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Enhancing bovine genome SNP call accuracy with autoencoder analysis of nucleotide impact with AI

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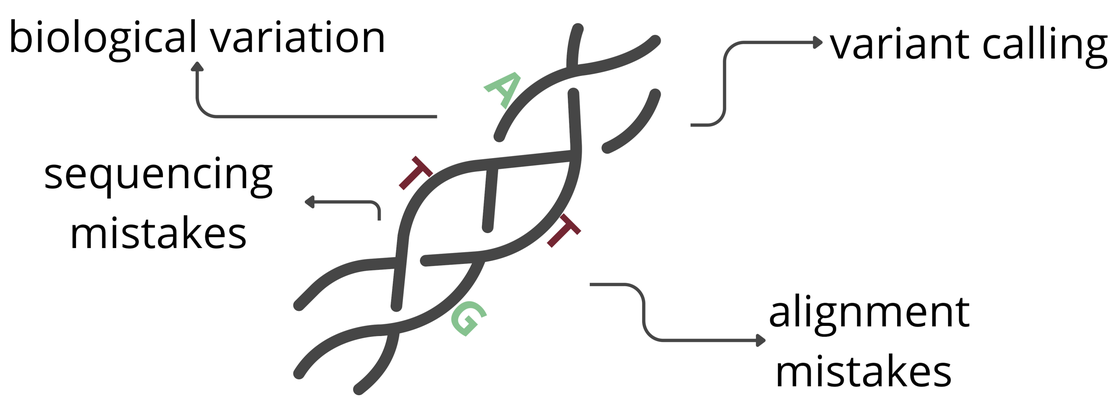
Objective

- Variant calling: critical step in the analysis of NGS data
- Potential mistakes for a number of reasons
- **<1% Incorrect calls: anomaly detection procedure implemented via autoencoder (AE) model**

Conclusions

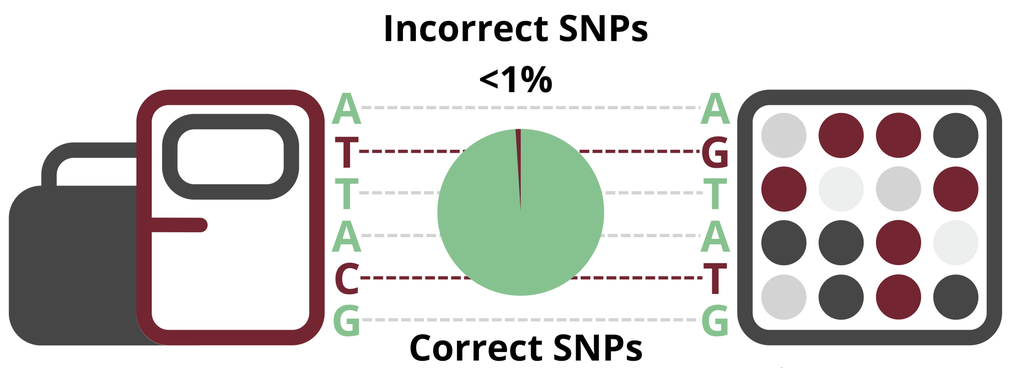
- **Autoencoders show promise in detecting anomalies and efficiently capturing complex patterns**
- Data with limited covariates variability can be represented by principal components
- The number of principal components and neural network architecture profoundly influence anomaly detection efficacy

Causes of mistakes



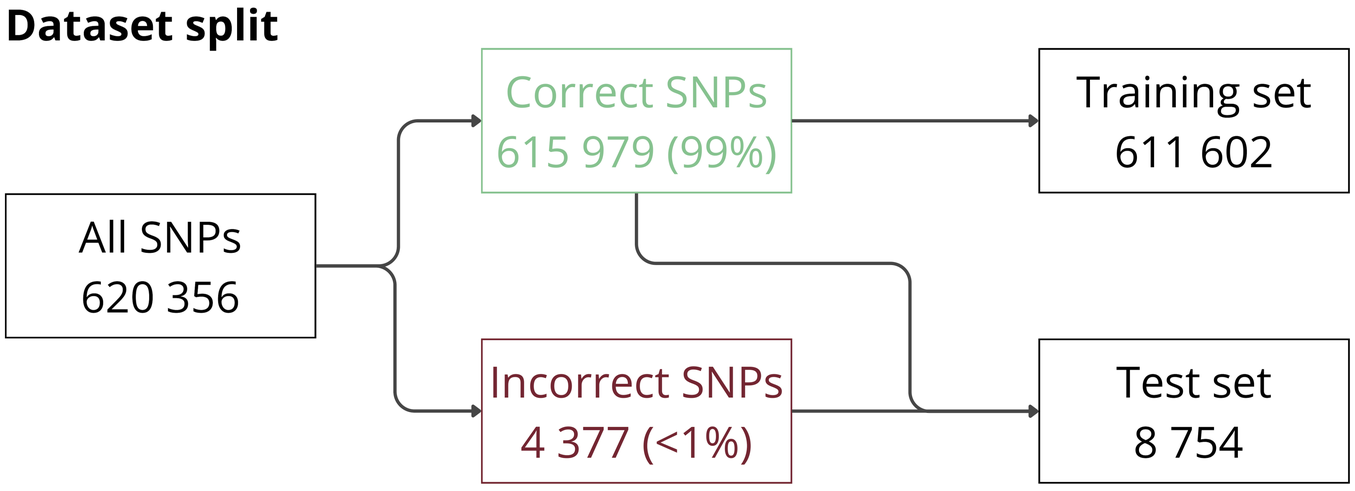
Incorrect calls

- 20 Polish Holstein-Friesian cows



Autoencoders

Dataset split

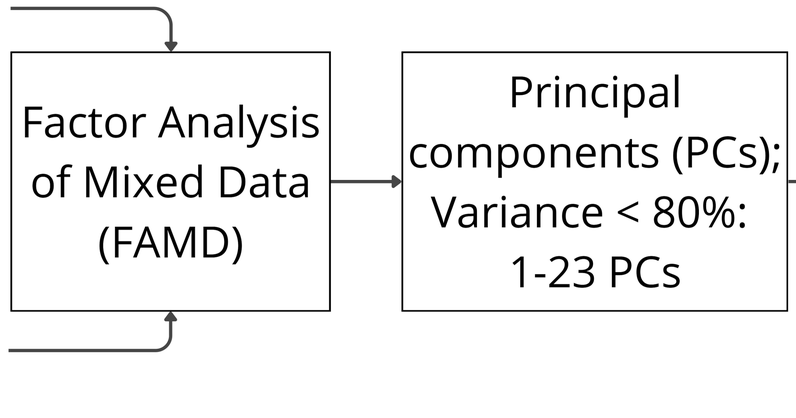


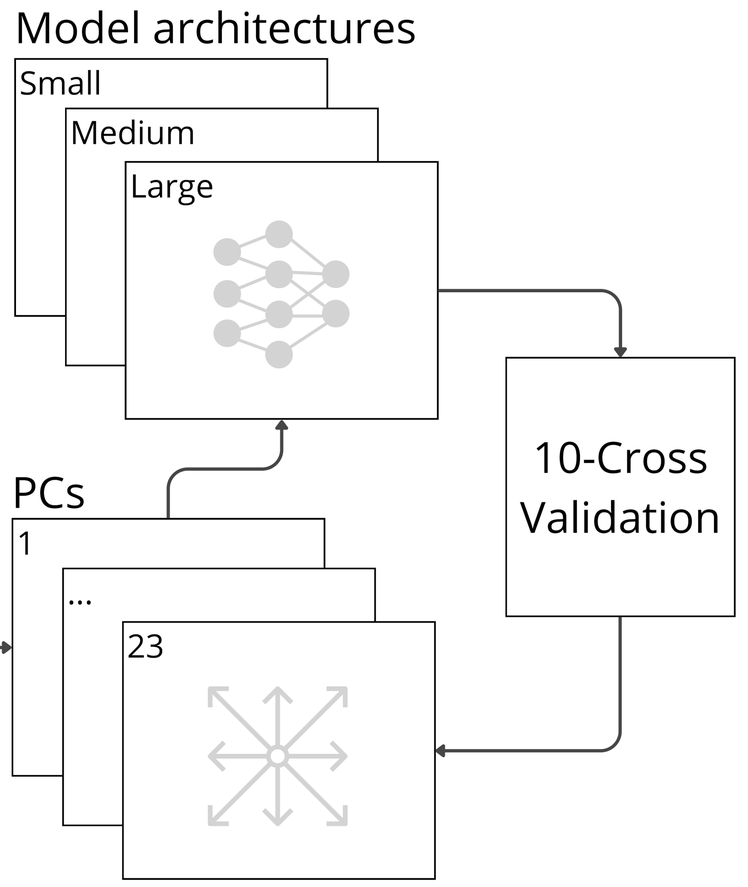
Model implementation

- Downstream: 1-4 bp
- Upstream: 1-4 bp

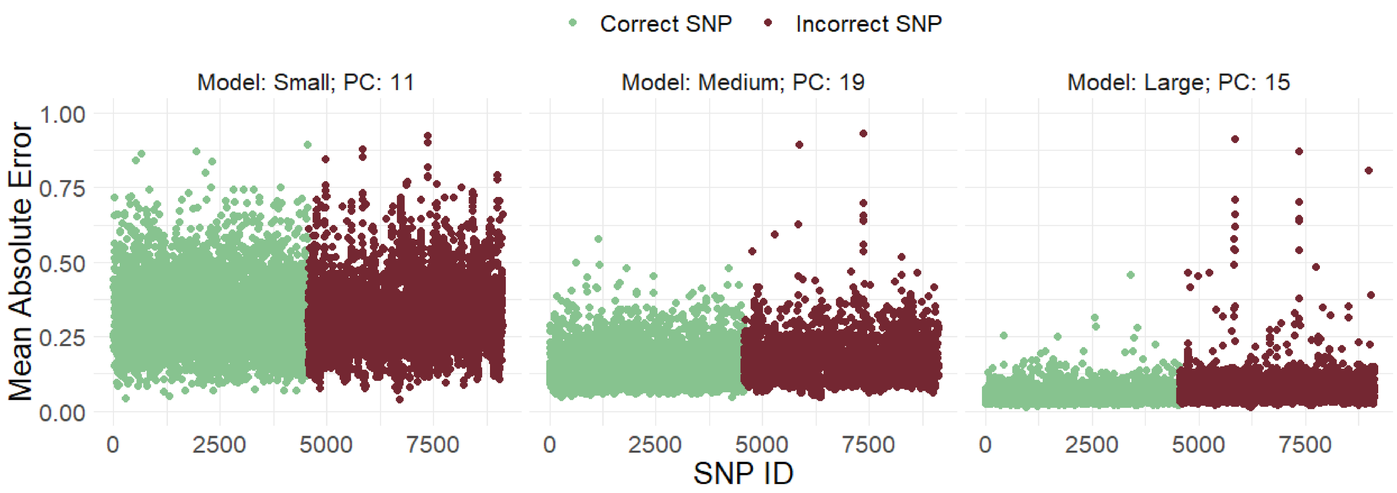
VCF

- Allele: A
- Allele: B
- Read depth: A
- Read depth: B
- Genotype quality





Results



Best model selection:

- Mann Whitney U test
- Sensitivity/Precision threshold

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