

Linkage disequilibrium pattern in Polish Holstein cattle

Adrian Drozd  
PhD supervisor:  
Joanna Szyda

Material

SNP chip  
SNP distribution  
SNP density  
SNP frequencies

Methods

LD measure  
LD blocks

Results

LD  
LD blocks

Conclusions

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20.05.2010

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## Animals

- Polish HF
- bulls
- 1'228
- 1997 to 2003

## SNP microarray

- Illumina BovineSNP50 BeadChip
- 54'001
- 66'259'227 total records
- 52'336 (96.9%) mapped to chromosomes
- 1'677 SNPs have no information on a chromosome location

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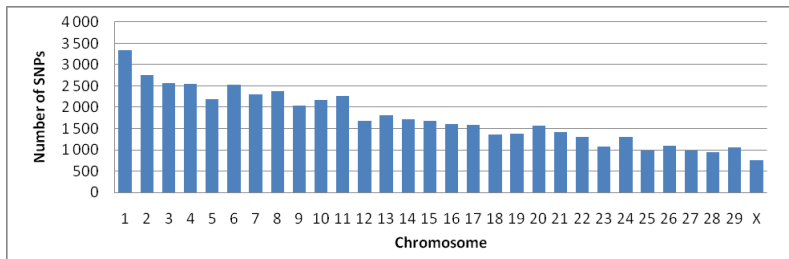
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<b>Chromosome</b>	<b>1</b>	<b>2</b>	<b>3</b>	<b>4</b>	<b>5</b>	<b>6</b>	<b>7</b>	<b>8</b>	<b>9</b>	<b>10</b>
<b>SNP</b>	3343	2764	2566	2541	2181	2535	2294	2369	2036	2179
<b>Chromosome</b>	<b>11</b>	<b>12</b>	<b>13</b>	<b>14</b>	<b>15</b>	<b>16</b>	<b>17</b>	<b>18</b>	<b>19</b>	<b>20</b>
<b>SNP</b>	267	1683	1802	1722	1688	1606	1585	1351	1378	1564
<b>Chromosome</b>	<b>21</b>	<b>22</b>	<b>23</b>	<b>24</b>	<b>25</b>	<b>26</b>	<b>27</b>	<b>28</b>	<b>29</b>	<b>X</b>
<b>SNP</b>	1419	1299	1083	1294	987	1086	977	942	1048	747

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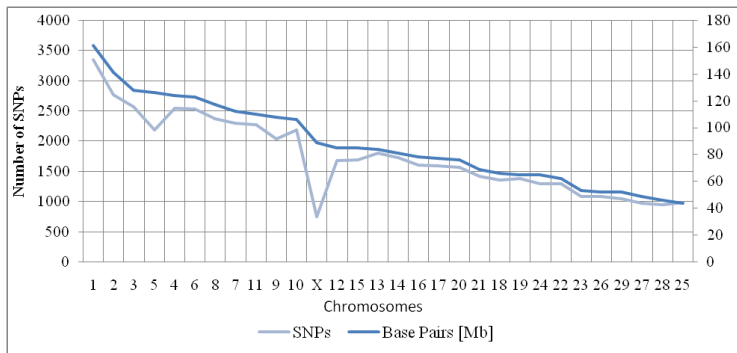


Figure: Left y axis: number of SNPs, right y axis: chromosome length [Mbp]

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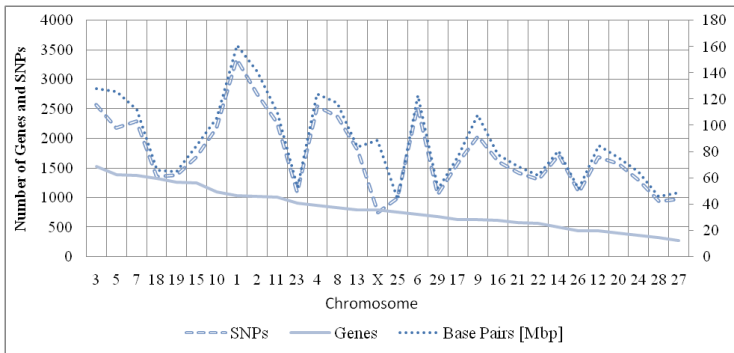


Figure: Left y axis: number of genes and SNPs, right y axis: chromosome length [Mbp]

# Marker distances

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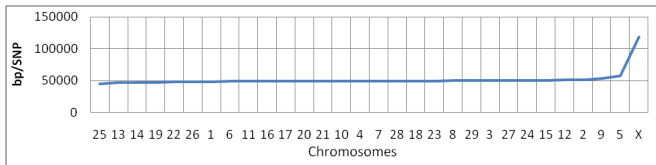
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## SNP microarray

- $51'486 \pm 12'969$  between two adjacent SNPs on average
- little variation in the average SNP spacing among autosomes
- 119'143 bp/SNP for BTAX



# Allele frequencies

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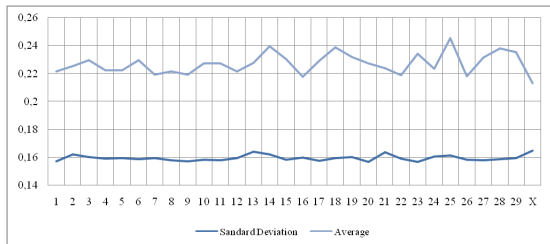
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## MAF

- All SNPs biallelic
- $0.227 \pm 0.0076$  average MAF for 30 chromosomes
- average MAF for each chromosome varied from 0.213 for BTAX to 0.245 for BTA25



# Allele frequencies

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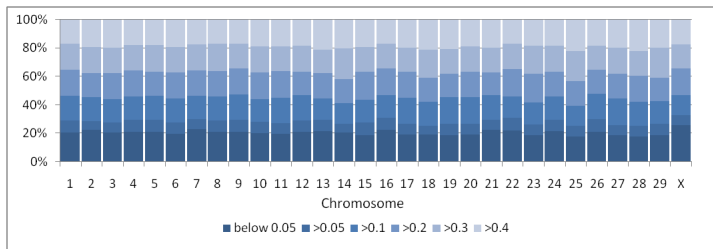


Figure: Y axis: % of MAF in each 6 classes

# Linkage Disequilibrium

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$r^2$  correlation

$$r^2 = \frac{D^2}{p_1q_1p_2q_2}$$

- PLINK 1.07
- $MAF \geq 0.05$
- measured separately for each chromosome

# Linkage Disequilibrium

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## LD block definition

- standard confidence intervals (e.g. Gabriel *et al.*) implemented in Haploview 4.2
  - confidence interval for strong LD: 0.7 - 0.98
  - fraction of strong LD in interval: 95%
- measured separately for each chromosome

# LD histogram

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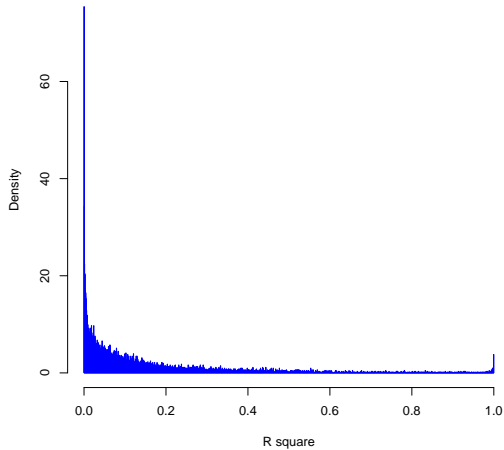
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**BTA14**



# LD histogram - BTA1-BTA9

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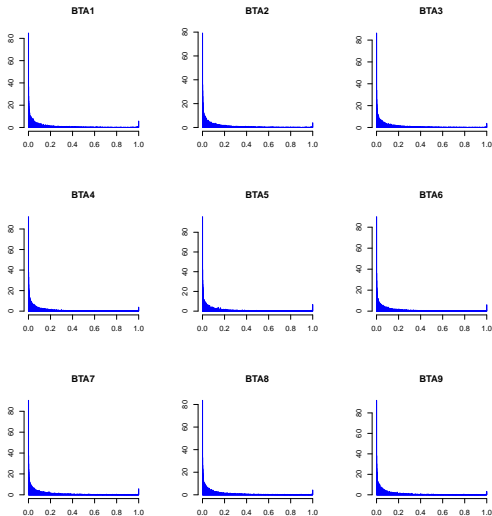
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# LD histogram - BTA10-BTA18

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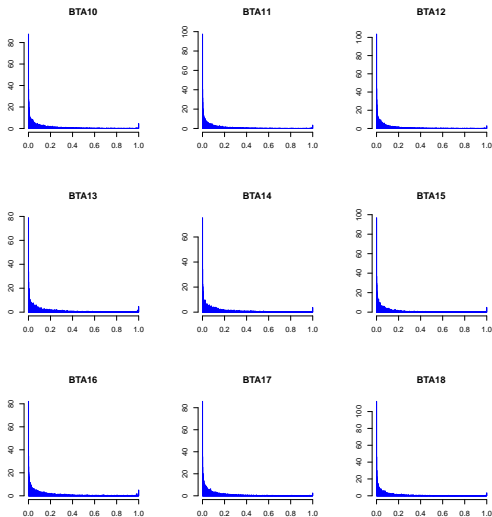
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# LD histogram - BTA19-BTA27

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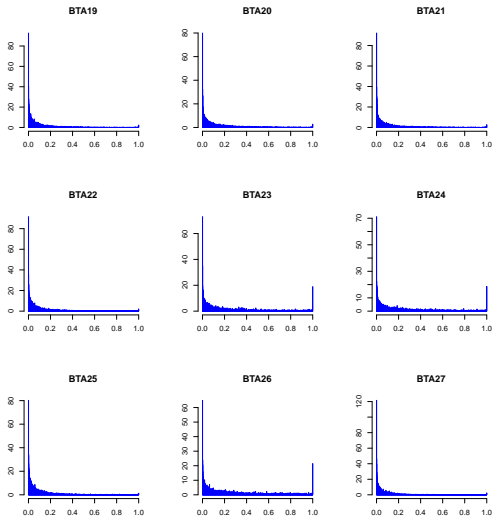
Material  
SNP chip  
SNP distribution  
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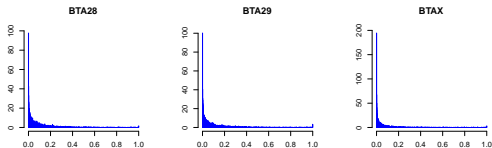
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# LD histogram - BTA28-BTAX



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# LD distribution

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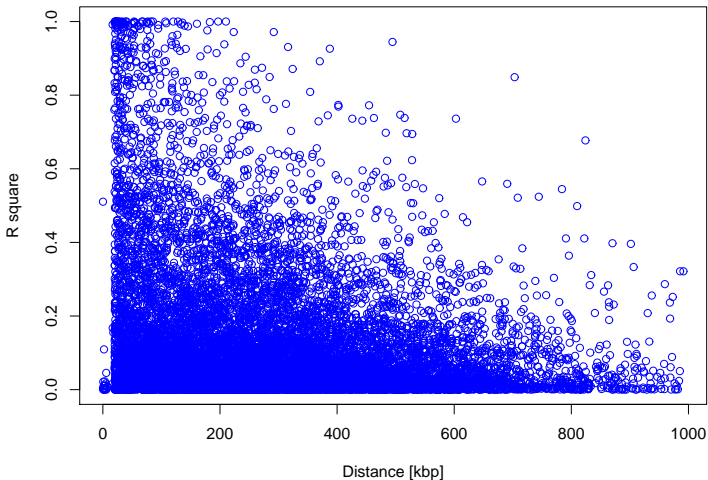
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**BTA14**



# LD distribution at low distances

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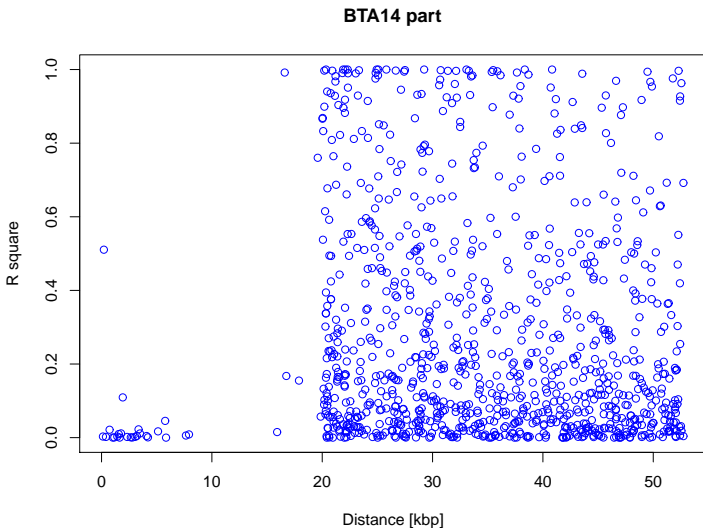
Adrian Drozd  
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# LD frequency

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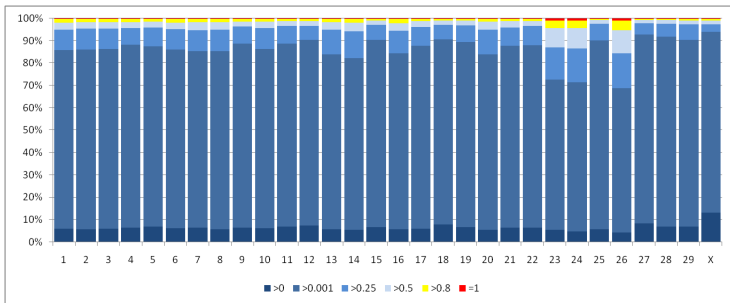


Figure: Six classes of  $r^2$

# LD blocks

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## BTA1

- number: 53
- average length: 186.2 kbp
- chromosome coverage: 6.21 %

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## LD distribution

- Low SNP density = lower LD
- many SNP pairs are equal to 1 = an evidence of a real LD blocks, even of natural/artificial selection

## LD blocks

- Bovine chip resolution is too low (compare to HapMap Project)
- reconstruction of a real LD blocks in cattle isn't possible

## Other

- $r^2$  describes a real LD patterns quite good

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Thank you . . .