



Mouse Diversity Array SNP Genotyping of C57BL/6JRj strains

- 1. Data and objectives**
- 2. Result summary**
- 3. Venn Diagrams**
- 4. Functional classes of SNPs**
- 5. Clustering of samples**
- 6. Conclusion**

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Data and objectives

► Data

Sample name	Short name	Sex	Origin
SNP_mDIV_A4-SNP11_477_090311	Jax_A4_477	Male	Jackson Laboratory
SNP_mDIV_A5-SNP11_478_090311	Jax_A5_478	Female	Jackson Laboratory
7840_9192.1314_20140624	BioLabs_9192	Male	Janvier Labs
7840_9193.1314_20140624	BioLabs_9193	Female	Janvier Labs

► Objectives

1. To determine the genetic profile of the 4 C57BL/6JRj strains listed above
2. Three comparisons:
 - Comparison of genetic profile of Jackson Laboratory samples (Jax_A4_477 and Jax_A5_478) vs. the reference C57BL/6J;
 - Comparison of genetic profile of Atlas BioLabs samples (BioLabs_9192 and BioLabs_9193) vs. the reference C57BL/6J;
 - Comparison of the Jackson Laboratory samples vs. Atlas BioLabs samples
3. Clustering of C57BL/6JRj strains with list of mouse strains selected by Janvier Labs

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Result summary

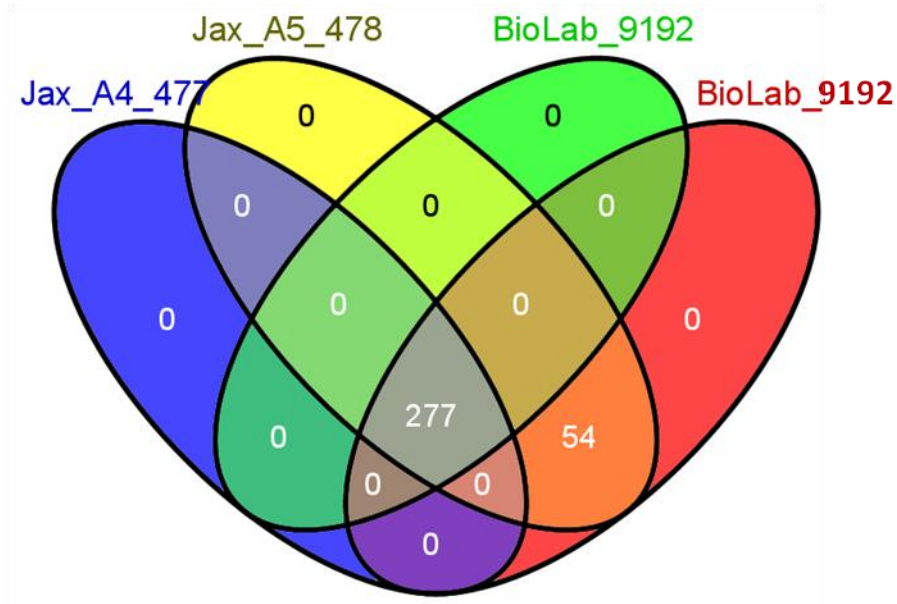
Sample ID	Total called SNPs	Nb of uncalled SNPs	Nb of called SNPs	Nb of Reference Homozygotes (Allele A)	Nb of Heterozygotes	Nb of Alternative Homozygotes (Allele B)
Jackson L	623065	117012	506053	504376 (99,67%)	277 (0,05%)	1400 (0,28%)
Jackson L	623065	116198	506867	505173 (99,67%)	331 (0,07%)	1363 (0,27%)
Janvier Labs	623065	117012	506053	504377 (99,67%)	277 (0,05%)	1399 (0,28%)
Janvier Labs	623065	116198	506867	505164 (99,66%)	331 (0,07%)	1372 (0,27%)

► The large majority of called SNPs are close to reference C57BL/6J (>99,6% homozygous A SNPs)

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Venn diagram for Heterozygous SNPs

Venn Diagram for SNPs with heterozygous genotype

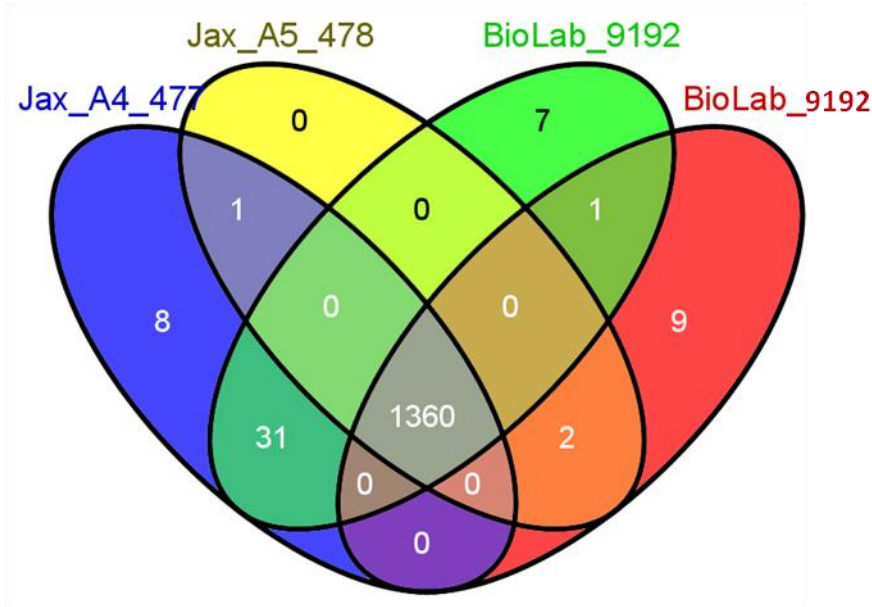


Sample ID	Total called SNPs	Nb of uncalled SNPs	Nb of called SNPs	Nb of Reference Homozygotes (Allele A)	Nb of Heterozygotes	Nb of Alternative Homozygotes (Allele B)
Jax_A4_477	623065	117012	506053	504376 (99,67%)	277 (0,05%)	1400 (0,28%)
Jax_A5_478	623065	116198	506867	505173 (99,67%)	331 (0,07%)	1363 (0,27%)
BioLabs_9192	623065	117012	506053	504377 (99,67%)	277 (0,05%)	1399 (0,28%)
BioLabs_9193	623065	116198	506867	505164 (99,66%)	331 (0,07%)	1372 (0,27%)

- ▶ All four samples share 277 common heterozygous SNPs
- ▶ Jax_A5_478 and BioLab_9193 share 54 common heterozygous SNPs which are specific to female samples

Venn diagram for Homozygous B SNPs

Venn Diagram for SNPs with alternative allele B (Homozygous B) genotype



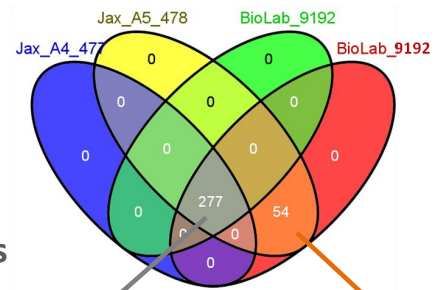
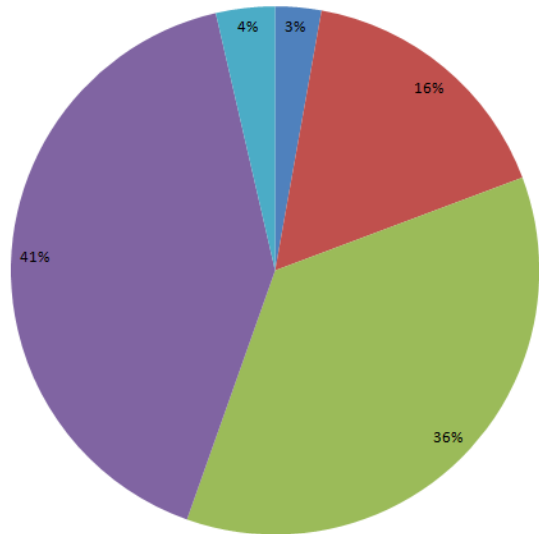
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BioLabs_9193	623065	116198	506867	505164 (99,66%)	331 (0,07%)	1372 (0,27%)

- ▶ All four samples share 1360 common homozygous B SNPs
- ▶ 31 SNPs are specific and common to male samples while 2 SNPs are specific and common to female samples
- ▶ Jackson samples share 1 common homozygous B SNP
- ▶ BioLabs samples share 1 common homozygous B SNP

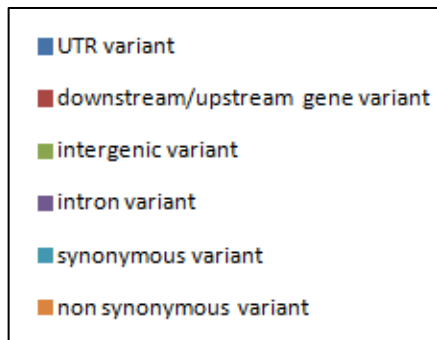
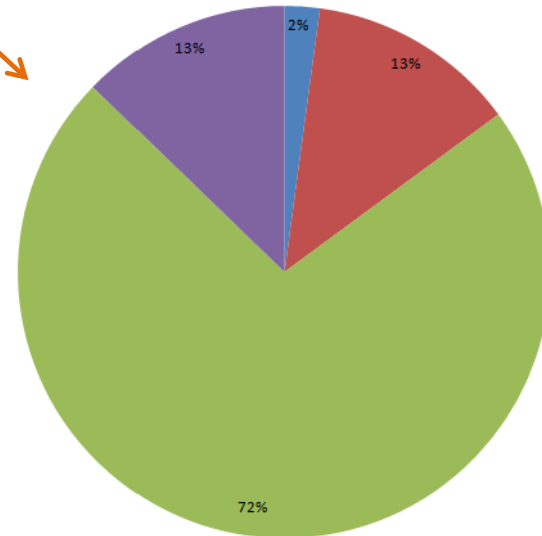
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Functional classes of Heterozygous SNPs

Functional classes of 277 heterozygous SNPs common to all samples



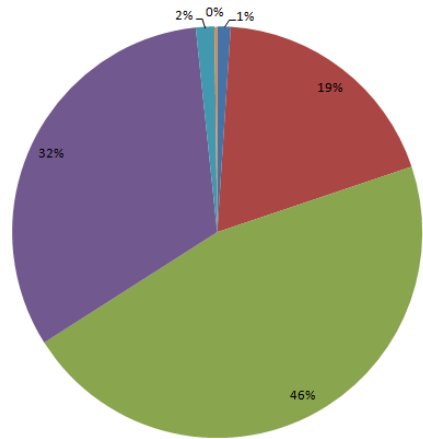
Functional classes of 54 heterozygous SNPs specific to female samples



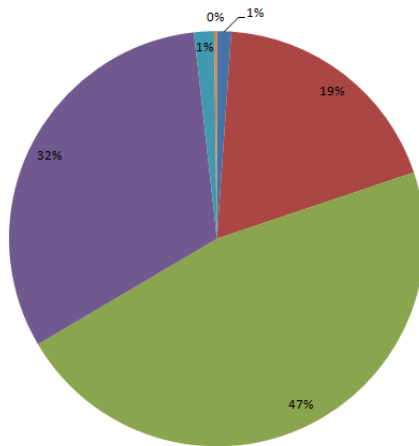
- ▶ The majority of SNPs fall into the intronic or intergenic regions
- ▶ There are no non synonymous heterozygous SNPs

Functional classes of Homozyzygous B SNPs

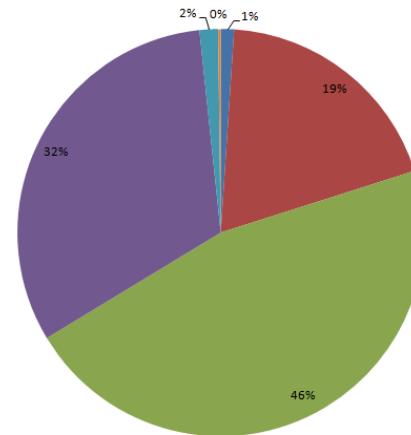
Jax_A4_477



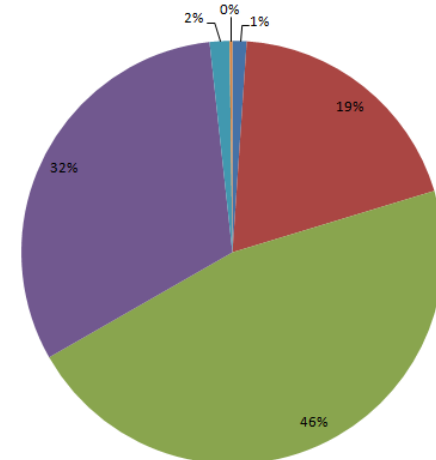
Jax_A5_478



BioLabs_9192

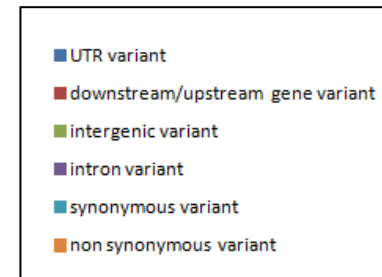


BioLabs_9193



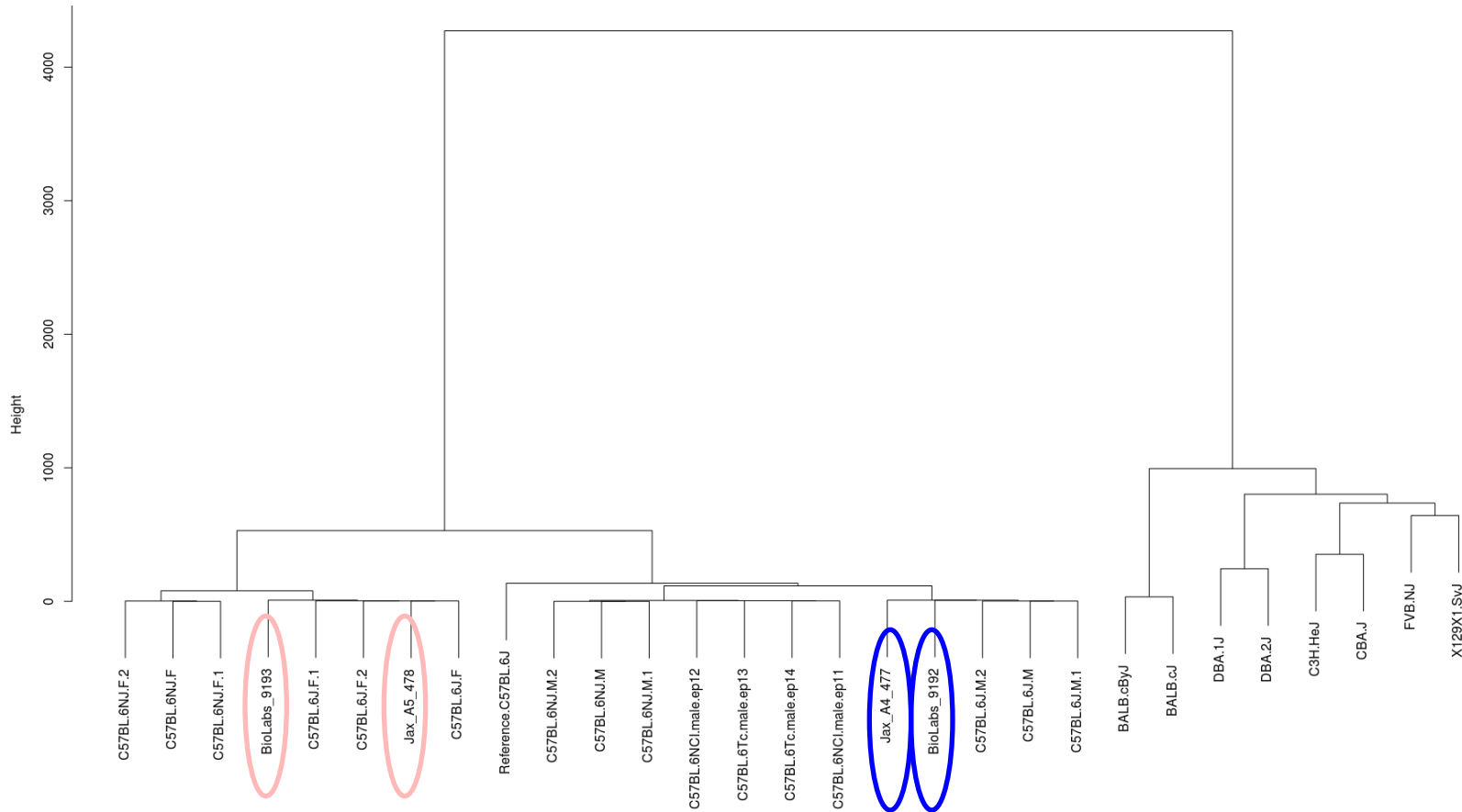
► The majority of SNPs fall into the intronic or intergenic regions

► There is only one missense homozygous B SNP (rs13479126) which is predicted to have no consequence on protein function



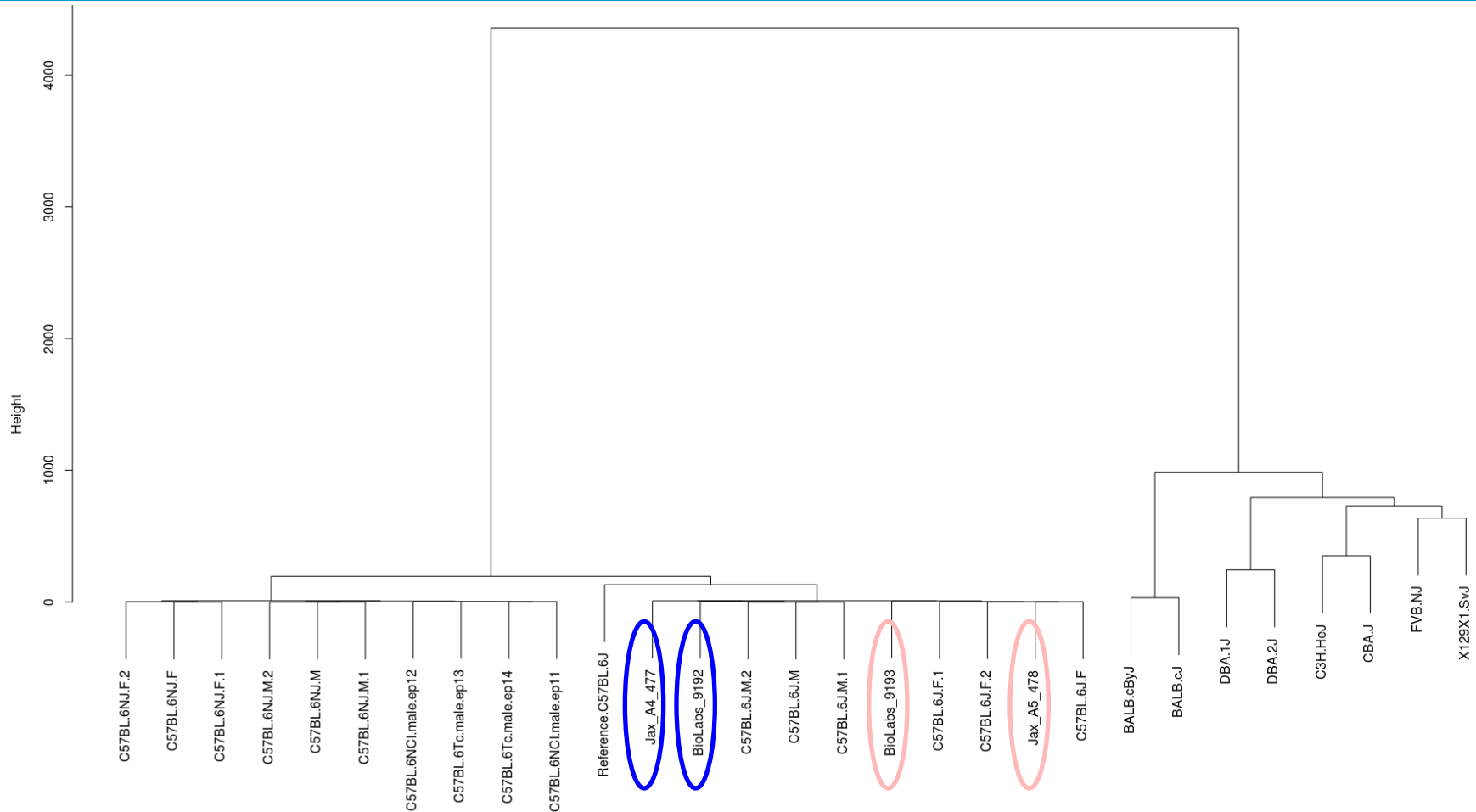
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Clustering of all samples



- ▶ A clustering was performed with all samples, taking chromosomes X and Y into account (uncalled SNPs were excluded from analysis)
- ▶ Reference.C57BL.6J represents a “fictional” reference genome sequence which contains allele A for every tested SNP
- ▶ Male samples Jax_A4_477 and BioLabs_9192 are closer to each other and to other male samples than to the reference C57BL/6J
- ▶ Female samples Jax_A5_478 and BioLabs_9193 are closer to each other and to other female samples than to the reference C57BL/6J

Clustering of all samples without chromosome X and Y



- ▶ A clustering was performed with all samples without taking chromosomes X and Y into account (uncalled SNPs were excluded from analysis)
- ▶ All four samples analysed clustered closer to each other than to the reference C57BL/6J
- ▶ Male samples Jax_A4_477 and BioLabs_9192 are clustered closer to each other than to the female samples
- ▶ Female samples Jax_A5_478 and BioLabs_9193 are clustered closer to each other than to the male samples

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Conclusion

1. The four samples are close to the reference C57BL/6J strain (>99,6% of called SNPs are homozygous for allele A in all samples)
2. The samples are closer to each other than to the reference C57BL/6J
 - Male samples Jackson laboratories and Janvier Labs are closer to each other than to female samples
 - Female samples Jackson Laboratories and Janvier Labs are closer to each other than to male samples
3. Apart from one missense SNP (rs13479126) which causes a change in amino acid but does not affect protein function, the other SNPs do not have any impact on the C57BL/6JRj mouse strains



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