Introduction to Bioinformatics and Biostatistics

Marta R. Hidalgo
Bioinformatics and Biostatistics Unit, CIPF

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Unidad de Bioinformática y Bioestadística

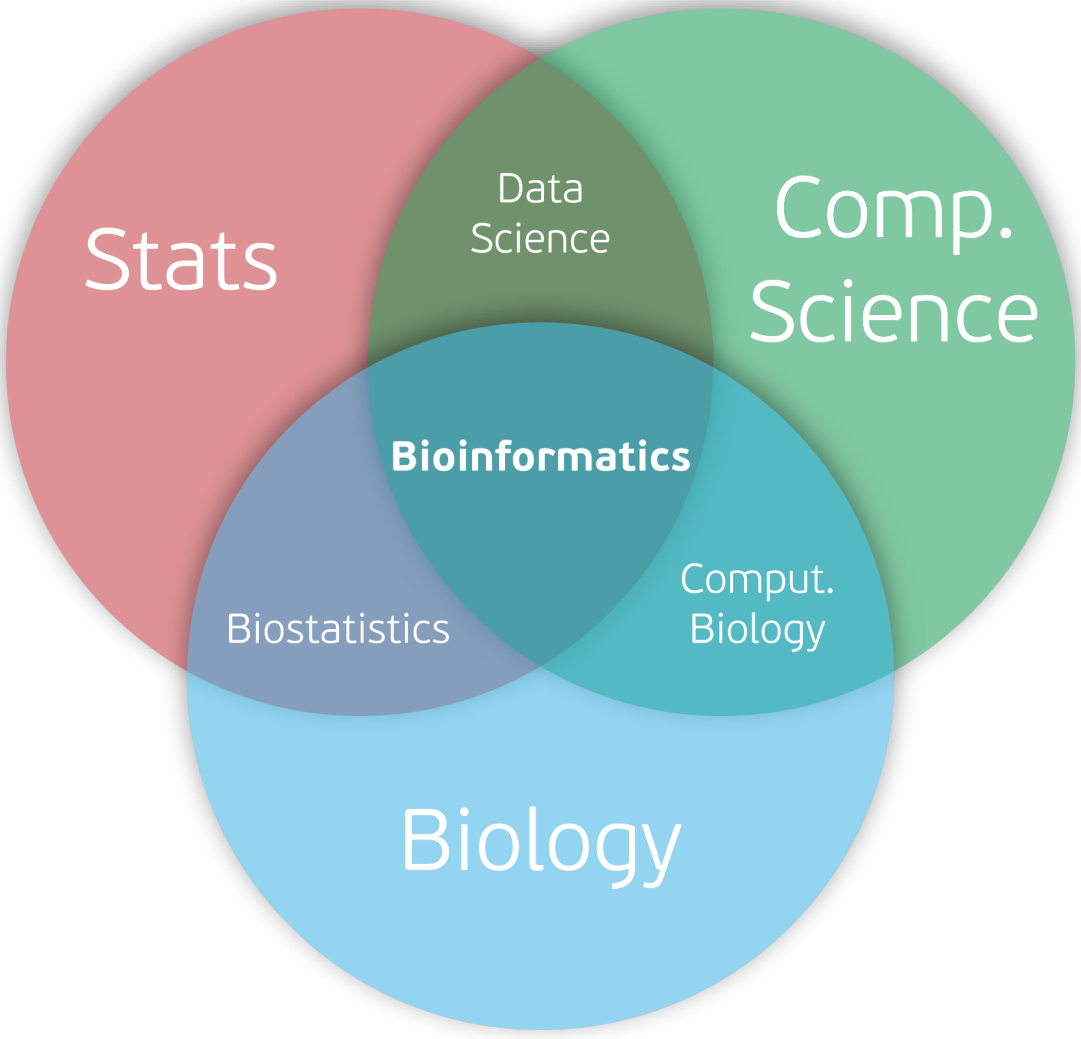
PRINCIPE FELIPE
CENTRO DE INVESTIGACION

WODA
WEB-BASED OMICS DATA ANALYSIS

Outline

Outline

What is Bioinformatics?



What is Bioinformatics?

Big Data

DNA contains 3.200M of bases
Each genome contains ~20.000 genes
Studies include up to 100.000 patients

Bioinformatics Database Building

Management of massive biological resource data and development of databases

Structural Bioinformatics

Prediction of the structure of a protein
Creation of new drugs

Bioinformatics analysis

Sequencing techniques:
- Disease mechanisms
- Phylogenetics
- Population genetics

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OMICS

Bioinformatics analysis

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What is Bioinformatics?

- Genomics
- Metabolomics
- Proteomics
- Transcriptomics

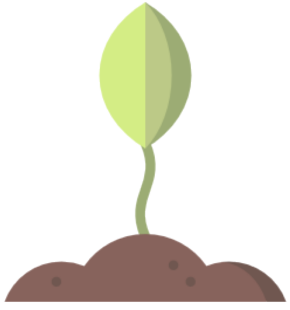
Data bases

What for?

- experiment data (raw / processed)
- variants
- pathways

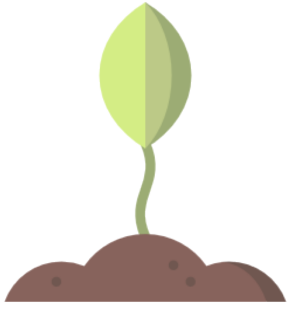
Outline

Variables



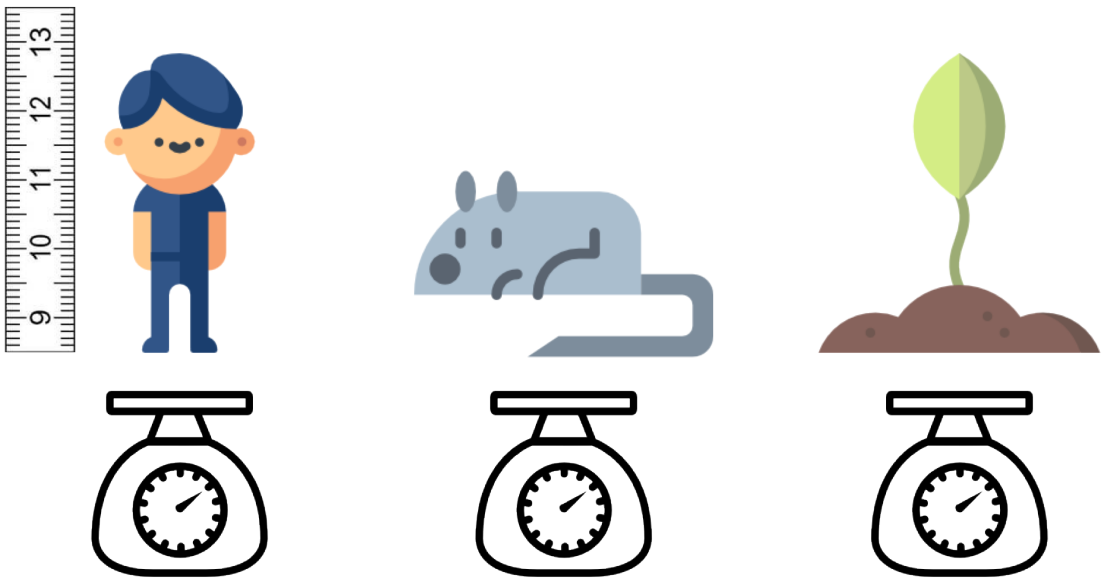
	Human	Mouse	Plant

Variables



	Human	Mouse	Plant
Height	170cm	25cm	10cm

Variables



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Height	170cm	25cm	10cm
Weight	68.000g	150g	10g

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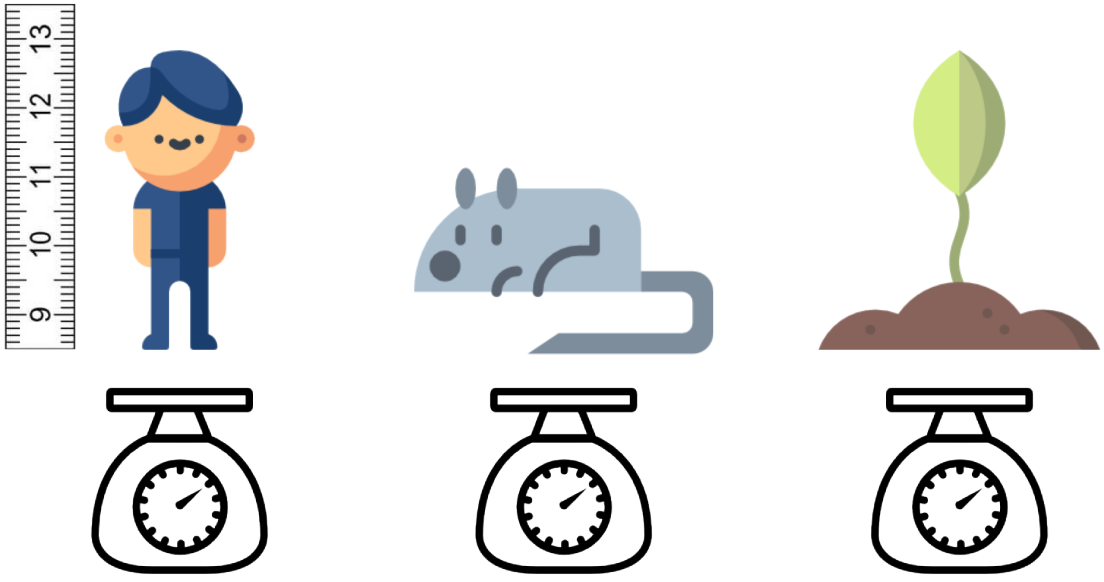
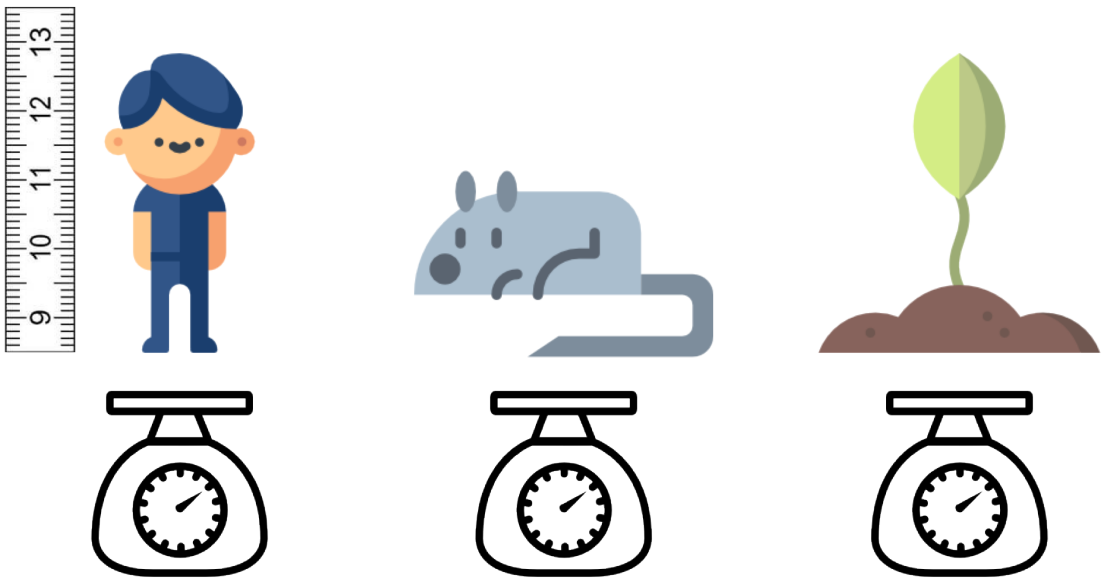


	Human	Mouse	Plant
Height	170cm	25cm	10cm
Weight	68.000g	150g	10g
Color eyes	Brown	Black	NA

Variables



	Human	Mouse	Plant
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Color eyes	Brown	Black	NA
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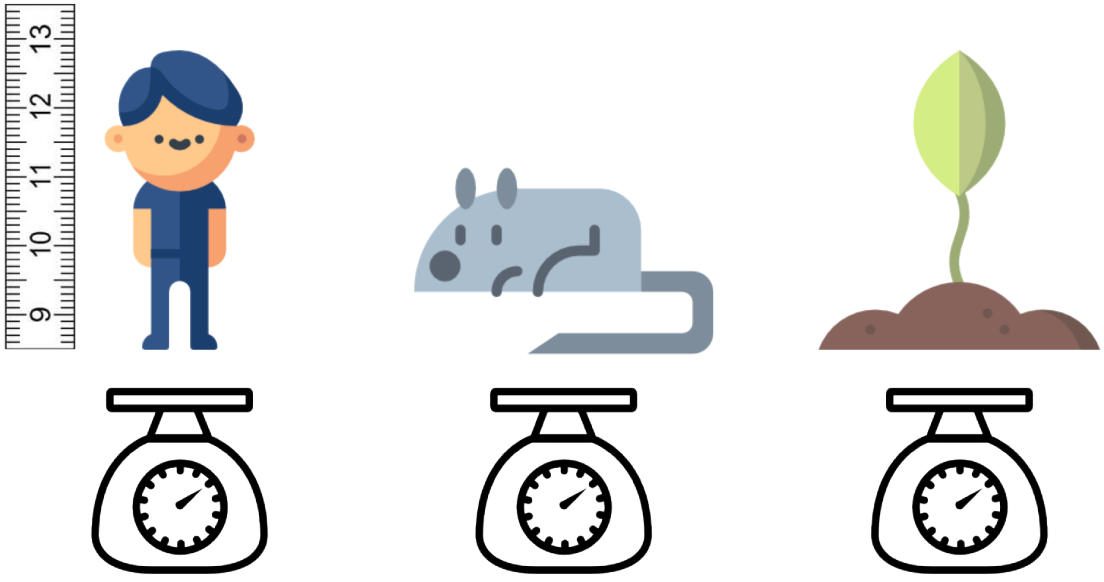
Variables



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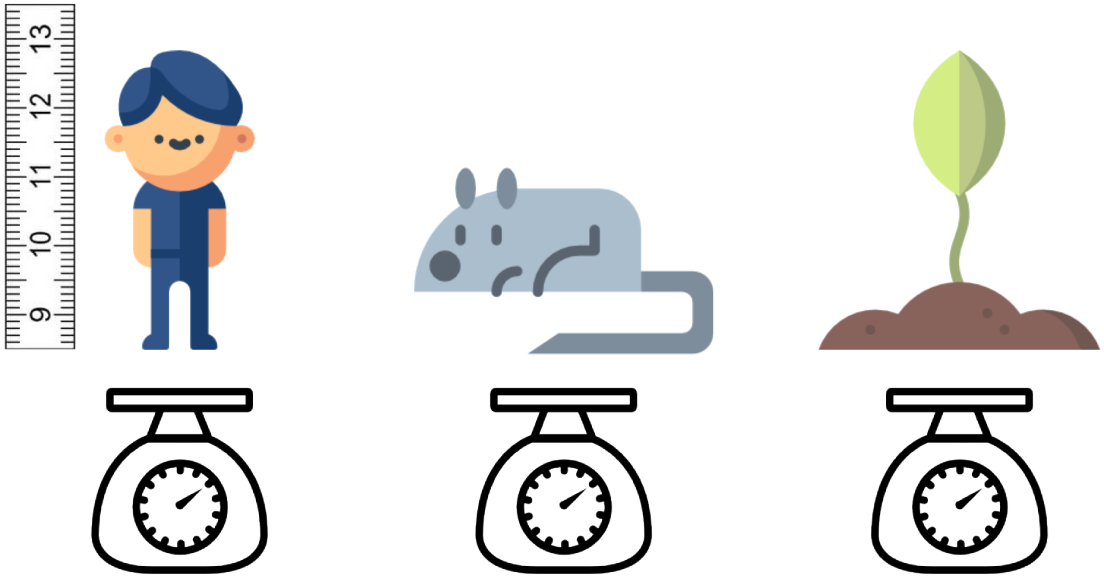
NUMERICAL

Variables



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NUMERICAL

CATEGORICAL

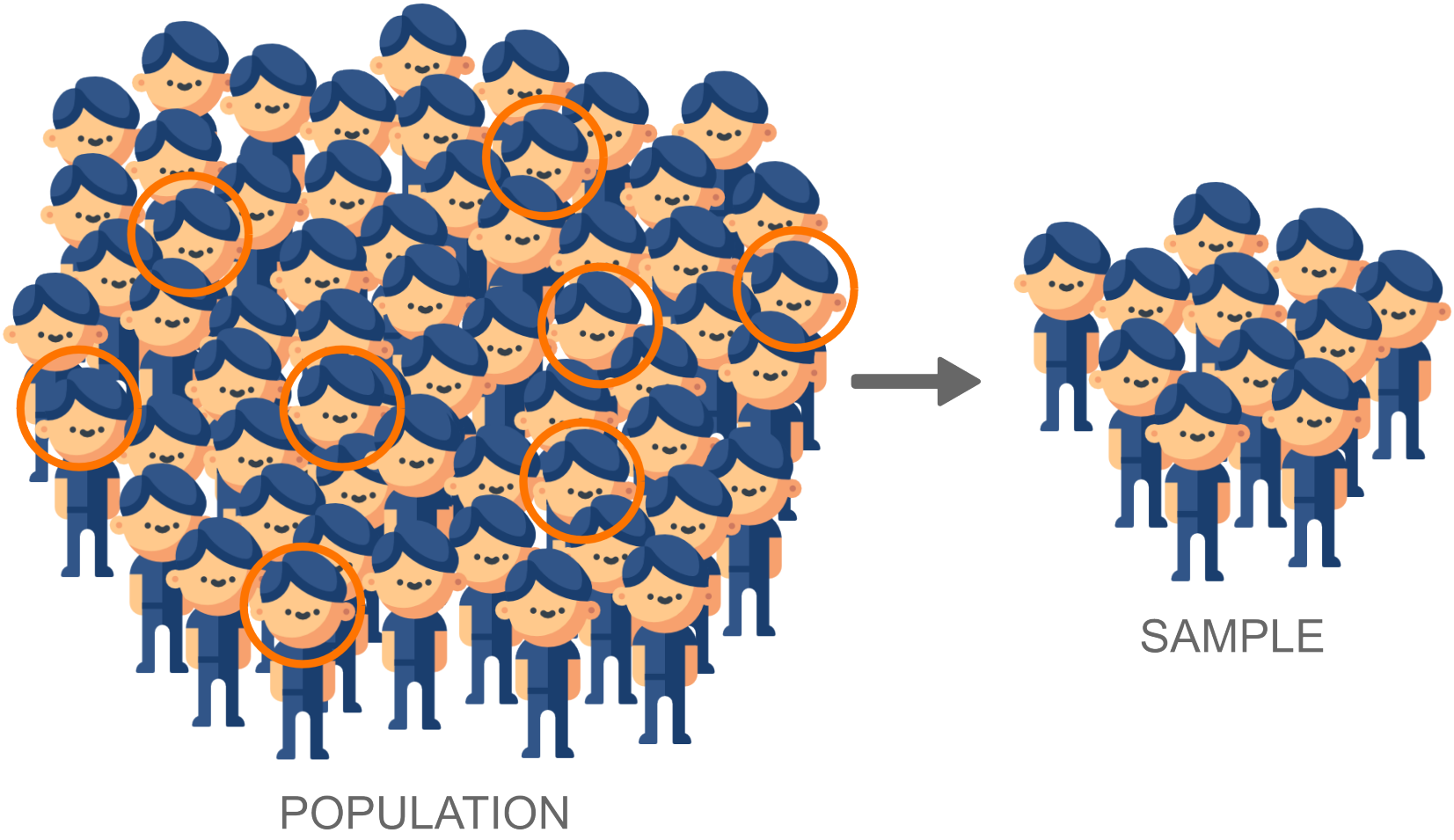
Population

POPULATION

Population

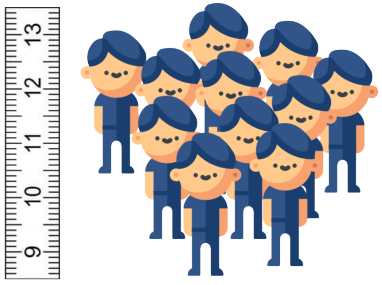
POPULATION

Population



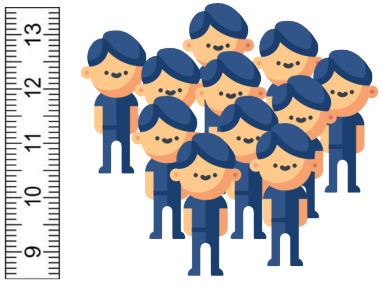
Numerical variable

Numerical variable



	Height
Human 1	1'70
Human 2	1'53
Human 3	2'01
Human 4	1'82
Human 5	1'65
Human 6	1'73
Human 7	1'91
Human 8	1.81

Numerical variable



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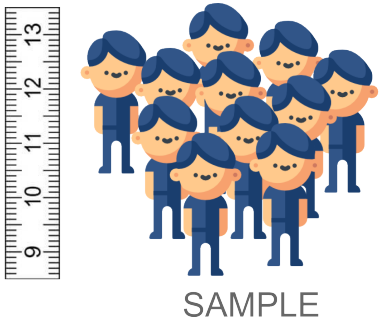
SAMPLING DISTRIBUTION

Numerical variable



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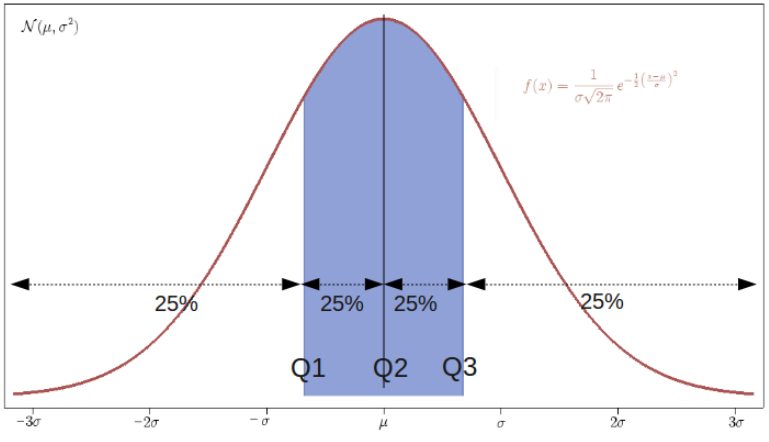
SAMPLING DISTRIBUTION

- mean
- median
- mode
- variance

Quantiles, Quartiles and Percentiles

Quantiles

Cut points dividing the observations in a sample in intervals of equal dimension.



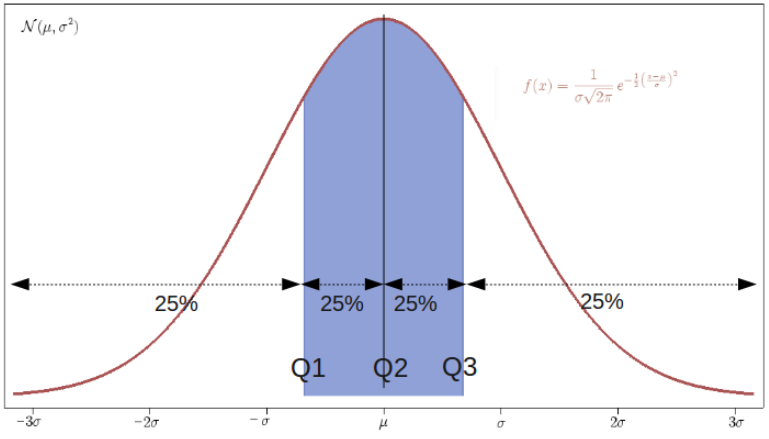
Quantiles, Quartiles and Percentiles

Quantiles

Cut points dividing the observations in a sample in intervals of equal dimension.

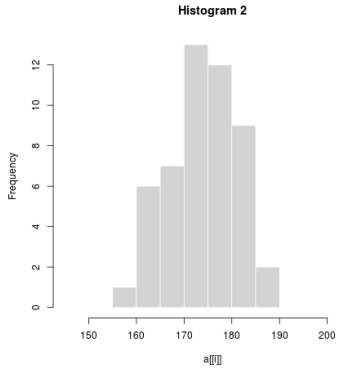
Special cases

- 2-quantile: median
- 4-quantile: quartiles
- 100-quantile: percentiles

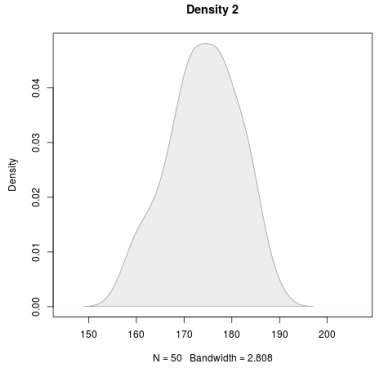


Representing Numerical variables

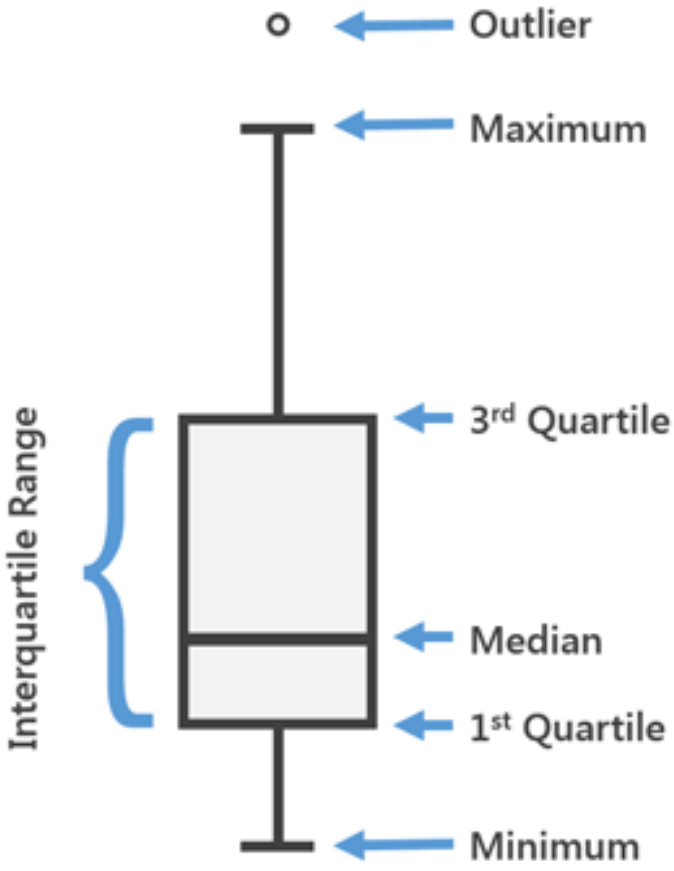
Histogram



Density



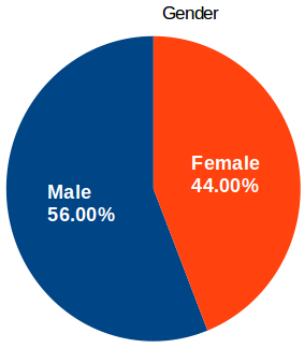
Boxplot



Representing cathegorical variables

Pie Chart

- One cathegorical variable



Representing cathegorical variables

Pie Chart

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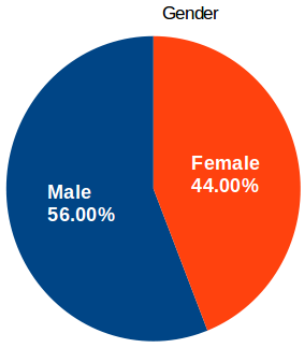
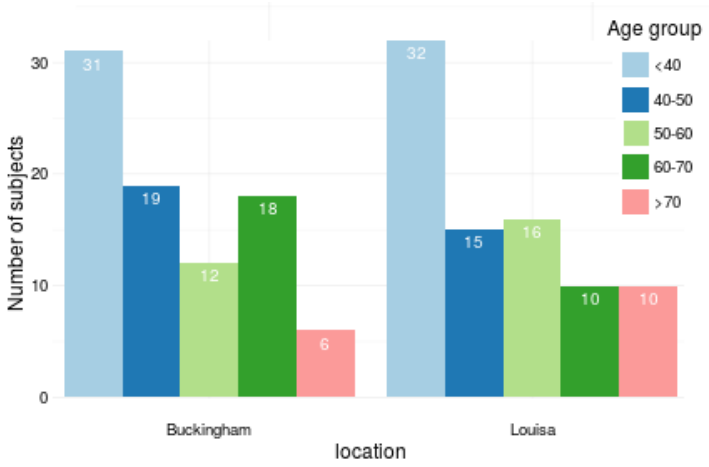
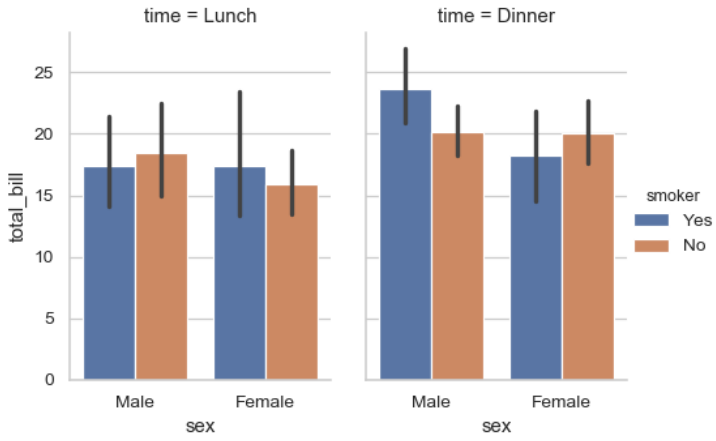


Bar plot

- Multiple cathegorical variables
- Relation cathegorical and numerical variables





Sampling distribution

SAMPLE

POPULATION

Sampling distribution

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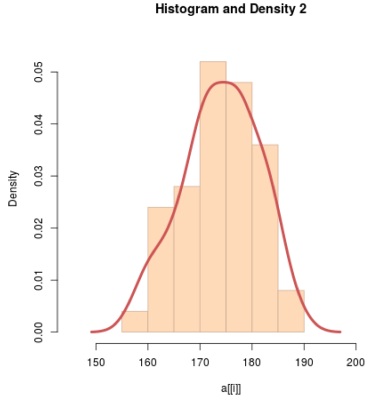
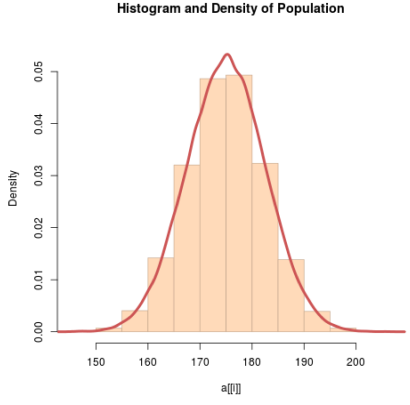
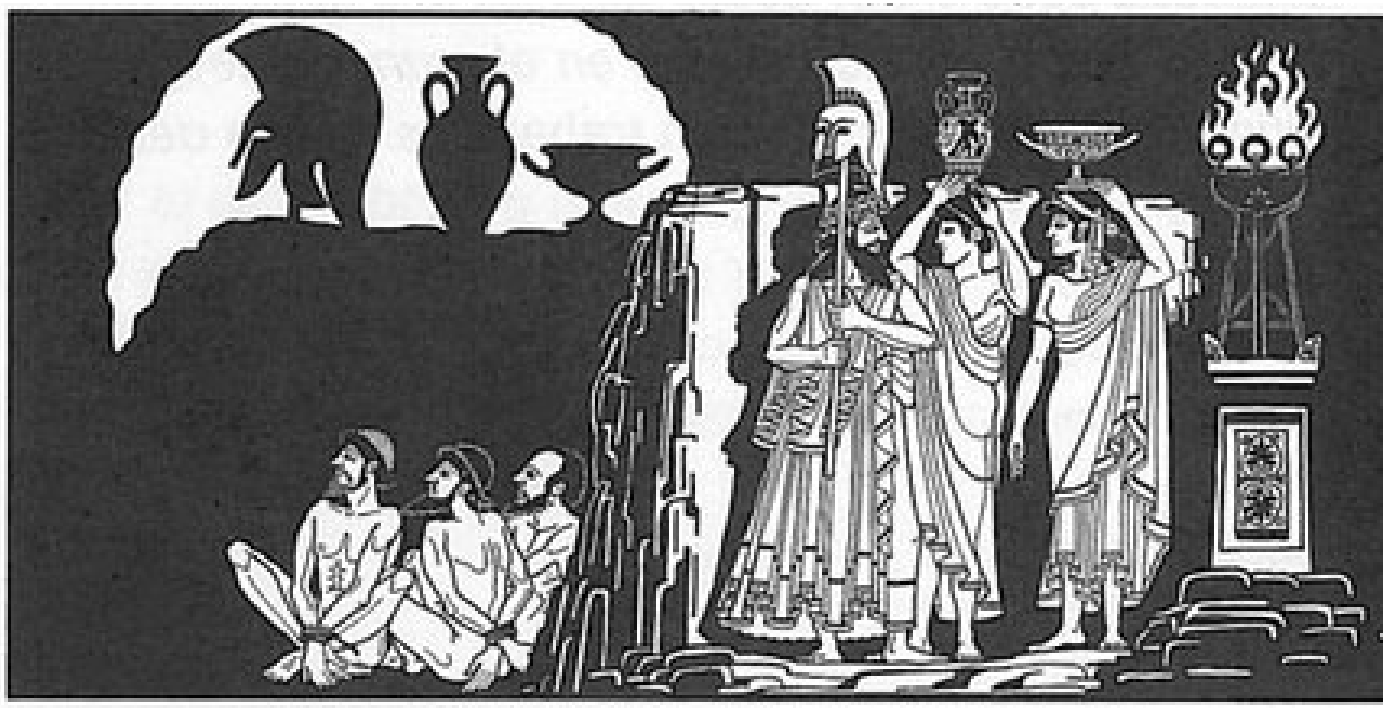




Sample / Population

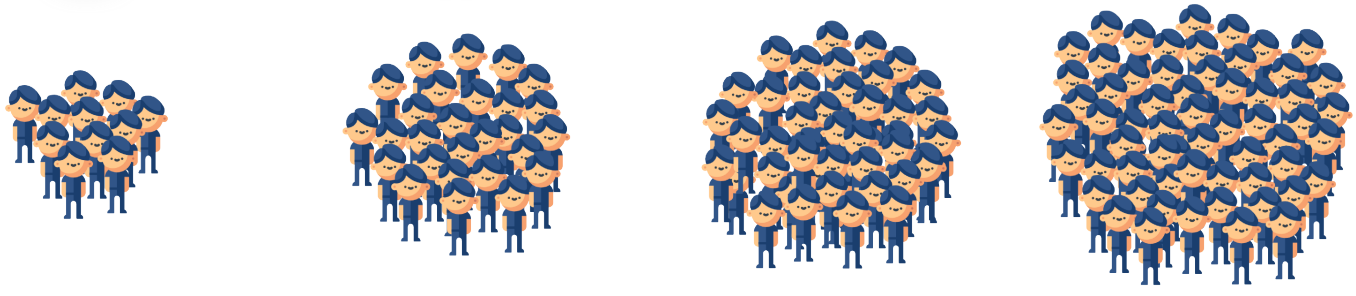
Plato's Cave Myth



Sample Size

Shadow

N



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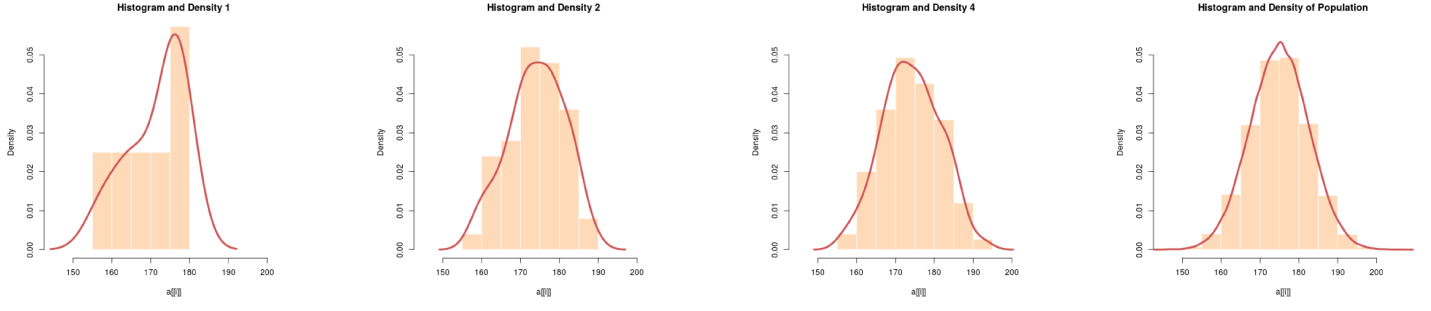
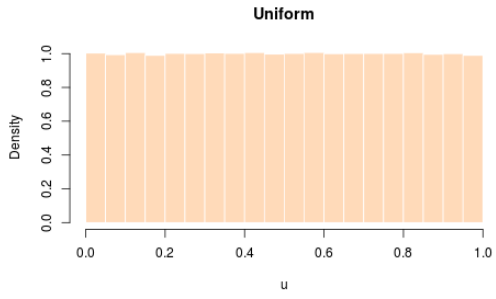


Distributions

Uniform

- All values equal probability
- Parameters: min, max



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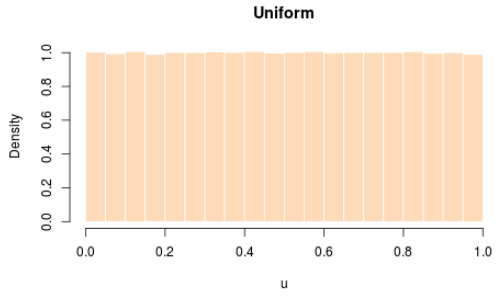
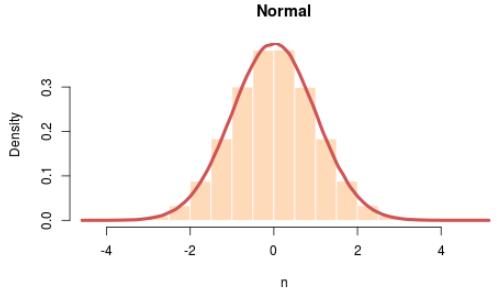


Normal

- Gauss Bell
- Parameters: μ, σ



Distributions

Uniform

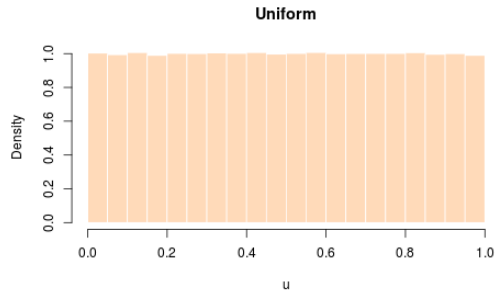
- All values equal probability
- Parameters: min, max

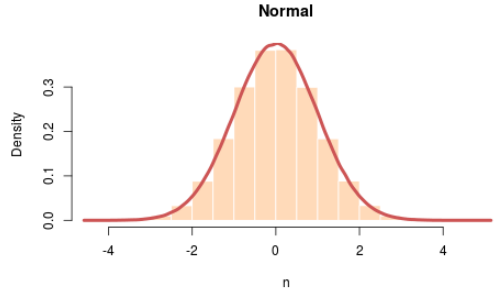
Normal

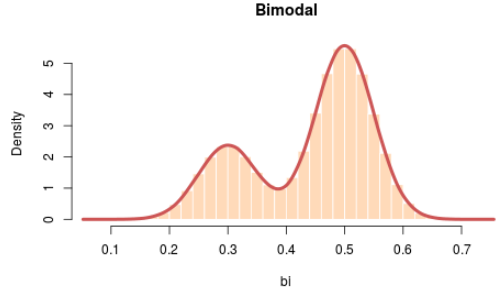
- Gauss Bell
- Parameters: μ, σ

Bimodal

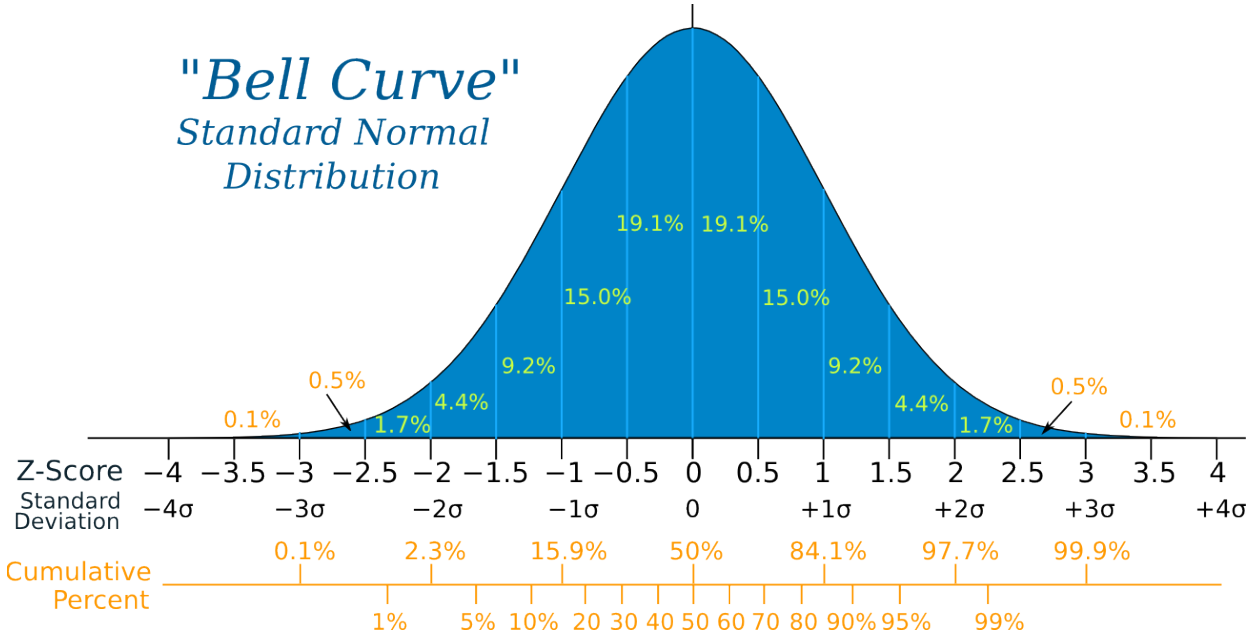
- Two different distributions combined
- Parameters: $(\mu_1, \sigma_1), (\mu_2, \sigma_2)$







Normal distribution



Inference

Inference

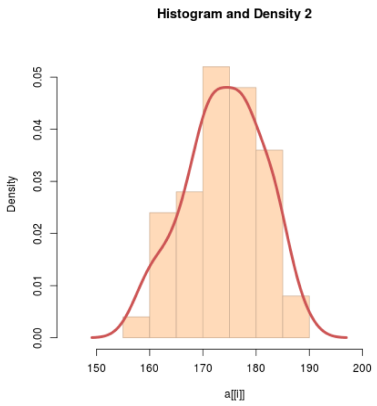
Conclude something about a parameter in the population from the data in the sample

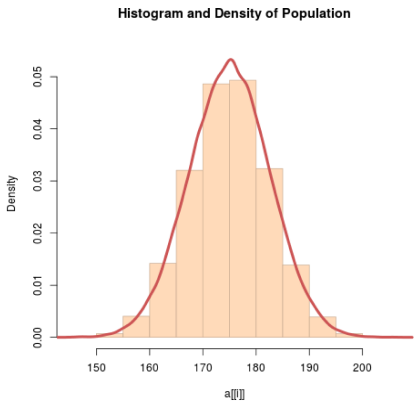
- Confidence intervals
- Hypothesis tests
- Specific tests

Confidence intervals

SAMPLE

POPULATION





Confidence intervals

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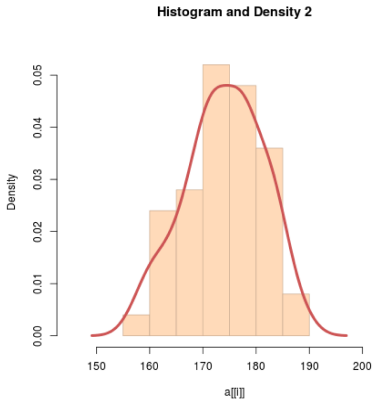

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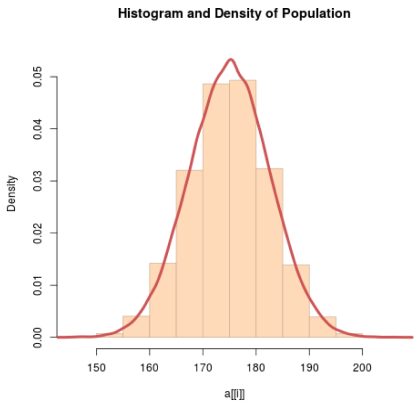

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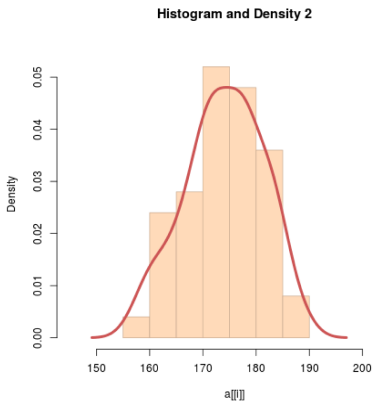
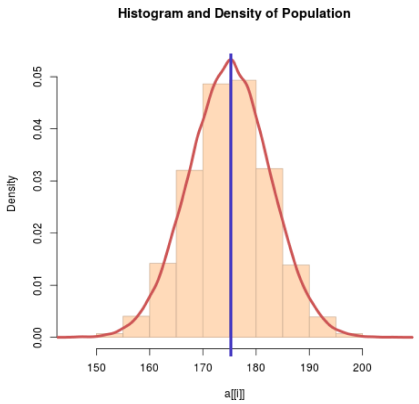


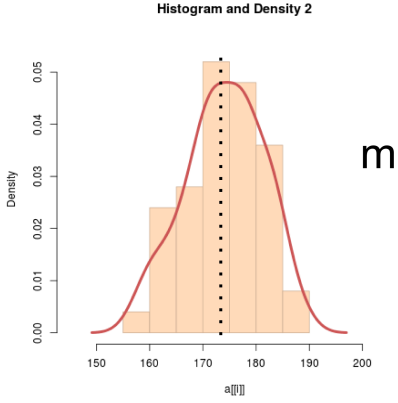


$\mu = 175$

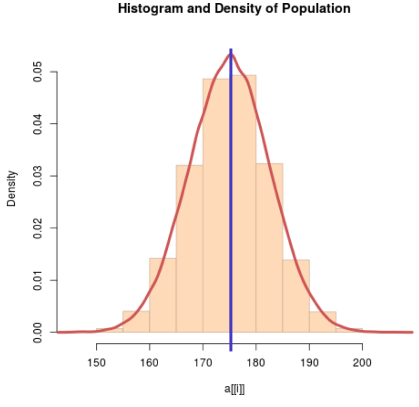
Confidence intervals

SAMPLE

POPULATION



mean = 173



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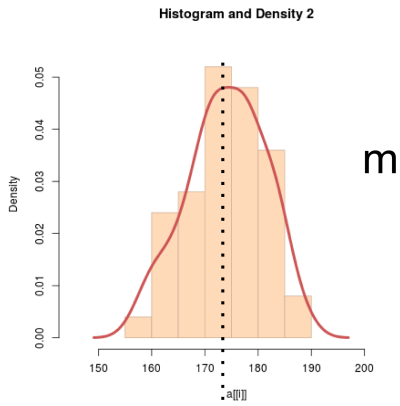
Confidence intervals

SAMPLE

POPULATION



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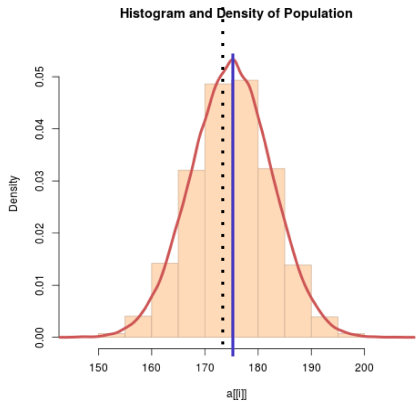

μ = 175

Confidence intervals

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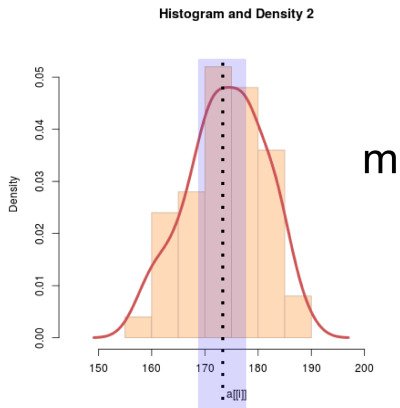
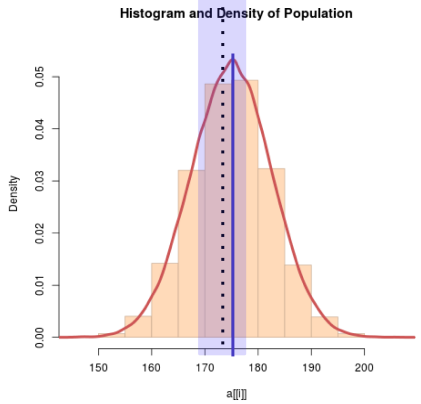
POPULATION



mean = 173

C.I. = [168, 178]



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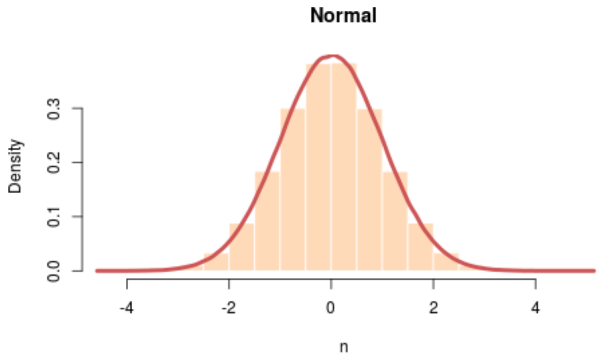
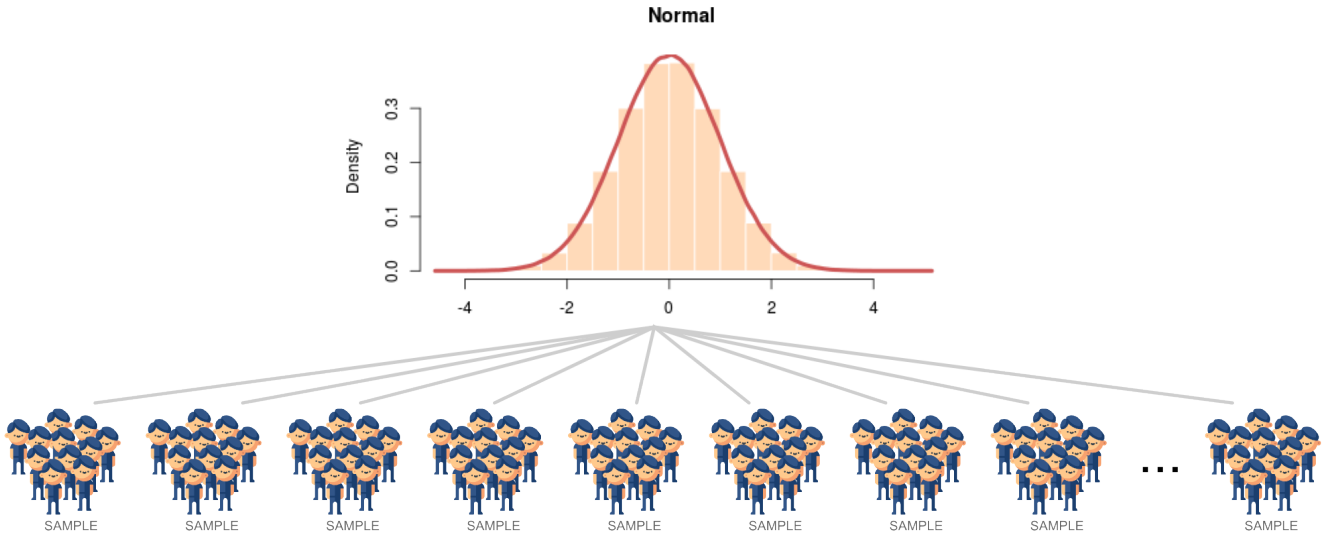
Confidence intervals

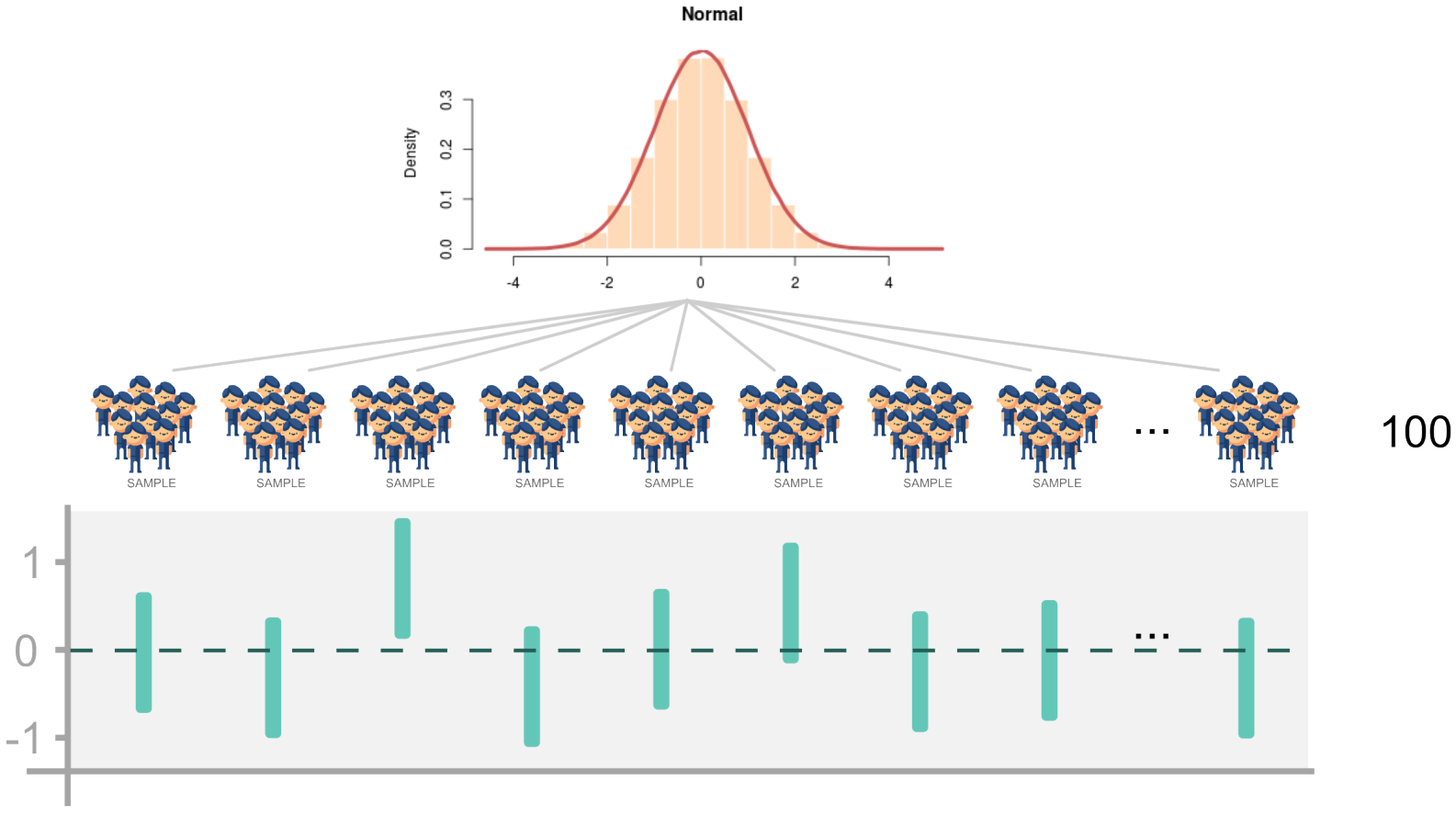


Confidence intervals



100

Confidence intervals



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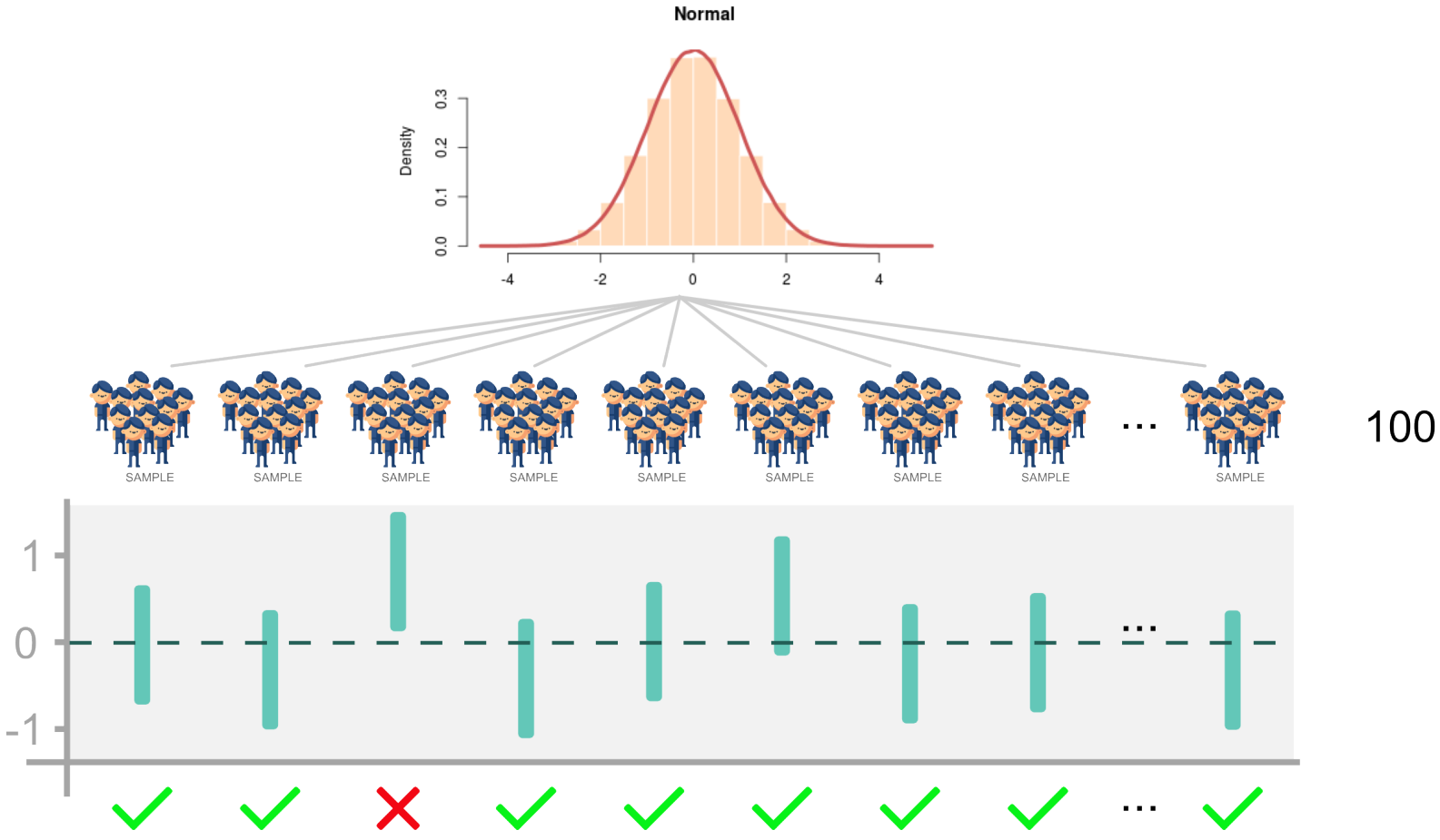

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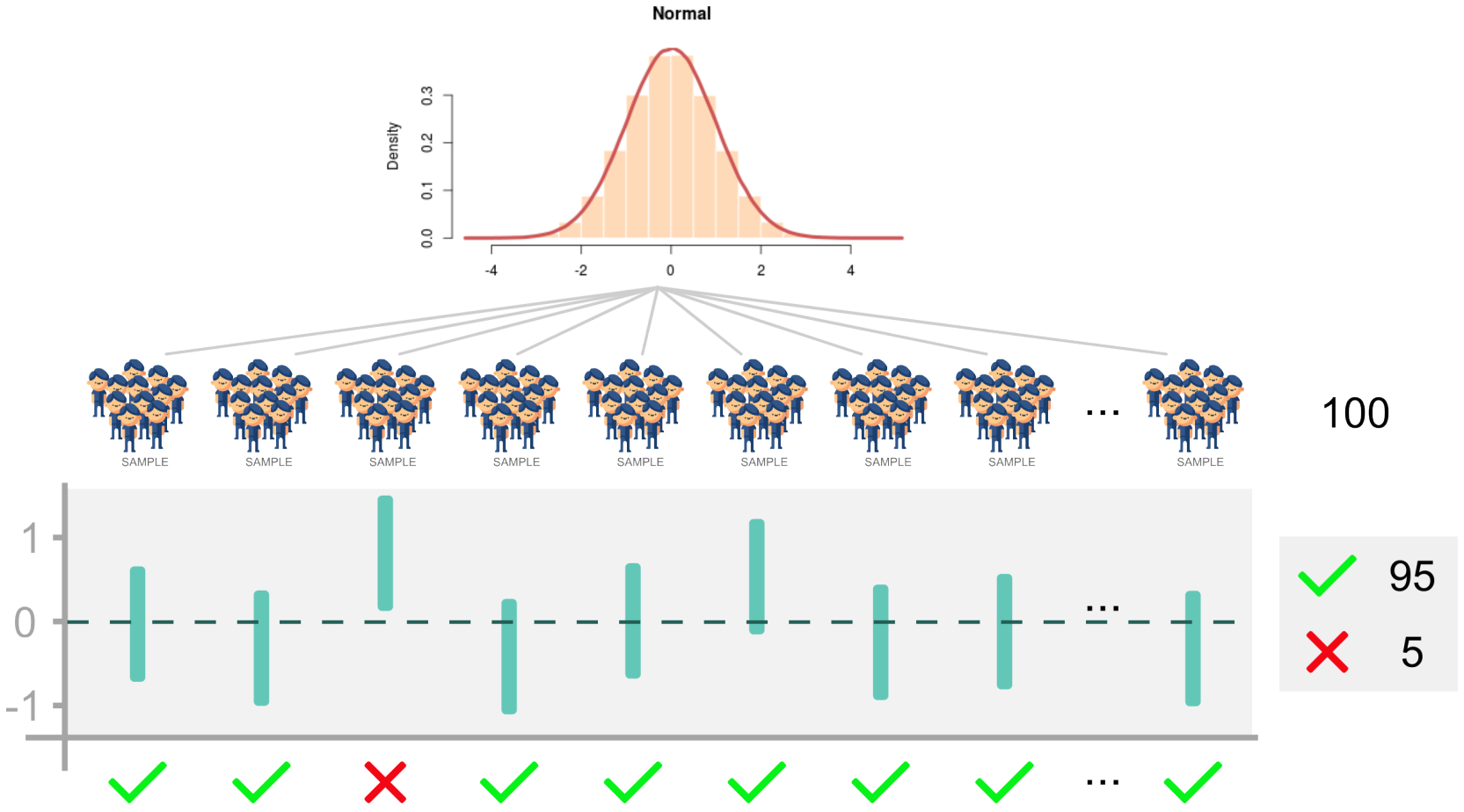


Hypothesis tests

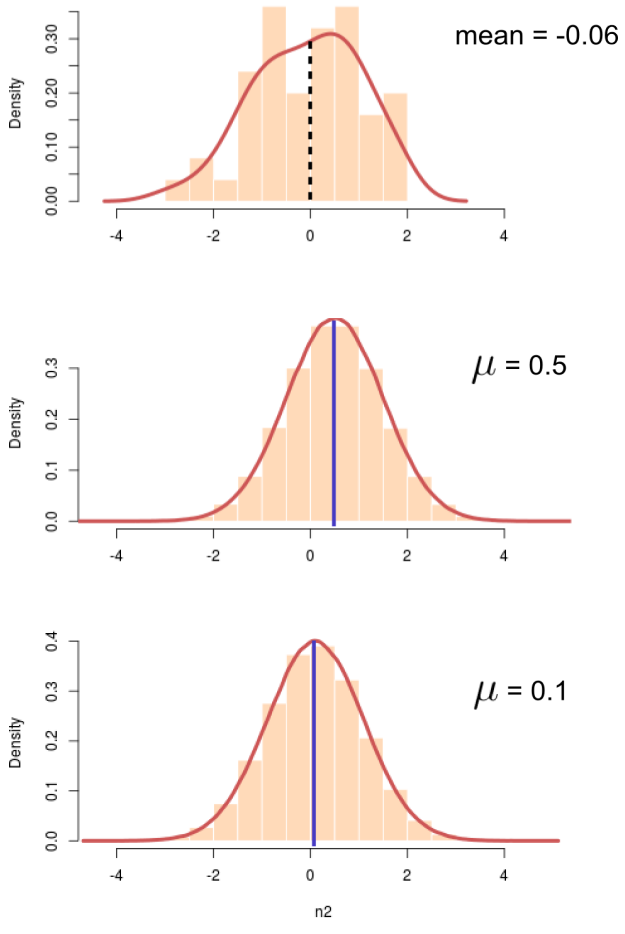
- Null hypothesis:

$$H_0 : \mu = 0.5$$

- Alternative hypothesis:

$$H_A : \mu \neq 0.5$$

- Compute confidence interval I.
- If $0.5 \notin I$, reject H_0.
- If $0.5 \in I$, H_0 is not rejected.



Hypothesis tests

t-test

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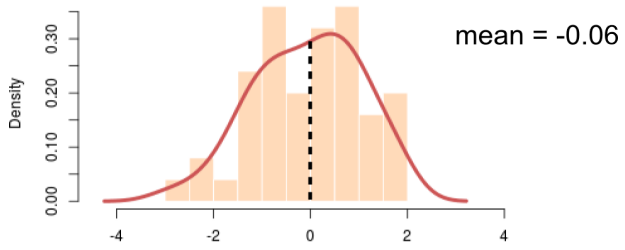

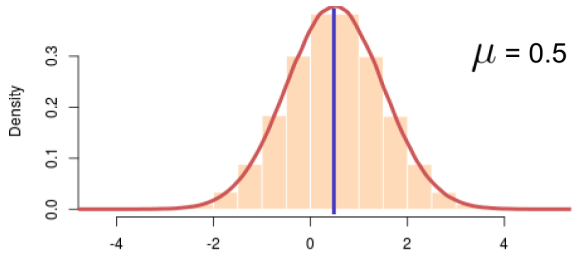

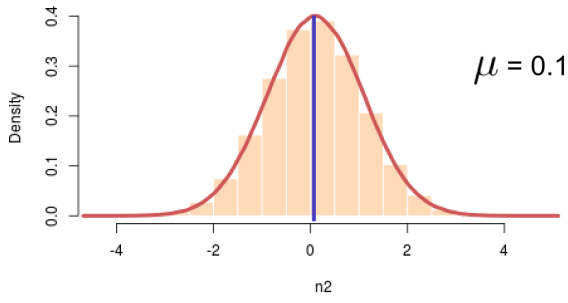

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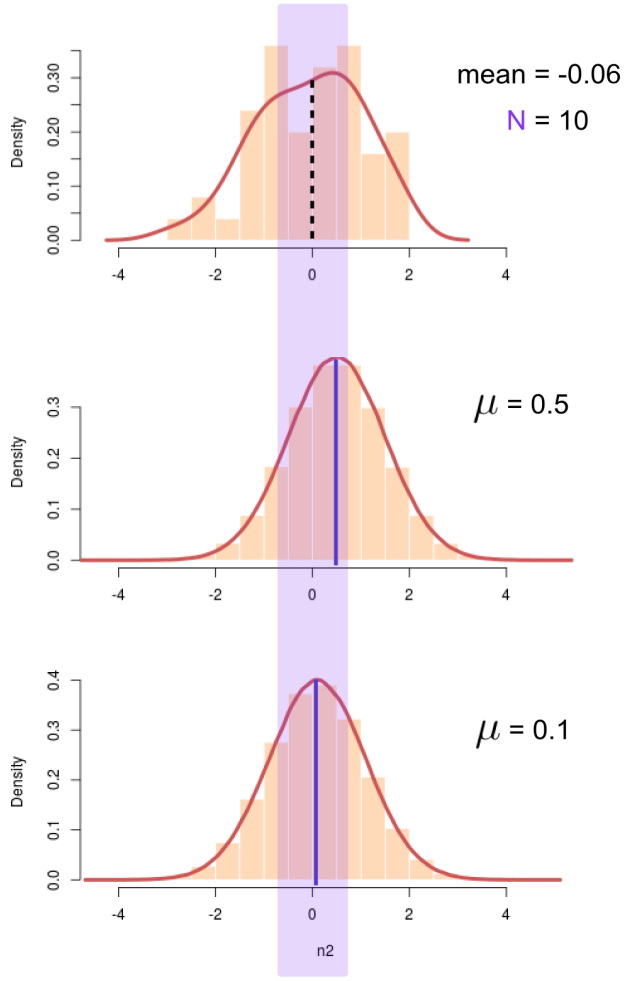

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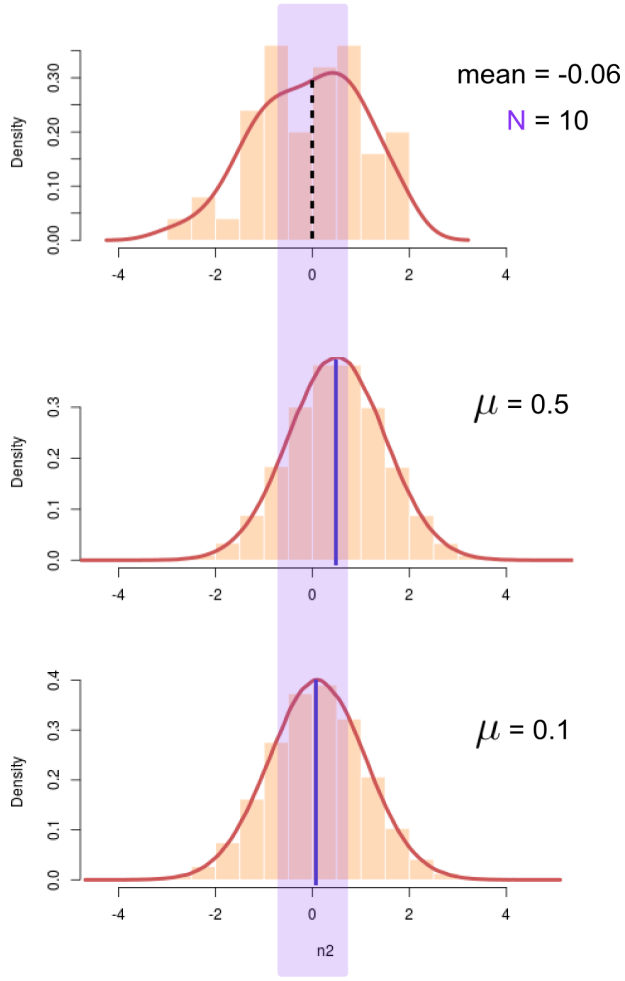

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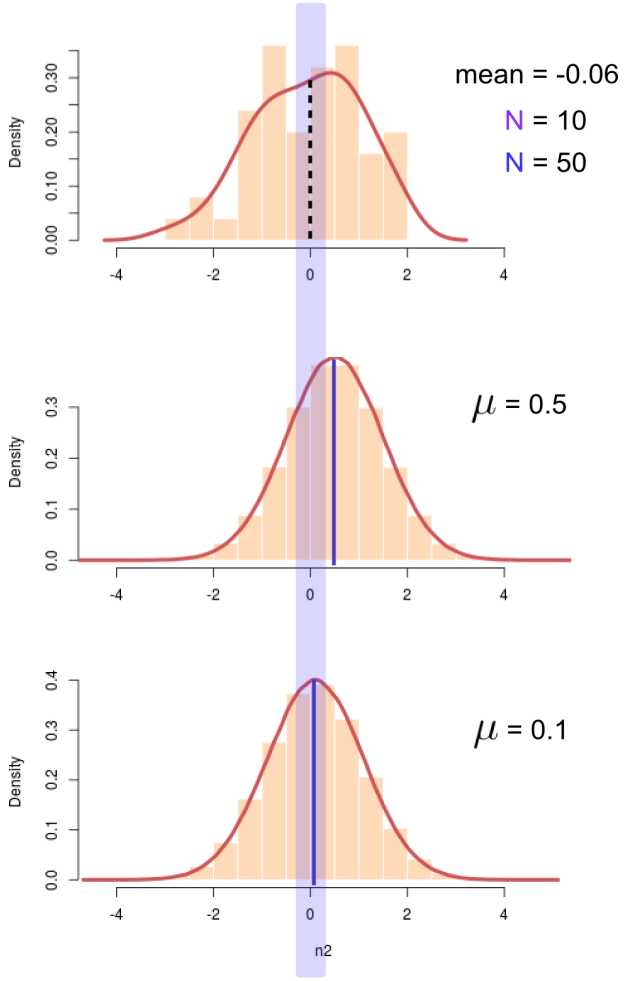

Mean comparison

- Null hypothesis:

$$H_0 : \mu_1 = \mu_2$$

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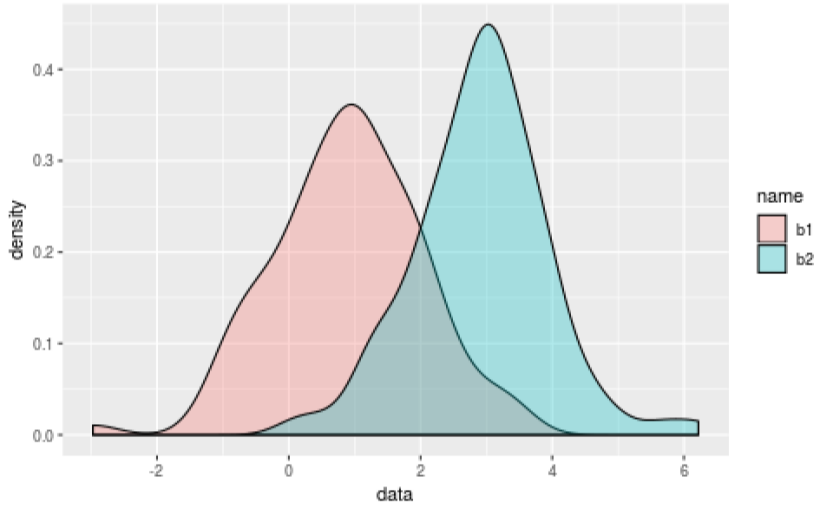
- t-test, Wilcoxon
- Paired samples?



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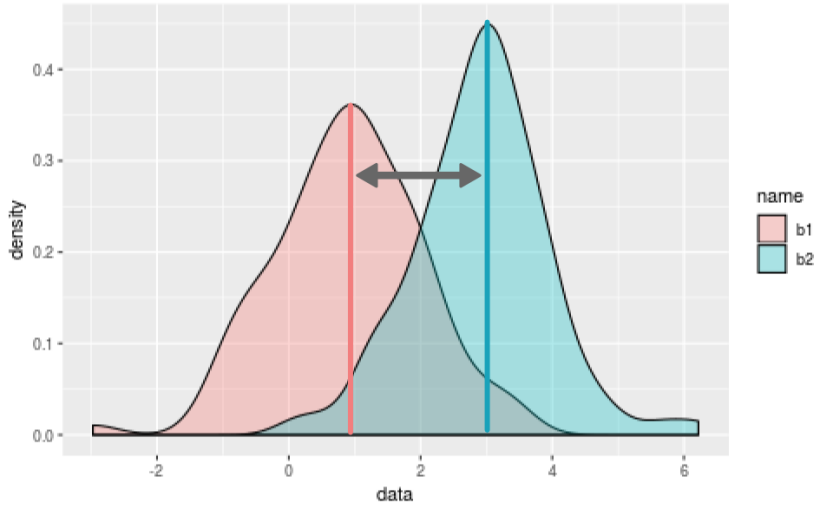

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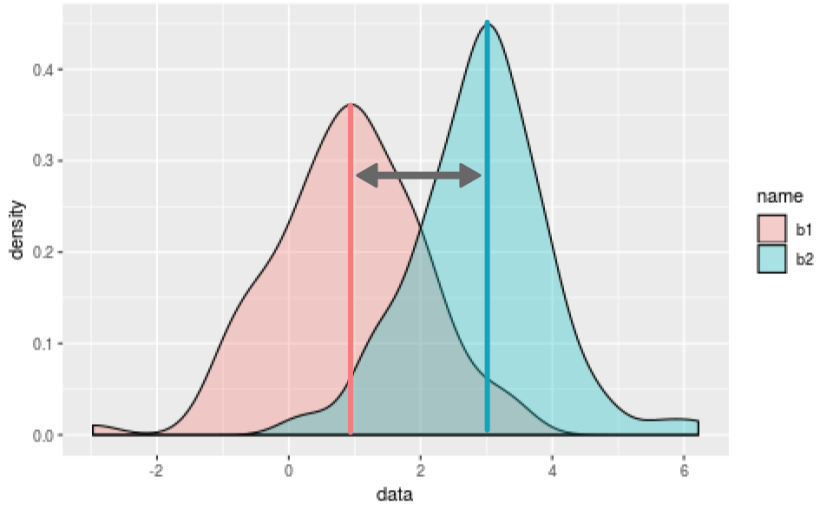

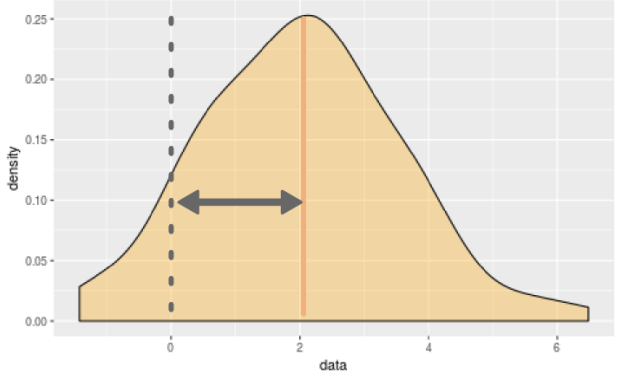

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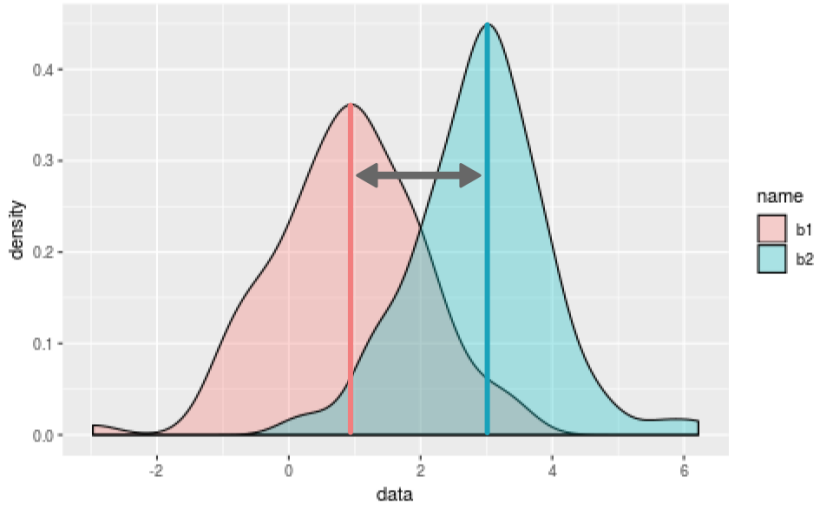

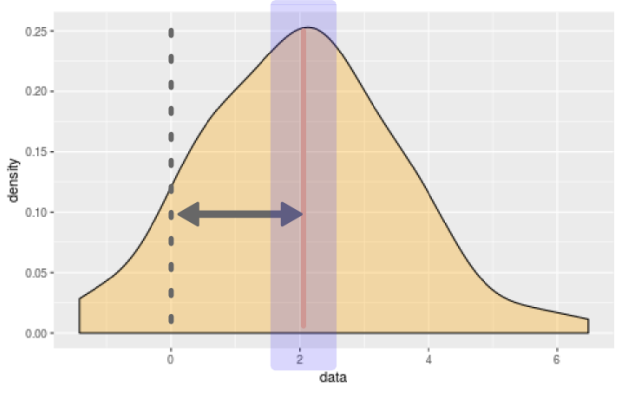

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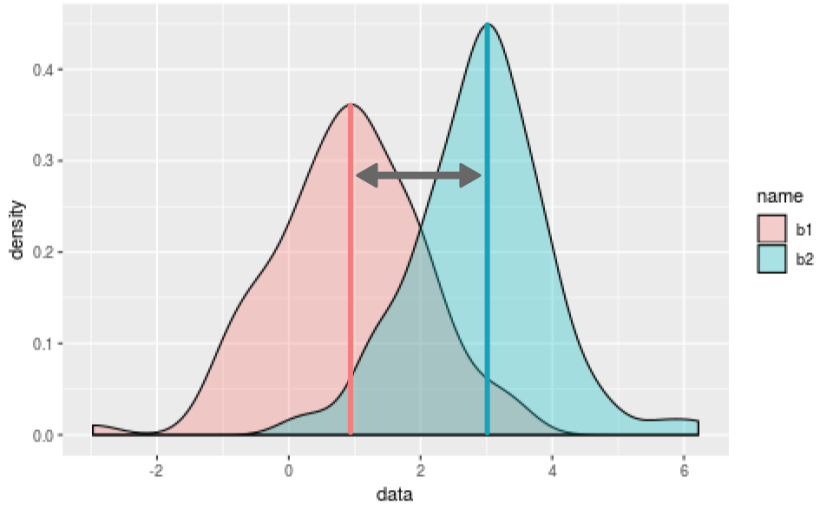

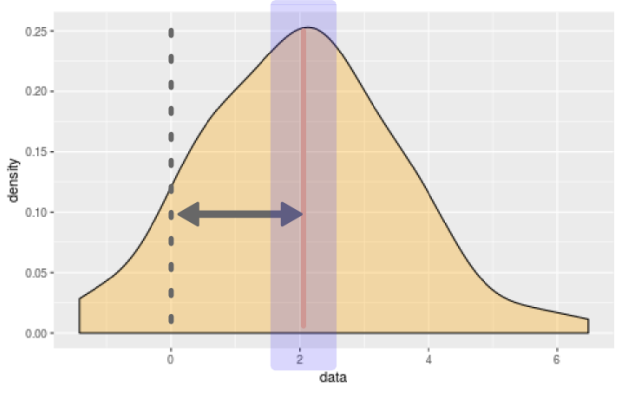

Fisher Test

Contingency table

Matrix that displays the frequency distribution of the variables

	Infected	Not infected	
Inoculated	3	276	279
Not inoculated	66	473	539
	69	749	818

Cholera Inoculation Study, 1894-96

- H_0 : Proportions are the same
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The end

Enjoy the course!!