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DIG (DEEP)ER

Deep learning algorithms for the imbalanced classification of correct and incorrect SNP genotypes from WGS pipelines

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THETA
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Institute of Animal Genetics

#1 MATERIALS

Whole-genome DNA sequence of four traditional Danish Red Dairy Cattle bulls:

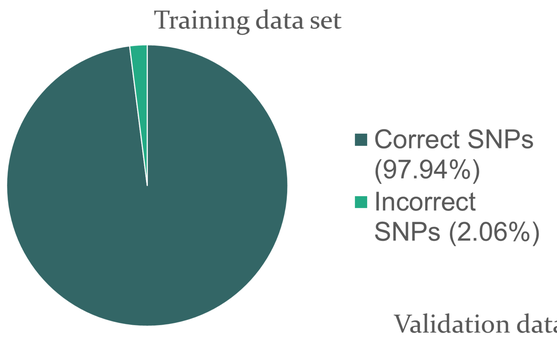
1) The training data set—**three animals**,
2) The validation data set—**the fourth animal**.

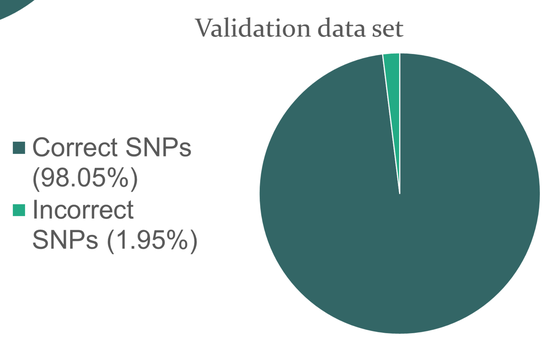
Correct SNPs (concordant WGS—Chip):

1) Training data set: 2 227 995 SNPs,
2) Validation data set: 749 506 SNPs.

Incorrect SNPs (discordant WGS—Chip):

1) Training data set: 46 920 SNPs,
2) Validation data set: 14 940 SNPs.





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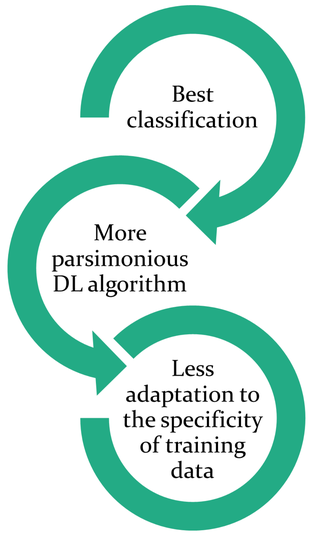
#4 CONCLUSIONS



#2 METHODS

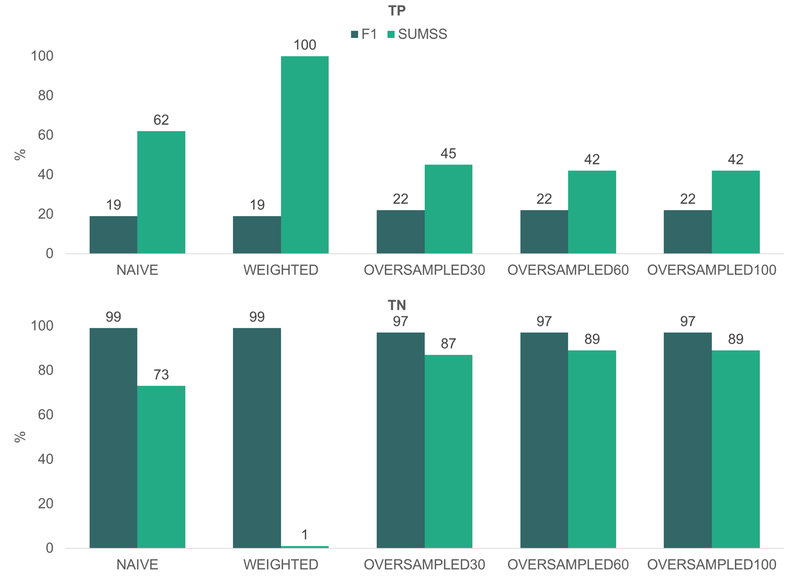
Deep Learning algorithms

1) Naïve algorithm
2) Weighted algorithm
3) Oversampled algorithm:
   - 30%
   - 60%
   - 100%

Cutoff points

1) The estimated cutoff points for each model by:
   - $F1 = \frac{2TP}{2TP+FN+FP}$
   - $SUMSS = \frac{TN}{TN+FP} + \frac{TP}{TP+FN}$

#3 RESULTS



Classification of **validation data** by the algorithms, based on the cutoff thresholds for the **F1** or **SUMSS** metrics.

1) **True positive (TP)**—an incorrect SNP classified as incorrect,
2) **False negative (FN)**—an incorrect SNP classified as correct,
3) **True negative (TN)**—a correct SNP classified as correct,
4) **False positive (FP)**—a correct SNP classified as incorrect,
5) **F1**—values of the F1 metric.