# Supervised learning: classification problems

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# Classification problems

# Classification problems

- the response variable **y** is **qualitative**
- e.g.: coat colour, type of rice (Tropical japonica, Indica, Temperate japonica, Aromatic, Aus)
- **y** = **label** (a.k.a. dependent variable)
- **X** = matrix of **features** (continuous, categorical)

# Classification problems

- **y** = **label** (a.k.a. dependent variable)
- **X** = matrix of **features** (continuous, categorical)
- we don't model the response (**y**) directly, rather its **probability**: **P(y=k|X)**
- probabilities lie in [0,1] (not +/- infinity)

# Classification problems

classifier:

- K classes ($k \in K$)

probabilities

classifier

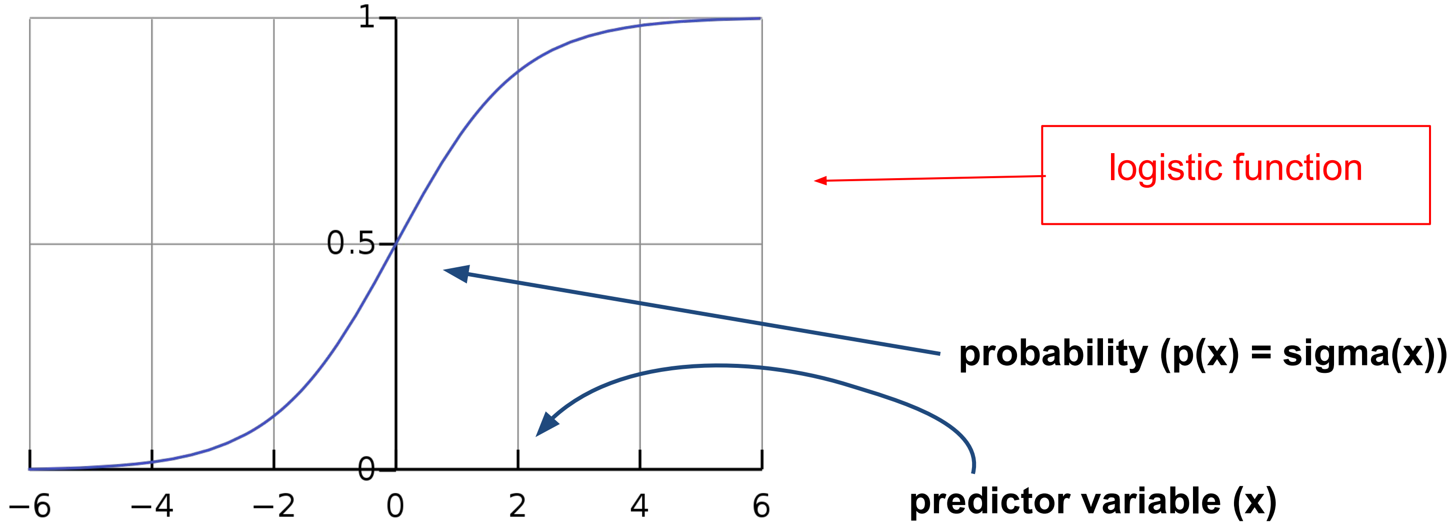
$$p_k(x) = Pr(y = k | X = x) = f(x)$$

$$C(x) = k, \text{if } p_k(x) = max\{p_1(x), p_2(x), \ldots, p_K(x)\}$$

# Binary classification problems

- “special” classification case → only two classes
- binary traits (e.g. cases/controls, resistant/susceptible, high/low, 0/1 etc.)
  - can you think of other examples?
- no need to model the probability of the two classes: one suffices → $P(y=1|x) = f(x)$

# Binary classification problems




$$\sigma(x) = \frac{1}{1+e^{-x}} = \frac{1}{1+\frac{1}{e^x}} = \frac{e^x}{1+e^x}$$

# Logistic regression

- the logistic function is the basis for **logistic regression**
- **P(y=1|x)** [also p(x)]
- P(y=1|z) → $\mathbf{Z = \beta_0 + \beta_1 x}$ (linear combination of variables)

$$p(y=1|x) = \sigma(z) = \frac{e^{\beta_0+\beta_1 x}}{1+e^{\beta_0+\beta_1 x}}$$

we see here the familiar **model coefficients** to be estimated and then used for predictions

# Logistic regression

- a little bit of algebra:

odds

$$p(x) = \frac{e^{\beta_0+\beta_1 x}}{1+e^{\beta_0+\beta_1 x}} \longrightarrow \frac{p(x)}{1-p(x)} = e^{\beta_0+\beta_1 x}$$

# Logistic regression

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log(odds): logit

$$log\left(\frac{p(x)}{1-p(x)}\right) = logit(p(x)) = \beta_0 + \beta_1 x$$

# Logistic regression

- the **logit function** (log(odds)) is the **link function** between a linear expression of X and the probabilities of Y
- linear X expression ($\beta_0+\beta_1$x) → logit scale (continuous)
- logistic function: converts values on the logit scale back to probabilities

$$\begin{cases} logit(p(x)) = \beta_0 + \beta_1 x \\ \sigma(\beta_0 + \beta_1 x) = p(x) \end{cases}$$

our objective!

# Logistic regression - recap

1. the **logistic function** allows us to **model probabilities** in [0,1] as **functions of variables** (features)
2. we need to **transform** the **non-linear logistic expression** to a manageable **linear expression** →the **logit link** function
3. finally, we use again the **logistic function** to **convert** unbounded results on the **logit scale** to **probabilities** (of belonging to a class given the variables/features)

# Estimating the coefficients

## how do we obtain the model coefficients β?

- similarly to linear regression, we need to define a **cost function** and then minimise it

| observations | predictions |
| --- | --- |
| **y** | $\hat{y} = \sigma(\beta_0 + \beta_1 x)$ |

difference between observed and predicted values → **LEAST SQUARES?**

# Estimating the coefficients

**how do we obtain the model coefficients β?**

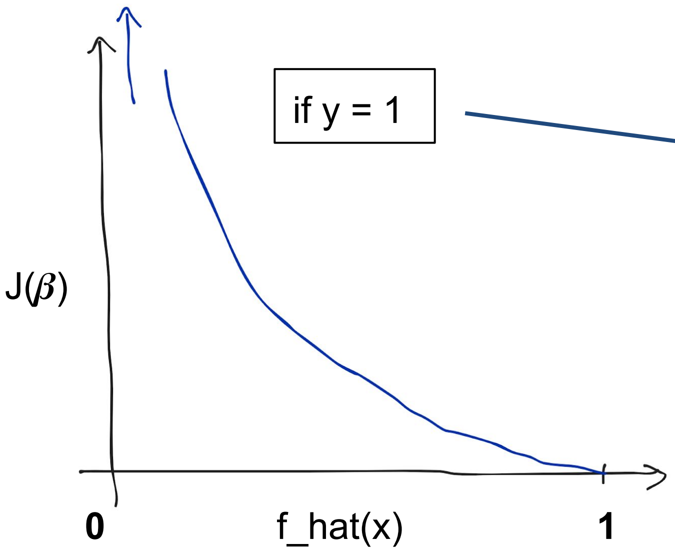
- similarly to linear regression, we need to define a **cost function** and then minimise it

$$J(\beta) = \mathrm{Cost}(\hat{y}, y) = \begin{cases} -log(\hat{y}) & \mathrm{if} y = 1 \\ -log(1-\hat{y}) & \mathrm{if} y = 0 \end{cases}$$

$$J(\beta) = \mathrm{Cost}(\hat{y}, y) = -\left(y \cdot log(\hat{y}) + (1-y) \cdot log(1-\hat{y})\right)$$

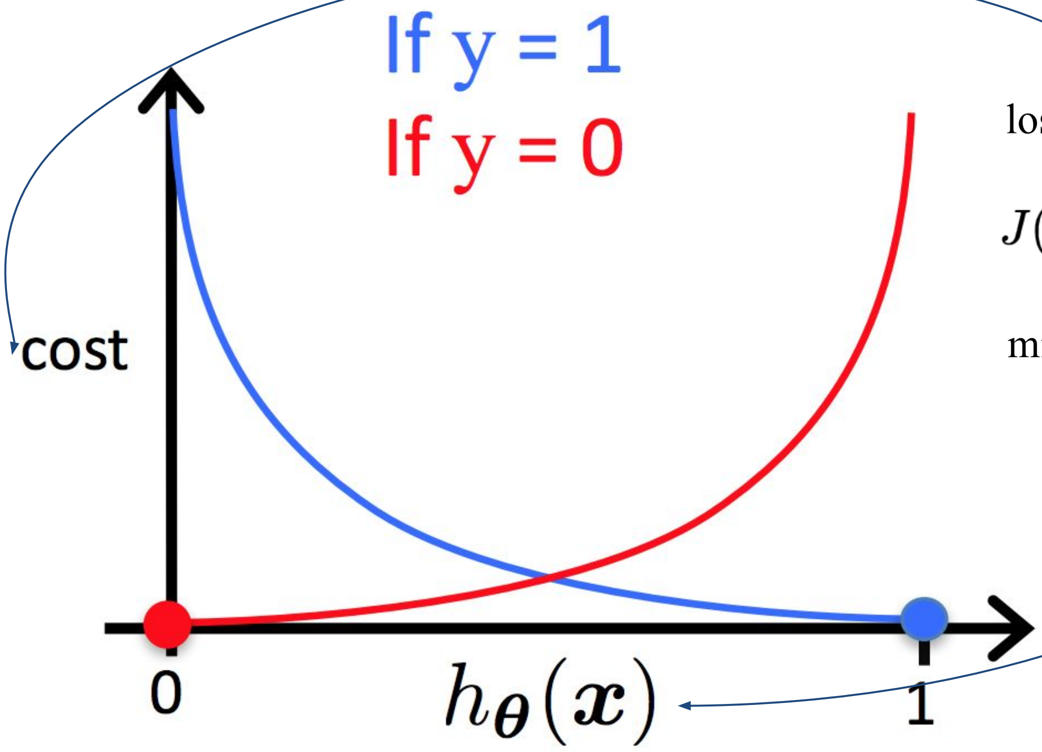
# Cost function for logistic regression

$$J(\beta) = \mathrm{Cost}(\hat{y}, y) = -(y \cdot log(\hat{y}) \cancel{+ (1-y) \cdot log(1-\hat{y})})$$




- if y_hat = 1, cost = 0
- if y_hat → 0 (but y = 1), cost → infinity
- the opposite holds if y = 0

# Loss function for logistic regression




$$\text{loss}\,(\hat{y}, y) = J(\beta)$$

$$J(\beta) = -\,(y \cdot \log\,(\hat{y}) + (1 - y) \cdot \log\,(1 - y))$$

$$\underset{\beta}{\text{minimize}}\, J(\beta)$$

From: https://datascience.stackexchange.com/questions/40982/logistic-regression-cost-function

# Minimising the cost function

- the defined cost function is convex
- can be minimised by **gradient descent**
- machine learning perspective: gradient descent is a general algorithm to solve models
- alternatively:
    - maximum likelihood
    - non-linear least squares

# Binary classification: measuring performance

- the most common metric to measure the performance of a binary classifier is the **error rate**:

$$\frac{1}{n}\sum_{i=1}^{n} I(y \neq \hat{y})$$

# Confusion matrix

| | | True observation | |
|---|---|---|---|
| | | 1 | 0 |
| Prediction | 1 | TP | FP |
| | 0 | FN | TN |

- **FPR** = FP/(FP+TN)
- **FNR** = FN/(FN+TP)
- TER = (FN+FP)/(FN+FP+TN+TP)

Not only total error rate!

# Introducing the dataset

# Genetic variants for cleft lip in dogs

binary phenotypes: **cleft lip** (presence/absence)

PLOS | GENETICS

RESEARCH ARTICLE

## Genome-Wide Association Studies in Dogs and Humans Identify *ADAMTS20* as a Risk Variant for Cleft Lip and Palate

Zena T. Wolf[1☯], Harrison A. Brand[2,3☯¤a], John R. Shaffer[3☯], Elizabeth J. Leslie[2], Boaz Arzi[4], Cali E. Willet[5], Timothy C. Cox[6,7,8], Toby McHenry[2], Nicole Narayan[9], Eleanor Feingold[3], Xioajing Wang[2¤b], Saundra Sliskovic[1], Nili Karmi[1], Noa Safra[1], Carla Sanchez[2], Frederic W. B. Deleyiannis[10], Jeffrey C. Murray[11], Claire M. Wade[5], Mary L. Marazita[2,12‡*], Danika L. Bannasch[1‡*]

# Genetic variants for cleft lip in dogs

binary phenotype: **cleft lip** (presence/absence)

- Nova Scotia Duck Tolling Retriever (NSDTR)
- 125 dogs:
    - 13 cases
    - 112 controls

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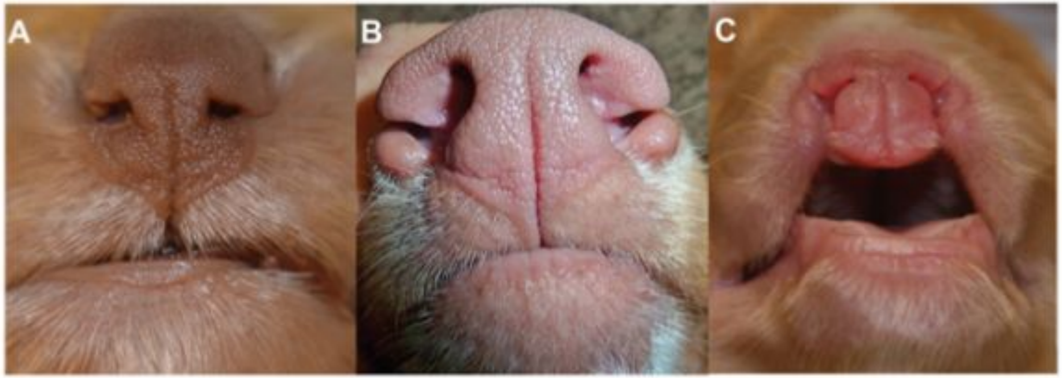

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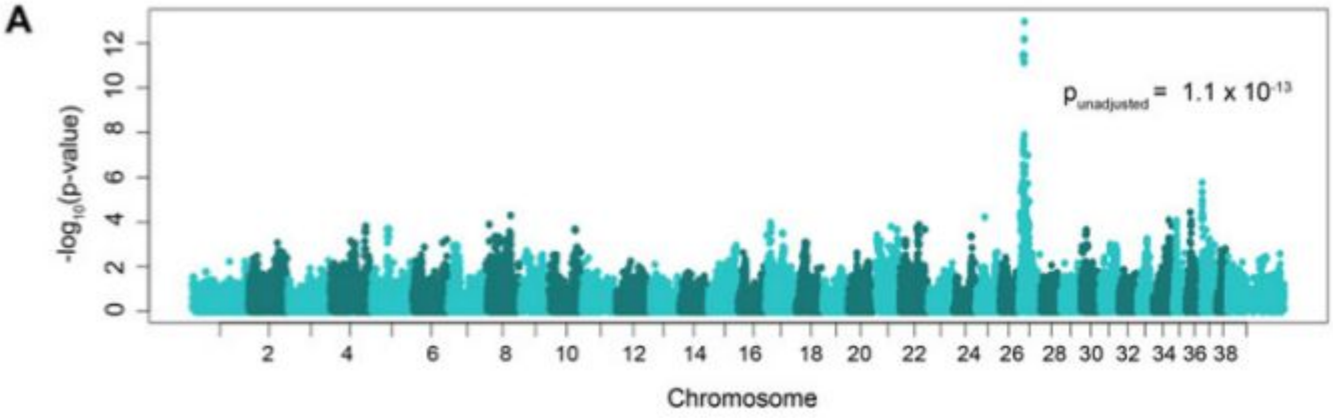



39 chromosomes

Strong signal of association on chromosome 27

# Logistic regression

- demonstration 4.1
- Exercise 4.1

→ 4.classification.ipynb

# ROC curves

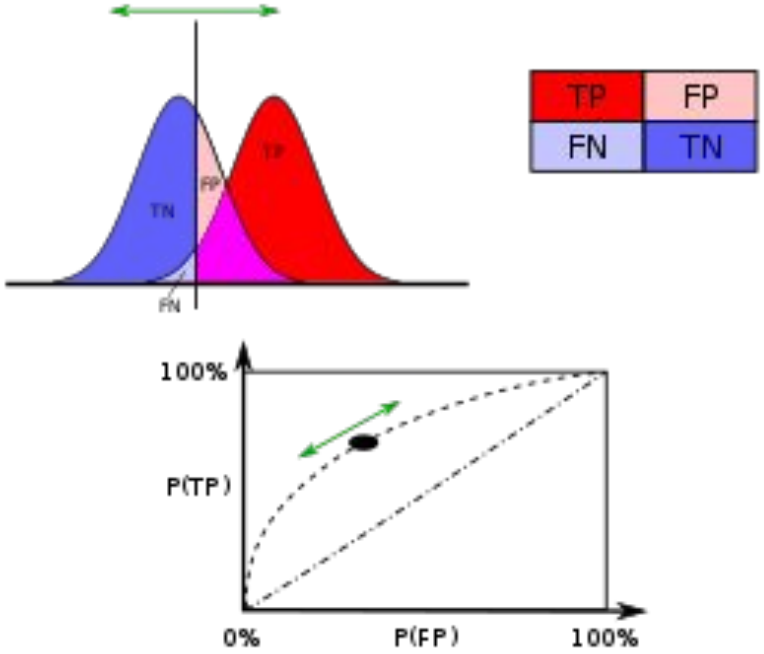
# Binary classification

| | | True observation | |
|---|---|---|---|
| | | 1 | 0 |
| Prediction | 1 | TP | FP |
| | 0 | FN | TN |

- classify observations in **two categories** (1/0)
- however, predictions are often **probabilities** ($P(y=1|x)$)
- different ***cut-offs*** (e.g. 0.5) will give different results

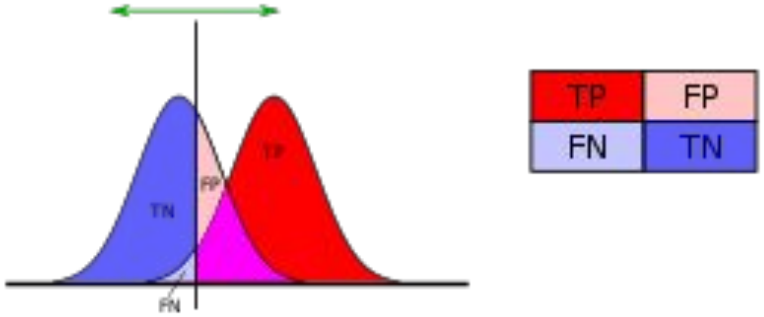
# ROC curves

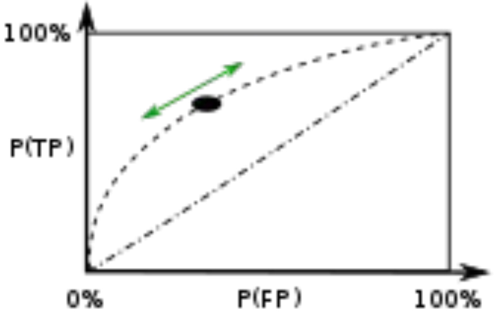
- Relationship between TPR and FPR
- The diagonal is chance classification (no predictive ability)




Source: https://en.wikipedia.org/wiki/Receiver_operating_characteristic

# ROC curves




- Relationship between TPR and FPR
- The diagonal is chance classification (no predictive ability)

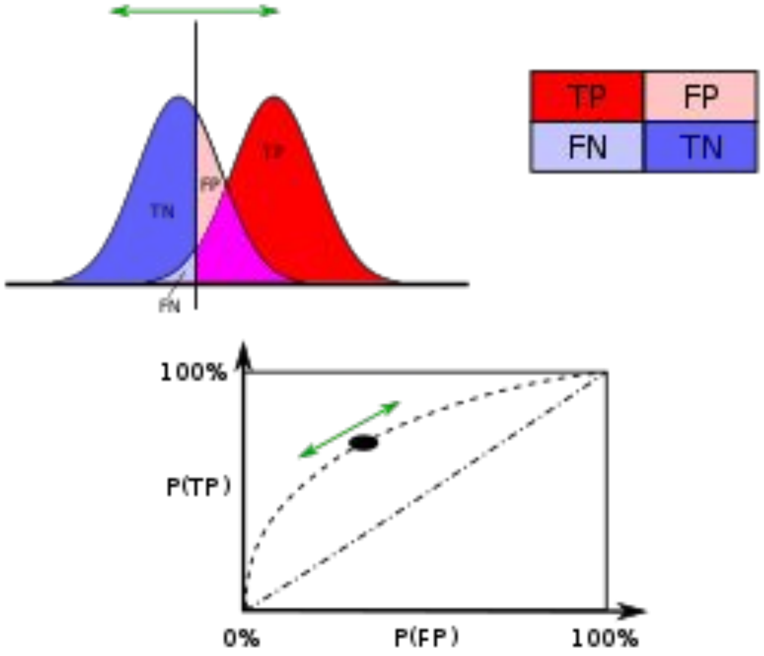
- Threshold for P(Y=1|x): 0
- No FN, no TN (all positive predictions)
- TPR = TP/(TP+FN) = TP/(TP+0) = TP/TP = 100%
- FPR = FP/(FP+TN) = FP/(FP+0) = FP/FP = 100%

| | | True observation | |
|---|---|---|---|
| | | 1 | 0 |
| Prediction | 1 | TP | FP |
| | 0 | 0 (FN) | 0 (TN) |

Source: https://en.wikipedia.org/wiki/Receiver_operating_characteristic

# ROC curves

- Relationship between TPR and FPR
- The diagonal is chance classification (no predictive ability)

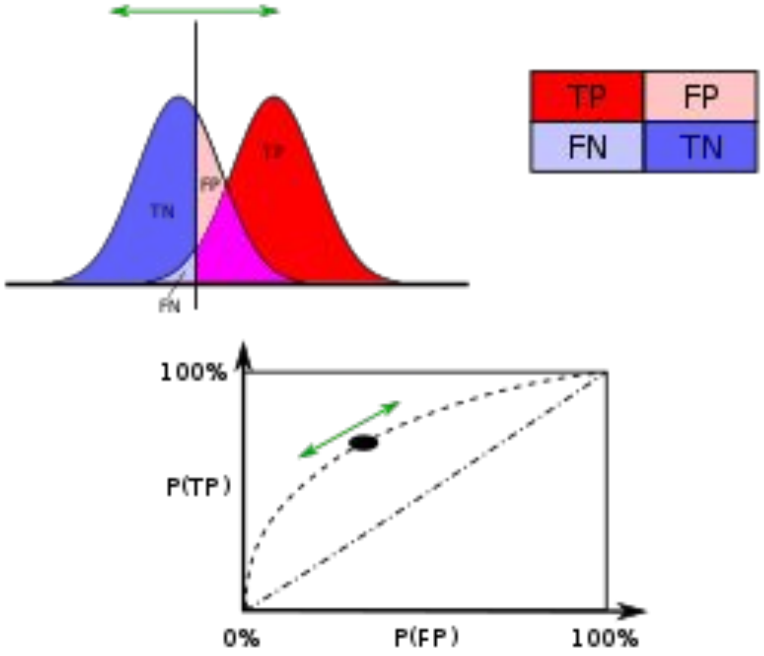



- Threshold for $P(Y=1|x)$: 1
- No TP, no FP (all negative predictions)
- TPR = (TP=0)/(TP=0+FN) = 0%;
- FPR = (FP=0)/(FP=0+TN) = 0%

| | | True observation | |
|---|---|---|---|
| | | 1 | 0 |
| Prediction | 1 | 0 | 0 |
| | 0 | FN | TN |

Source: https://en.wikipedia.org/wiki/Receiver_operating_characteristic

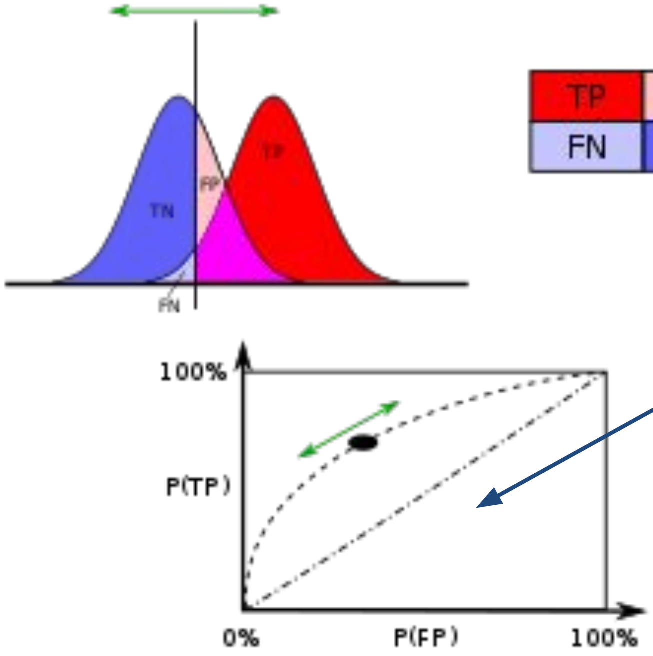
# ROC curves




- Relationship between TPR and FPR
- The diagonal is chance classification (no predictive ability)
- **The best is towards the left upper corner (TPR → 100%, FPR → 0%)**

Source: https://en.wikipedia.org/wiki/Receiver_operating_characteristic

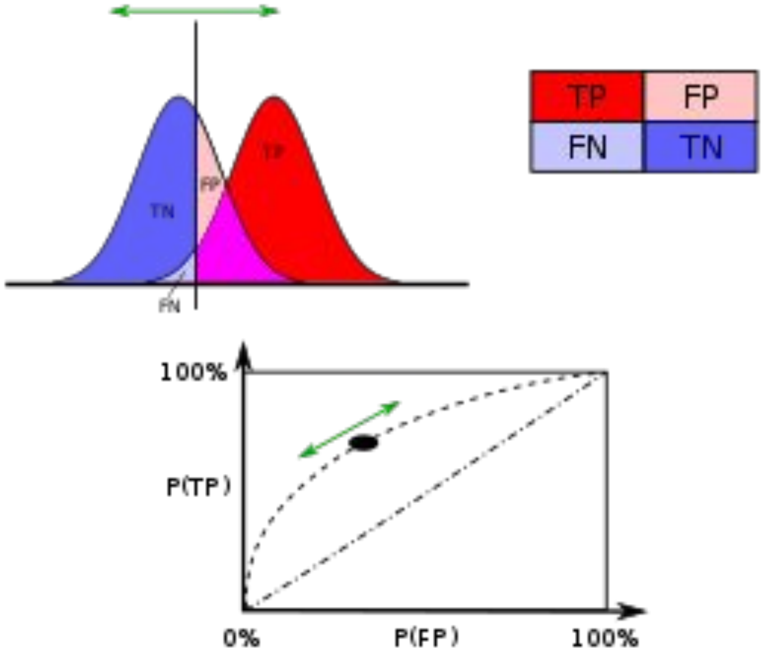
# Area under the curve (AUC)




- AUC = 0.5: random guessing
- AUC = 1: perfect classifier
- AUC > 0.8: good classifier

Source: https://en.wikipedia.org/wiki/Receiver_operating_characteristic

# Cut-off thresholds




- Two types of error: **FP**, **FN**: sometimes one error may be more critical than the other
- e.g. for a bank may be more important to correctly identify borrowers who will default at the expense of an increase of false positives (and of the total error rate) → lower cut-off for $P(y=1|x)$
- e.g. in a pandemic, you may want to be sure about detecting carriers, even if this means increasing the FPR

Source: https://en.wikipedia.org/wiki/Receiver_operating_characteristic

# ROC curves

- demonstration 4.2

→ 4.classification.Rmd