minimum	Maximum	Lower and upper 95% CI
0-50	22618	**0.225 ± 0.226**	0.01	0.93	0.222 ± 0.228
50-100	6564	**0.184 ± 0.196**	0.01	0.92	0.179 ± 0.188
100-150	1226	**0.155 ± 0.167**	0.01	0.9	0.146 ± 0.165
150-200	378	**0.144 ± 0.163**	0.01	0.86	0.127 ± 0.160
200-250	154	**0.138 ± 0.153**	0.01	0.76	0.114 ± 0.162
< 250	123	**0.117 ± 0.122**	0.01	0.59	0.095 ± 0.139

Results

0-50 kb
0.2-0.26

BTA 27
BTA 21

150-200 kb
0.07-0.33

BTA 13
BTA 23

Chromosome-wise average r^2-values between adjacent SNPs and the decay of linkage disequilibrium with increasing physical distances for the four smallest distance bins evaluated
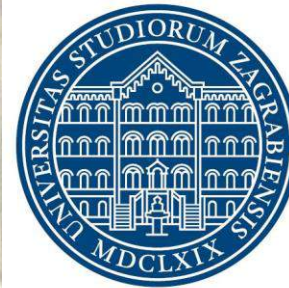
Conclusions

- the higher extent of LD across chromosomes is caused by smaller number of SNPs and few pairs located on the same genes (e.g. **KHDRBS2** on chromosome 23 and **LPHN3** on chromosome 6)

=> physical distance is not a perfect indicator of the strength of linkage, especially in the case if adjacent pairs are located on the same gene

- the application of bovine genotyping arrays is usable tool for genetic diversity parameters estimation

- presented methodology of LD extent examination is applicable for every livestock population

Further investigation

- LD – syntenic SNP pairs
- ESP
- other breeds for comparison
  - Holstein, Simmental, Austrian Pinzgau, Ayrshire, MRI (Meuse Rhine Issel)

- Introgression of other breeds
- Age of admixture

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Thank you for your attention! ;)