

## Università degli Studi di Padova

# Random subsampling techniques for sea bass mortality prediction

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#### Overview



- Motivation: Identifying impactful SNPs in sea bass mortality
- Dataset: Genomic SNP data, mortality outcomes, and annotations
- Method: Subsampling techniques with XGBoost
- Results: Accuracy/F1 vs. subsampling rate
- Conclusion: Subsampling preserves predictive power

## SNPs and Sea Bass Mortality



- VNN is a widespread lethal disease in sea life.
- SNPs may increase or decrease the chance of death.
- Goal: Predict mortality based on SNP profiles.

#### Challenges with Genomic Data



- Each fish: over 6 million SNP positions.
- Sample size: only 990 sea bass individuals.
- Traditional models overfit due to data dimensionality.

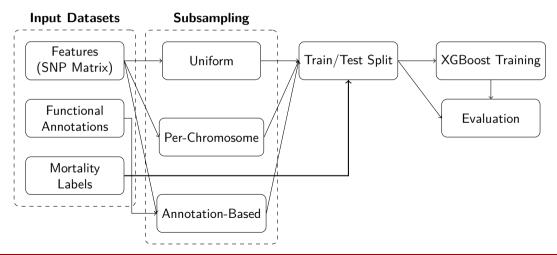
## Machine Learning Approach



- Use XGBoost classifier for mortality prediction.
- Need to reduce feature space: apply subsampling.
- Evaluate performance on subsampled datasets.

#### Pipeline Overview





#### SNP Dataset Structure



- 990 rows (fish), each with 6,072,853 SNP features.
- SNP values: 0 (no mutation), 1 (heterozygous), 2 (homozygous alt).
- Each fish is paired with a mortality label.

#### Annotation Metadata



- Annotations include function: Promoter, Enhancer, Open Chromatin.
- Tissue number (0–25) indicates location-specific relevance.

#### **Uniform Subsampling**



- Randomly sample a fixed proportion p of all SNPs.
- Simple but may cause imbalance across chromosomes.

### Per-Chromosome Subsampling



- Ensures balanced representation from each chromosome.
- Randomly sample same number of SNPs per chromosome.

#### Annotation-Based Subsampling



- Filter SNPs by biological annotation.
- Then apply uniform subsampling to relevant regions.

### Subsampling Strategy Comparison



- Trade-offs in simplicity, biological interpretability, and balance.
- Aim: maximize predictive power while reducing dimensionality.

### XGBoost Training



- Training/testing split is fixed before subsampling.
- Trained multiple times per method/rate to average performance.

#### Control of Randomness



- XGBoost random seed fixed.
- Subsampling is the only random step.

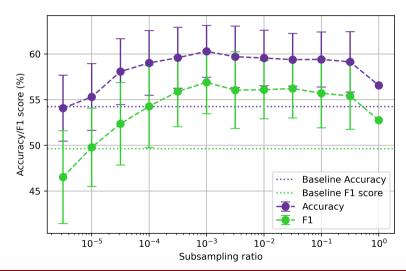
#### Subsampling Ratios



- Subsampled with multiple p values (e.g. 0.01, 0.05, 0.1, 0.2).
- Trained model for each combination.

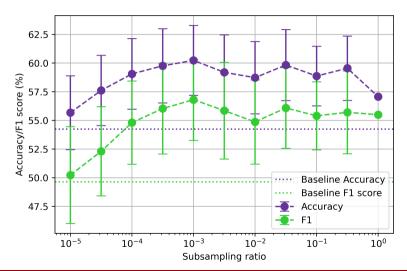
#### Results: uniform subsampling





# Results: uniform subsampling on each chromosome





#### Results: annotated subsampling



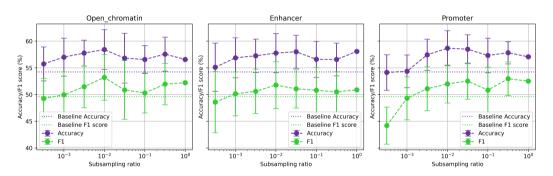


Figure: Plot of accuracy and F1 scores when subsampling uniformly on each chromosome.

#### Results: annotated subsampling



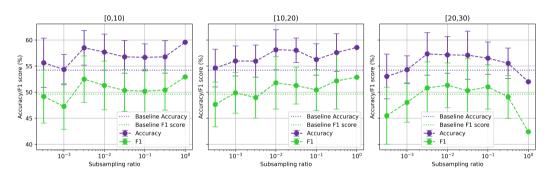


Figure: Plot of accuracy and F1 scores when subsampling uniformly on each chromosome.

#### Conclusions



- Random SNP subsampling retains model effectiveness.
- No strong trend between rate and accuracy (outside extremes).
- Enables faster, scalable experimentation for genomic prediction.

## Thank You / Questions?



## Questions?

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