Supervised learning: Classification

Marta Belchior Lopes

NOVAMATH
CENTER FOR MATHEMATICS
+ APPLICATIONS

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Where I am

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DE LISBOA

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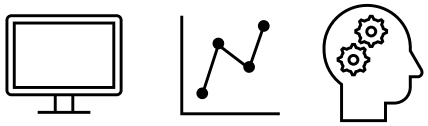
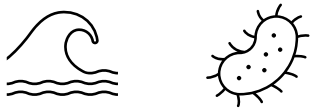
unidemi NOVA | RESEARCH & DEVELOPMENT UNIT FOR MECHANICAL AND INDUSTRIAL ENGINEERING

Biostatistics, Machine Learning and Bioinformatics



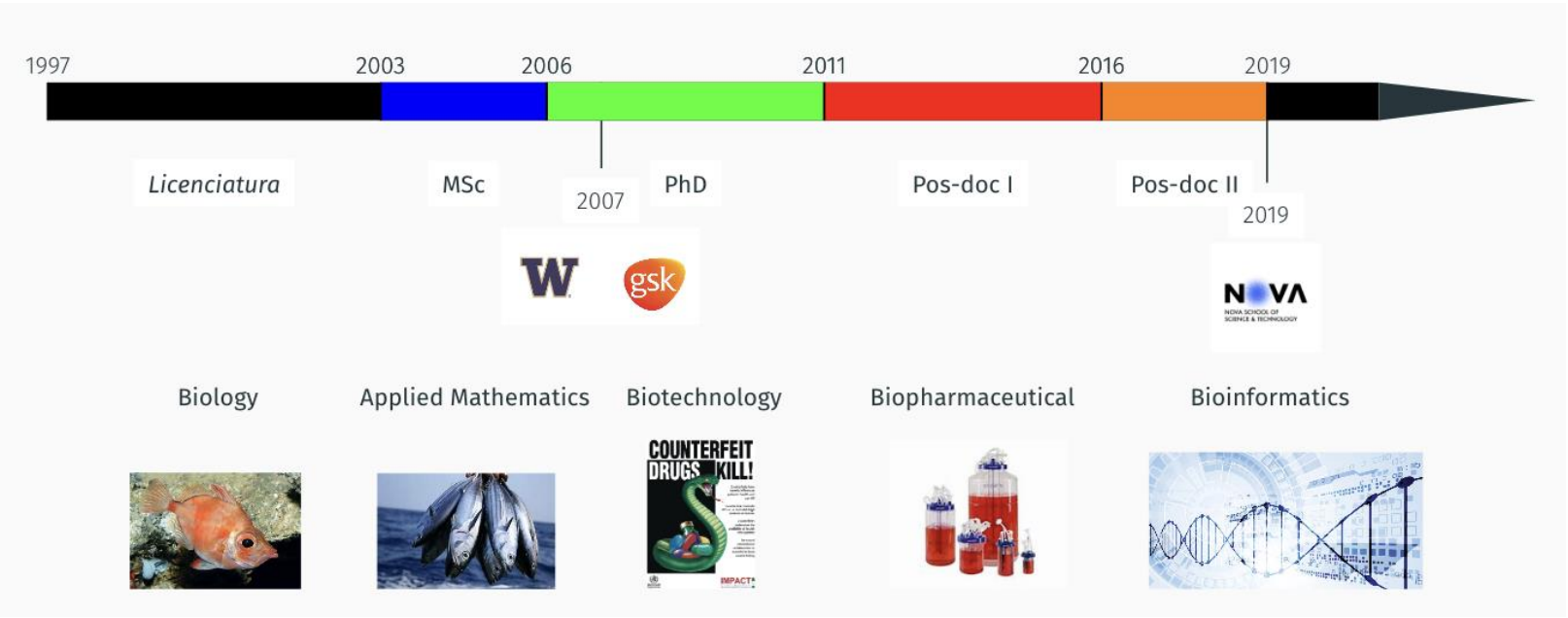
- Precision Medicine

- Environmental



- Ecology

My academic and research timeline



Generalized Linear Models

Linear regression

Multiple linear regression

- Numerical response
- Numerical and categorical predictors

Generalized Linear Models

Linear regression

Multiple linear regression

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Other type of response?
(e.g., categorical, count data)

Generalized Linear Models

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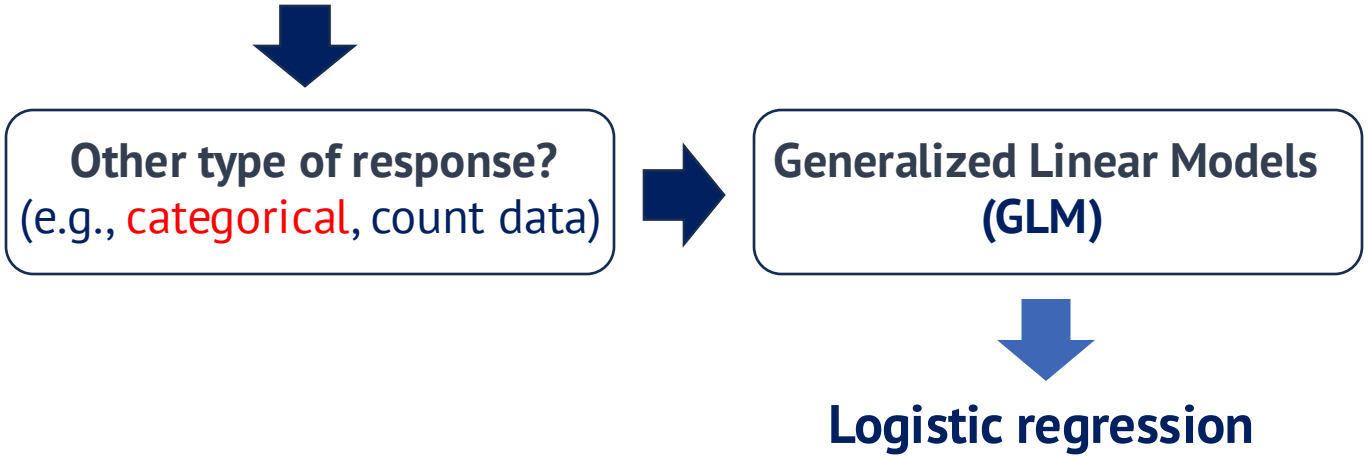
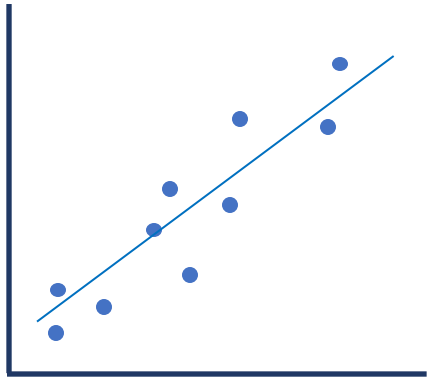


Logistic regression



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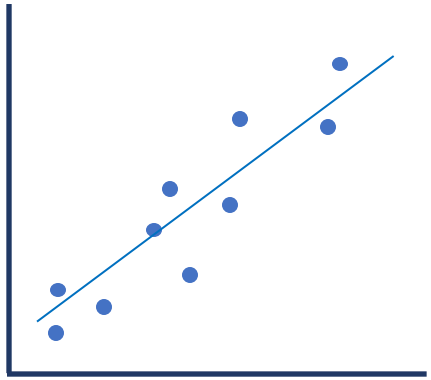

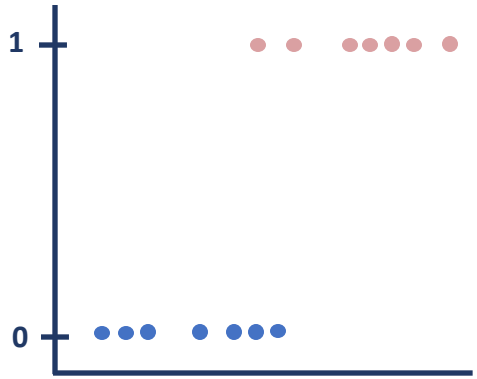

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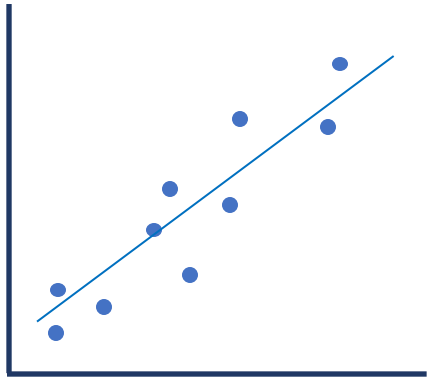

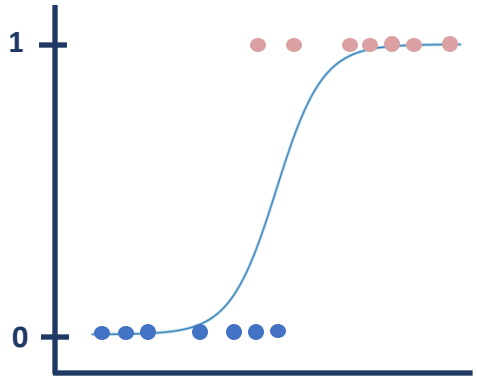

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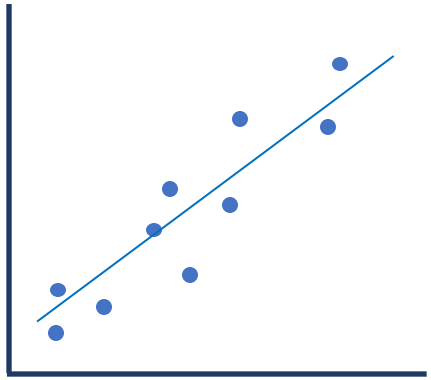

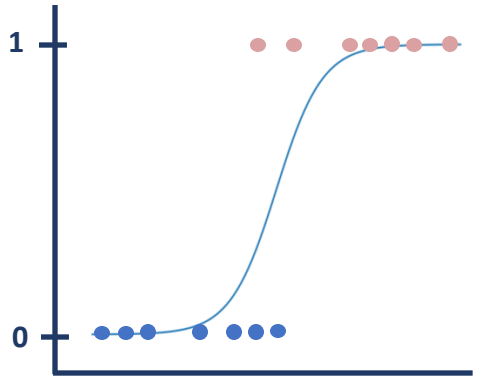

GLM uses a **link** function $g(.)$

Connects the linear predictor to the expected value of the response variable

Logistic regression

The **logit** function

$$\text{logit}(p) = \ln\left(\frac{p}{1-p}\right) = \beta_0 + \beta_1 X_1 + \beta_2 X_2 + \ldots + \beta_p X_p$$

p is the probability of success (event occurrence)

$1 - p$ is the probability of failure (non-event occurrence)

β_0 is the intercept term

$X_1, X_2, \ldots, X_p$ are the predictor variables

Logistic regression

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The **logistic** function

$$p = P(Y = 1) = \frac{1}{1 + e^{-(\beta_0 + \beta_1 X_1 + \beta_2 X_2 + \ldots + \beta_p X_p)}}$$

$P(Y = 1)$ is the probability that the outcome variable Y is equal to 1 (event)

Logistic regression

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$$l(\beta_0, \beta) = -\frac{1}{N} \sum_{i=1}^{N} \left[y_i \log p_i + (1 - y_i) \log(1 - p_i) \right]$$

Logistic regression

BIOLOGY, ECOLOGY & HEALTH

Presence or **absence** of a species in a given *habitat* based on environmental variables (e.g., temperature, humidity, vegetation cover)

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Logistic regression

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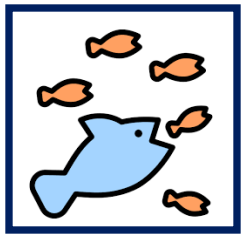

Predator-prey relationships, to predict predatory events based on variables such as predator abundance, environmental conditions, prey density

Logistic regression

BIOLOGY, ECOLOGY & HEALTH

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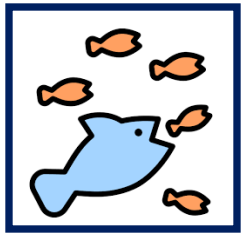

Predator-prey relationships, to predict predatory events based on variables such as predator abundance, environmental conditions, prey density

Contamination events, based on biological and environmental variables (pollution, contaminant, climate)

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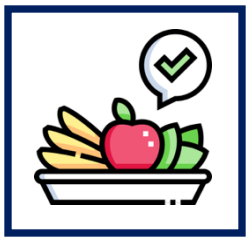
Logistic regression

BIOLOGY, ECOLOGY & HEALTH



Food safety, based variables measured by analytical testing in the laboratory

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Logistic regression

BIOLOGY, ECOLOGY & HEALTH

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Disease presence-absence in a given population based on e.g., clinical, demographic and environmental variables

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BIOLOGY, ECOLOGY & HEALTH

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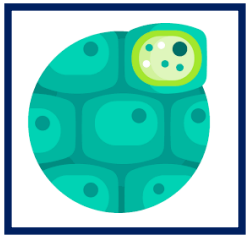

Disease/cell subtype, based on molecular variables (e.g., gene expression)

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Logistic regression

Model training and testing

- Data split into training and test sets

Logistic regression

Model training and testing

- Data split into training and test sets
- Train the logistic regression model

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Logistic regression

Model training and testing

- Data split into training and test sets
- Train the logistic regression model
- Make predictions on the test set
- Evaluate the model

Logistic regression

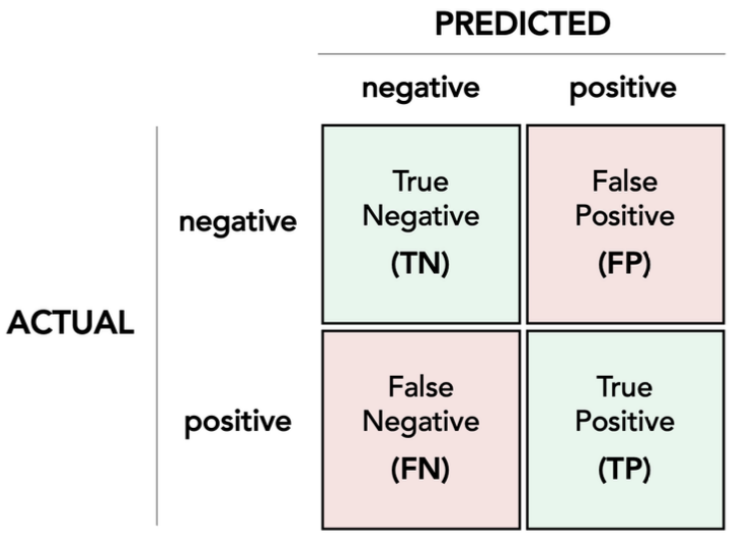
Model evaluation

$$\text{accuracy} = \frac{TP + TN}{TP + TN + FP + FN}$$

$$\text{precision} = \frac{TP}{TP + FP}$$

$$\text{recall} = \frac{TP}{TP + FN} \qquad \text{FPR} = \frac{FP}{FP + TN}$$

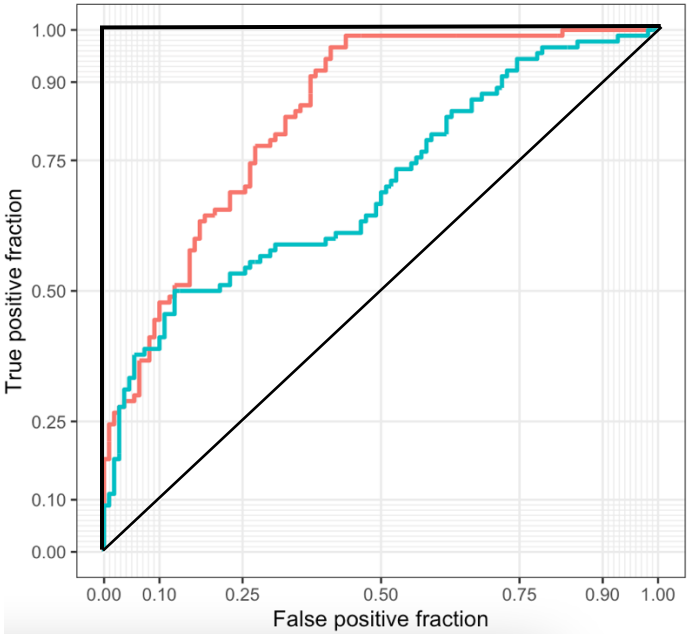
$$F1 = 2 * \frac{\text{precision} * \text{recall}}{\text{precision} + \text{recall}} = \frac{2TP}{2TP + FP + FN}$$



Logistic regression

Model evaluation

- Receiver Operating Characteristic (**ROC**) curve
- Area under the ROC curve (**AUC**)



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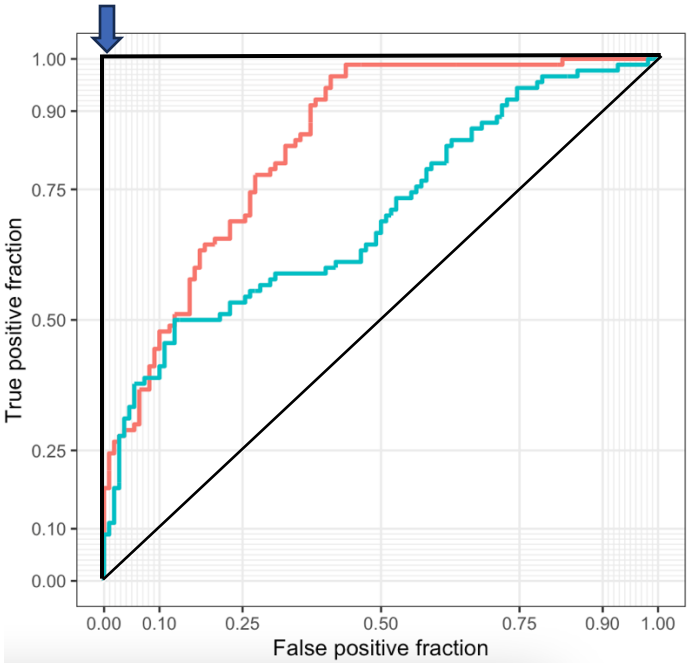
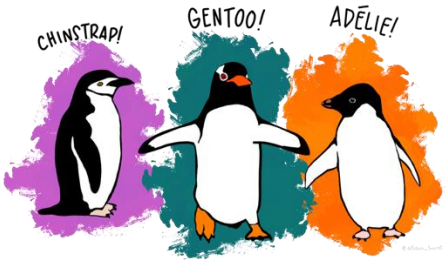


Logistic regression

Penguin species (Palmer Archipelago)

Horst AM, Hill AP, Gorman KB (2020). palmerpenguins: Palmer Archipelago (Antarctica) penguin data. R package version 0.1.0.



- bill_length_mm
- flipper_length_mm
- body_mass_g

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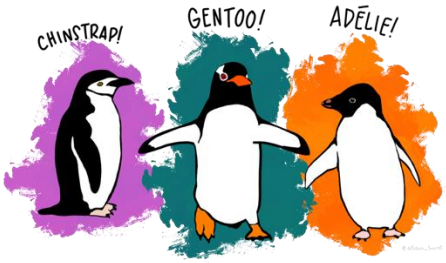

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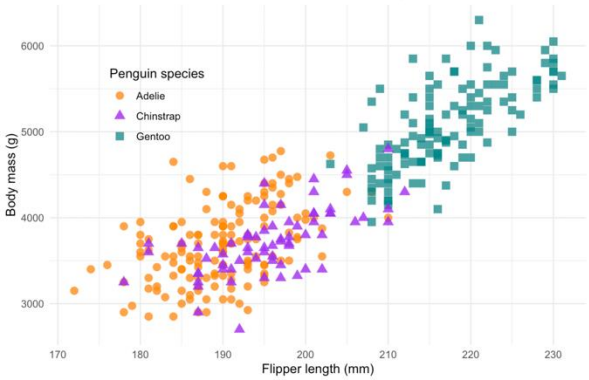

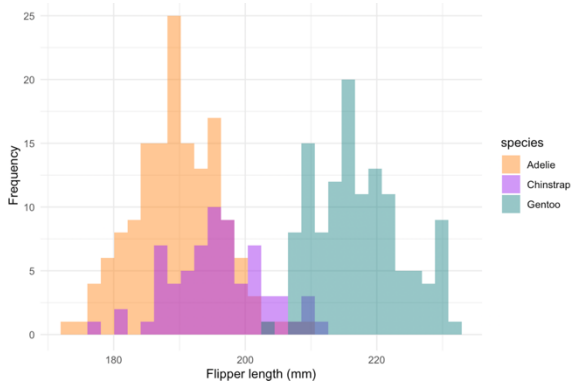

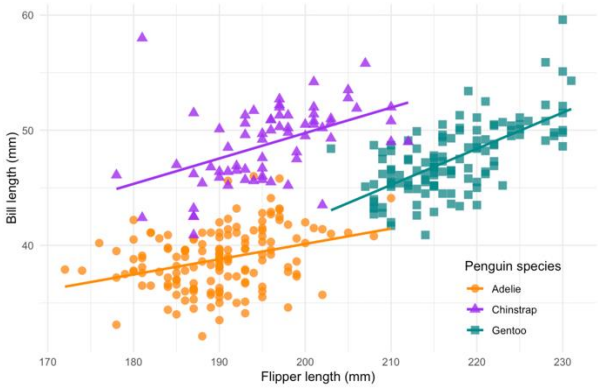

Logistic regression

Penguin species (Palmer Archipelago)

- Create a binary outcome variable “IsGentoo”
- Partition the data into a training and a test set
- Build a binary logistic regression with multiple predictor variables
- Make predictions on the test set
- Evaluate the model using the confusion matrix and AUC

Logistic regression

Penguin species (Palmer Archipelago)

```r
# Install packages and load the penguins dataset
install.packages("caret")
install.packages("pROC")
install.packages("palmerpenguins")

library("palmerpenguins")
data("penguins")
dim(penguins)
head(penguins,10)
penguins <- na.omit(penguins)  # remove rows with missing values

# Create a binary outcome variable, TRUE if species is Gentoo
penguins$IsGentoo <- ifelse(penguins$species == "Gentoo", 1, 0)

# Partition the data into a training set and a test set, preserving class distribution
set.seed(123)  # For reproducibility
library("caret")
partition <- createDataPartition(penguins$IsGentoo, p = 0.7, list = FALSE)
train_data <- penguins[partition, ]
test_data <- penguins[-partition, ]

# Create a binary logistic regression model with multiple predictor variables
model <- glm(IsGentoo ~ bill_length_mm + flipper_length_mm + body_mass_g,
             data = train_data, family = binomial)

# Make predictions on the test set
test_predictions <- predict(model, newdata = test_data, type = "response")
```

Logistic regression

Penguin species (Palmer Archipelago)

```r
## Evaluate the model

# Convert probabilities to class labels (threshold = 0.5)
test_labels <- ifelse(test_predictions > 0.5, TRUE, FALSE)

confusion_matrix <- table(Predicted = test_labels,
                          Actual = test_data$IsGentoo)
print(confusion_matrix)

# Compute the AUC (Area Under the ROC Curve)
library(pROC)
roc_obj <- roc(test_data$IsGentoo, test_predictions)  # probs from glm
auc_value <- auc(roc_obj)
auc_value

plot(roc_obj, main = "ROC Curve")
```

Logistic regression

Penguin species (Palmer Archipelago)

```
> summary(model)


  Call:
  glm(formula = IsGentoo ~ bill_length_mm + flipper_length_mm +
      body_mass_g, family = binomial, data = train_data)

  Coefficients:
                     Estimate Std. Error z value Pr(>|z|)
  (Intercept)       -1.230e+02  3.201e+01  -3.844 0.000121 ***
  bill_length_mm    -3.340e-01  1.693e-01  -1.973 0.048492 *
  flipper_length_mm  5.631e-01  1.610e-01   3.498 0.000469 ***
  body_mass_g        4.954e-03  2.086e-03   2.376 0.017518 *
  ---
  Signif. codes:  0 ‘***’ 0.001 ‘**’ 0.01 ‘*’ 0.05 ‘.’ 0.1 ‘ ’ 1

  (Dispersion parameter for binomial family taken to be 1)

      Null deviance: 308.835  on 233  degrees of freedom
  Residual deviance:  25.393  on 230  degrees of freedom
  AIC: 33.393
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Logistic regression

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bill_length = 45 mm
flipper_length = 220 mm
body_mass = 5500 g

$$\log\left(\frac{p}{1-p}\right) = 13.1 \Rightarrow p = 0.99$$

Logistic regression

Penguin species (Palmer Archipelago)

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$$\log\left(\frac{p}{1-p}\right) = \mathbf{13.1} \Rightarrow p = \mathbf{0.99}$$

bill_length = 45 mm
flipper_length = 195 mm
body_mass = 5500 g

$$\log\left(\frac{p}{1-p}\right) = \mathbf{-0.98} \Rightarrow p = \mathbf{0.27}$$

Summary

- **Logistic regression** for binary outcome variables
- **Examples** of application in Biology, Ecology and Health
- **Mathematical** formulation and interpretation
- **R code** example

Fundamentals of
Biodata Analysis with R

December 10-13, 2025

FNS, University of Tirana, Albania

Dimensionality reduction and feature selection

Marta Belchior Lopes

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Dimensionality reduction

Why reducing data dimension?

- Not all features are important (e.g., less impact on the outcome or noise features)
- High number of features increase model complexity ➡ need to simplify
- Measuring can be expensive and time-consuming

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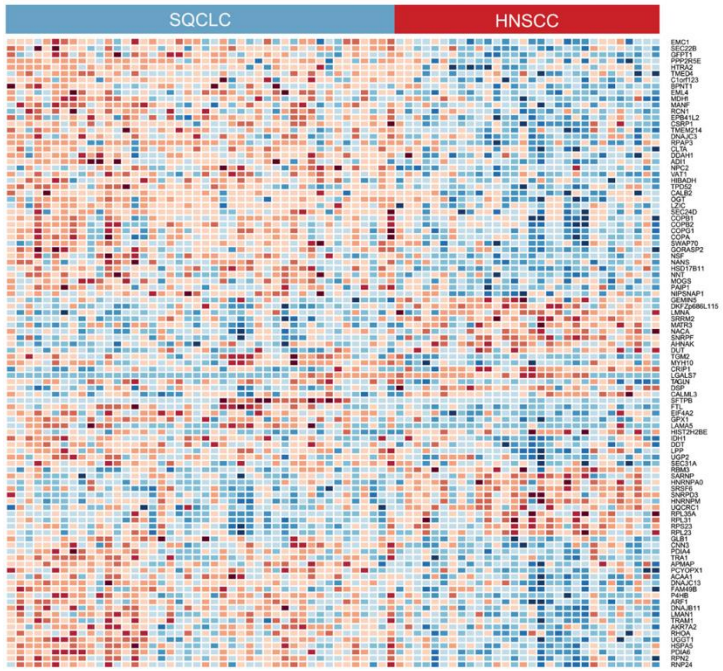
FEATURE SELECTION

➡ **Improve model performance, interpretability** and **efficiency**

Feature selection

Examples

- **Genomics and Proteomics** in disease modeling

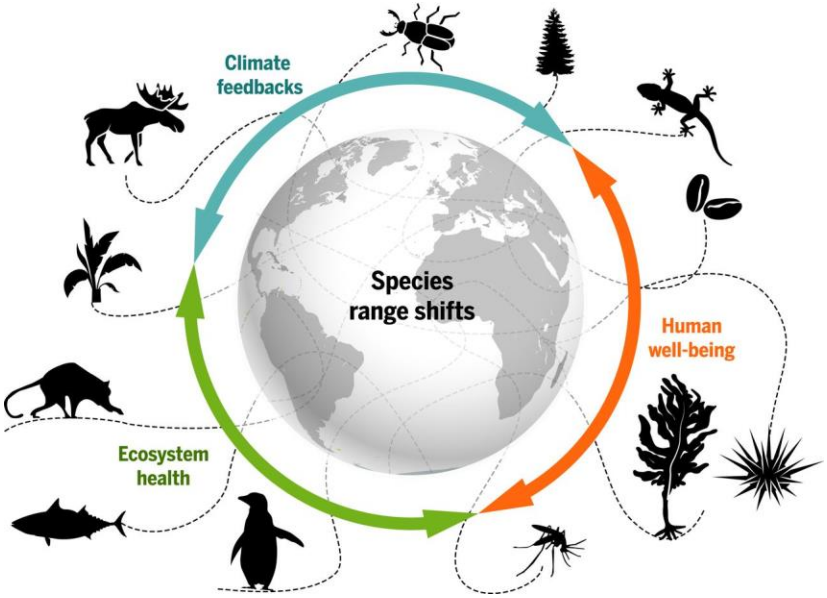


EMBO Mol Med (2018)10:e8428

Feature selection

Examples

- **Genomics and Proteomics** in disease modeling
- **Ecological** studies
  - Species distribution
  - Biodiversity



Science **355**, eaai9214 (2017)

Feature selection

Examples

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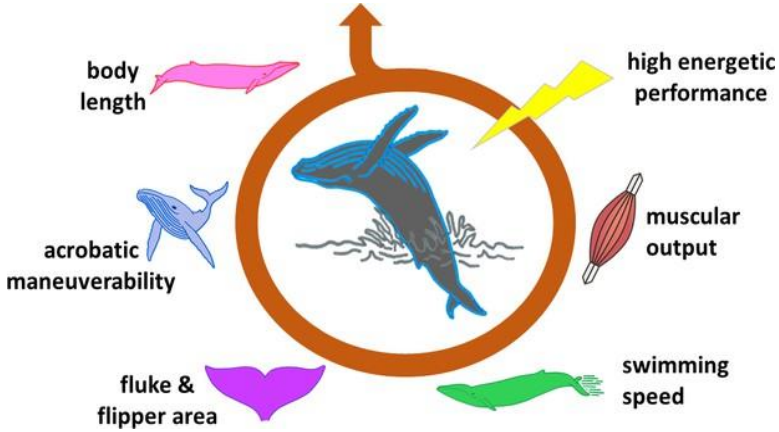
  - Biodiversity
  - Behaviour ecology

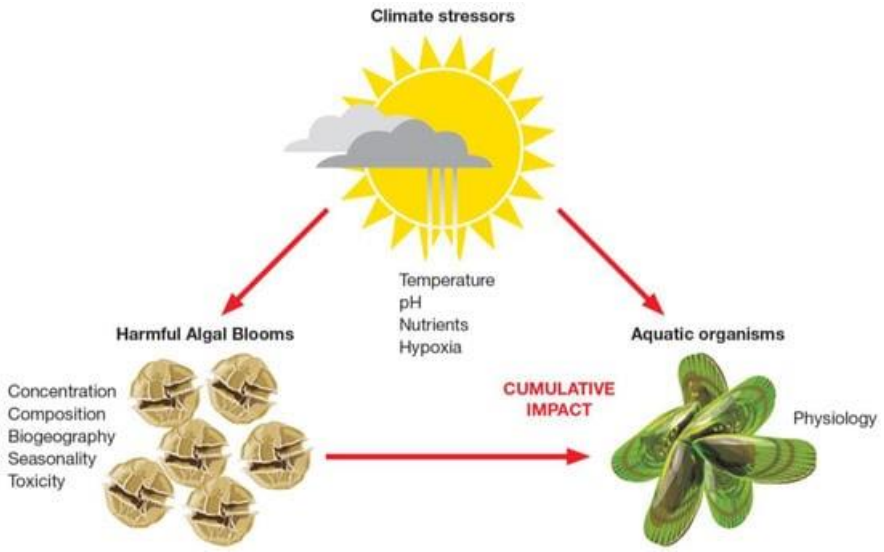


eLife **9**: e55722 (2022)

Feature selection

Examples

- **Genomics and Proteomics** in disease modeling
- **Ecological** studies
- **Toxicology** studies



Toxins **2022**, *14*, 341

Feature selection

Strategies

- **Filter** methods: independent of the learning algorithm used for model building (e.g., statistical properties of the variables)
- **Wrapper** methods: evaluate feature subsets using a learning algorithm and select features based on the performance of the algorithm
- **Embedded** methods: combine feature selection and model training, selecting features as part of the model-building process

Feature selection

Filter methods

- Independence on the learning algorithm, and used as a **preprocessing** step
- Computationally **efficient**
- Features assessed based on **statistical** measures

Feature selection

Filter methods

- Univariate (unsupervised and supervised)
    - Variance
    - Correlation
    - Mutual information
    - Chi-square test
    - Analysis of variance (ANOVA)

Feature selection

Filter methods

- Univariate (unsupervised and supervised)
    - Variance
    - Correlation
    - Mutual information
    - Chi-square test
    - Analysis of variance (ANOVA)
- Multivariate
    - Correlation-based

Feature selection

Wrapper methods

- **Dependence** on the learning algorithm
- **Iterative** process
- **Model performance** metrics as criterion (e.g., accuracy)
- Computationally **expensive** (inadequate for high-dimenisonal data)

Feature selection

Wrapper methods

- **Dependence** on the learning algorithm
- **Iterative** process
- **Model performance** metrics as criterion (e.g., accuracy)
- Computationally **expensive** (inadequate for high-dimenisonal data)
    - Forward selection
    - Backward elimination
    - Recursive feature elimination

Feature selection

Embedded methods

- Combined **feature selection** and **model training**
- More **efficient** than wrapper methods
    - L1 regularization (LASSO)
    - Tree-based methods

Feature selection

Embedded methods

- Combined **feature selection** and **model training**
- More **efficient** than wrapper methods
    - L1 regularization (LASSO)
    - Tree-based methods

Sparse logistic regression

$$l(\beta_0, \boldsymbol{\beta}) = -\frac{1}{N}\sum_{i=1}^{N}\left[y_i \log \pi_i + (1-y_i)\log(1-\pi_i)\right] + \lambda F_\alpha(\boldsymbol{\beta})$$

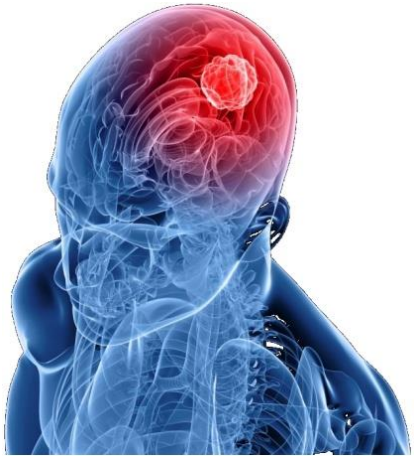
$$F_\alpha(\boldsymbol{\beta}) = \left(\alpha \left\|\boldsymbol{\beta}\right\|_1 + \frac{1-\alpha}{2}\left\|\boldsymbol{\beta}\right\|_2^2\right), \quad \boldsymbol{\beta} = (\beta_1, \ldots, \beta_p)$$

Elastic net & LASSO

Feature selection

Single-cell gene expression data from gliomas

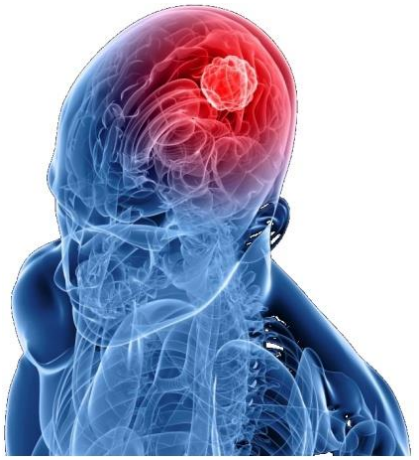
Gliomas – the most common brain tumors



Feature selection

Single-cell gene expression data from gliomas

Gliomas – the most common brain tumors



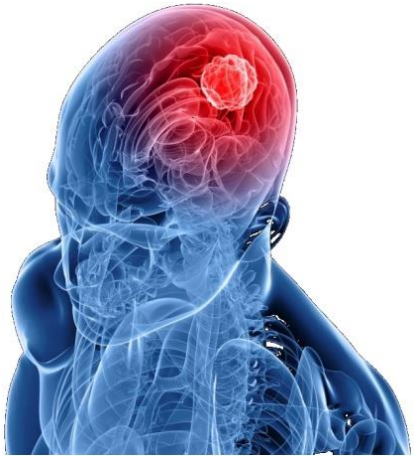
Intertumoral heterogeneity

- Astrocytoma
- Oligodendroglioma
- Glioblastoma

Feature selection

Single-cell gene expression data from gliomas

Gliomas – the most common brain tumors



Intertumoral heterogeneity

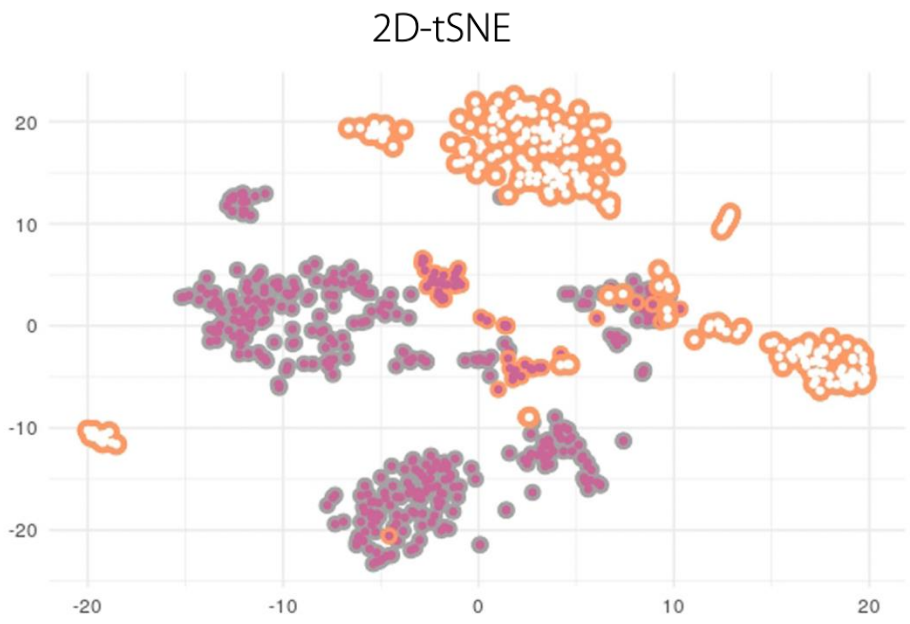
- Astrocytoma
- Oligodendroglioma
- Glioblastoma

Intratumoral heterogeneity

Therapy failure and tumor relapse

Feature selection

Glioblastoma single-cell gene expression data



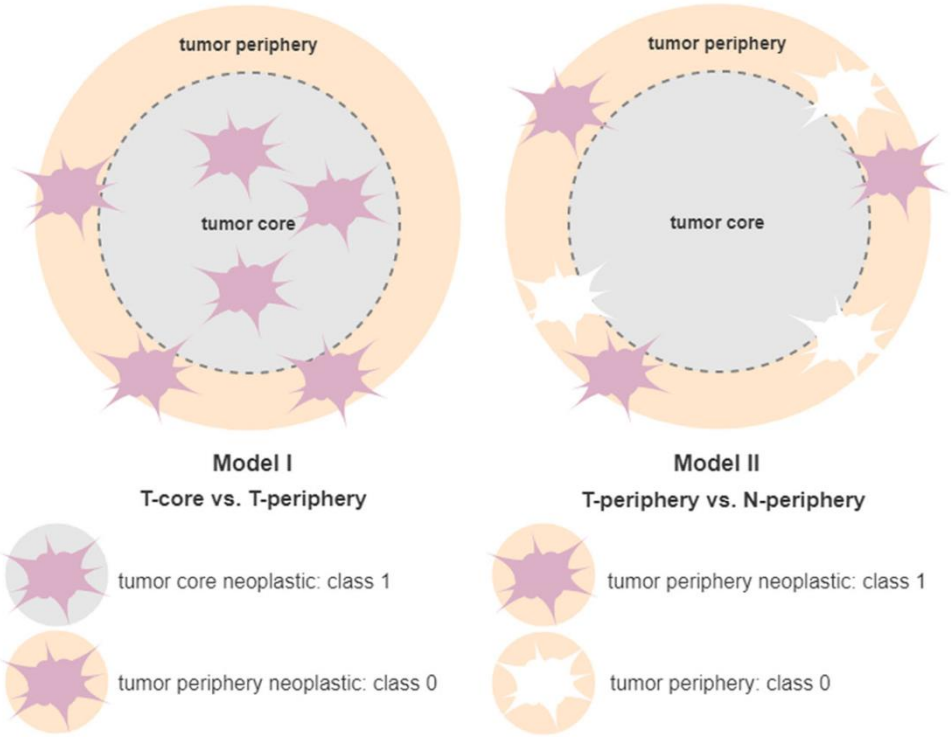
- Single-cell RNA-seq data
- Four primary glioblastoma patients
- **3,589 cells** and **23,368 genes**

tumor core neoplastic
tumor periphery neoplastic
tumor periphery normal

Lopes and Vinga *BMC Bioinformatics* (2020) 21:59
https://doi.org/10.1186/s12859-020-3390-4

Feature selection

Glioblastoma single-cell gene expression data



- **Sparse** logistic regression
- **Elastic net** penalty

Lopes and Vinga *BMC Bioinformatics* (2020) 21:59
https://doi.org/10.1186/s12859-020-3390-4

Feature selection

Glioblastoma single-cell gene expression data

Model performance

Classes	Vars	Miscl		AUC	
		Train	Test	Train	Test
I - T-core vs. T-periphery	83	10	7	0.97	0.94
II - T-periphery vs. N-periphery	85	3	4	0.99	0.96

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Feature selection

Glioblastoma single-cell gene expression data

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Genes selected

Model I - T-core vs. T-periphery				
∗***ATP1A2***	***CLDN10***	***ECHDC2***	***FGFR3***	***GRM3***
HERC6	***HIF3A***	***HSPB8***	***NPL***	***PCSK1N***
PPM1K	∗***PRODH***	***SCG3***	***SPARCL1***	***TMSB10***
Model II - T-periphery vs. N-periphery				
ADAMTS3	***ADAMTSL1***	∗***ANXA1***	***COL28A1***	***CRNDE***
∗***EGFR***	***EMP1***	***F2R***	***GNG5***	***HES6***
HLA-A	***HOXB3***	***HSPB6***	∗***HTRA1***	***ID3***
∗***IFI44L***	***IGFBP2***	***IQCE***	***LINC00475***	***MGLL***
PSPH	∗***PTGDS***	***SEC61G***	***SPOCK1***	***VIM***

Lopes and Vinga *BMC Bioinformatics* (2020) 21:59
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Summary

- **Need** for data dimensionality reduction
- **Methods** for feature selection
- **Embedded** feature selection
- **Example** of application in Biomedicine