

# Università degli Studi di Padova

# Random subsampling techniques for sea bass mortality prediction

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#### Overview

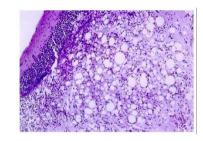


- Motivation: Identifying impactful SNPs in sea bass mortality
- Dataset: Genomic SNP data, mortality outcomes, and annotations
- Method: Subsampling techniques with XGBoost
- Results: Accuracy/F1 vs. subsampling rate
- Conclusion: Subsampling preserves predictive power

#### VNN and Sea Basses



**Viral nervous necrosis** (**VNN**) is a highly spread disease among sealife.

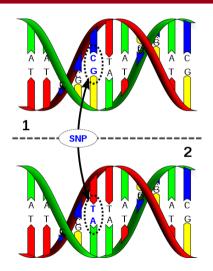




We concentrate our efforts on predicting the mortality of a population of **sea basses** affected by VNN.

## SNPs for predicting mortality





The **genome** might be useful to predict mortality.

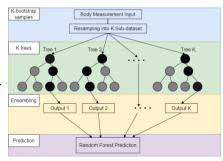
**SNPs**: Single nucleotide polymorphisms.

# Machine Learning Approach



Predict if a sea bass will die by watching its genome: Machine Learning.

In particular, we use the **XGBoost** classifier.



#### Challenges with Genomic Data



- Each fish: over 6 million SNP positions.
- Sample size: only 990 sea bass individuals.
- Traditional models may overfit due to high dimensionality.
- We mitigate through subsampling.



#### Pipeline Overview



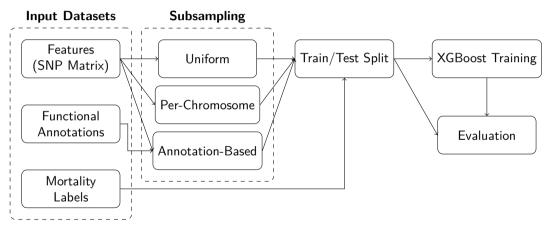


Figure: Pipeline of the model training after subsampling of the data

#### SNP Dataset Structure



•	990 rows	(fish).	each with	6.072.853	SNP	features.
•	990 IOWS	(11511),	each with	0.012.000	2111	reatures.

- SNP values: 0 (no mutation), 1 (heterozygous), 2 (homozygous alt).
- Each fish is paired with a mortality label.

id	mortality
PL06-B12	1
PL06-B06	1
PL06-E06	1
PL08n-B05	0
PL08n-G09	0
:	:

	CAJNNU010000001.1:299	CAJNNU010000001.1:903	CAJNNU010000001.1:986	
PL04-A06	1	0	0	
PL04-A08	0	0	1	
PL04-A09	0	1	1	
PL04-A10	0	0	0	
PL04-A11	2	2	1	
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#### Annotation Metadata



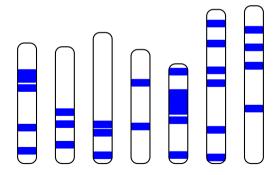
- Annotations include function: Promoter, Enhancer, Open Chromatin.
- Tissue number (0–25) indicates location-specific relevance.

snp_id	funct	n_tissue
CAJNNU010000001.1:7825	Open_chromatin	7
CAJNNU010000001.1:7865	Open_chromatin	7
CAJNNU010000001.1:8046	Open_chromatin	7
CAJNNU010000001.1:8084	Open_chromatin	7
CAJNNU010000001.1:8116	Open_chromatin	7
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## **Uniform Subsampling**



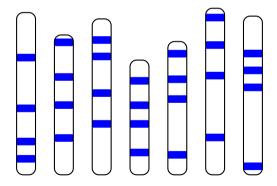
- Randomly sample a fixed proportion p of all SNPs.
- Simple but may cause imbalance across chromosomes.



## Per-Chromosome Subsampling



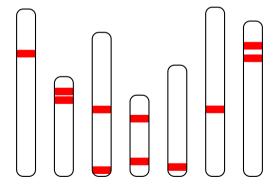
- Ensures balanced representation from each chromosome.
- Randomly sample same number of SNPs per chromosome.



#### Annotation-Based Subsampling



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



## Subsampling Strategy Comparison



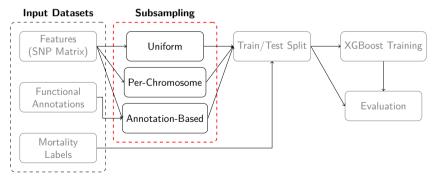
- Trade-offs in simplicity, biological interpretability, and balance.
- Aim: maximize predictive power while reducing dimensionality.

#### Control of Randomness



In order to limit the variance of results we impose:

- XGBoost random seed, train-test split fixed
- Subsampling is the only random component varying between experiments.



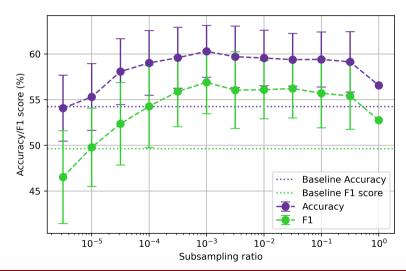
#### Subsampling Ratios and baseline results



- ullet Subsampled with multiple p values: log-spaced varying from the whole genome to few SNPs.
- Trained model for each combination of model and subsample rate.
- Multiple runs for each pair of parameters.
- Comparison with baseline results from a "dumb" classifier, always guessing the most common class.

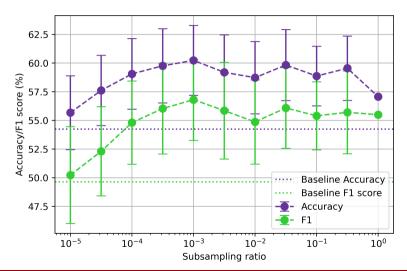
#### Results: uniform subsampling





# Results: uniform subsampling on each chromosome





## Results: annotated subsampling (function)



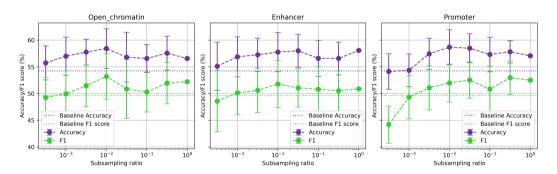


Figure: Plot of accuracy and F1 scores when subsampling uniformly on each chromosome.

# Results: annotated subsampling (tissue number)



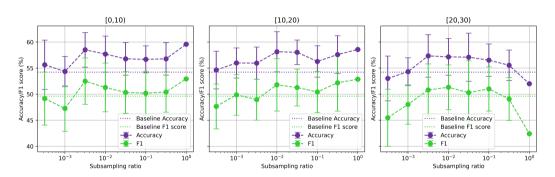


Figure: Plot of accuracy and F1 scores when subsampling uniformly on each chromosome.

#### Conclusions



- Random SNP subsampling retains model effectiveness.
- No strong trend between rate and accuracy (outside extremes): this may be good.
- There doesn't seem to be specific regions of the genome containing the information determining the disease effects.



The results don't show any meaningful trend

We showed that subsampling allows the use of more complex models

# Thank You!