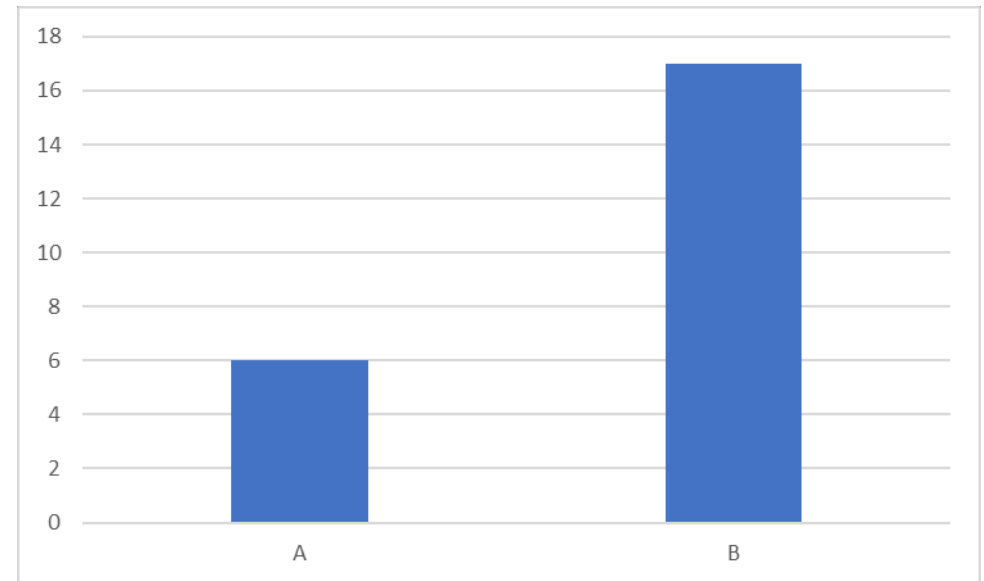
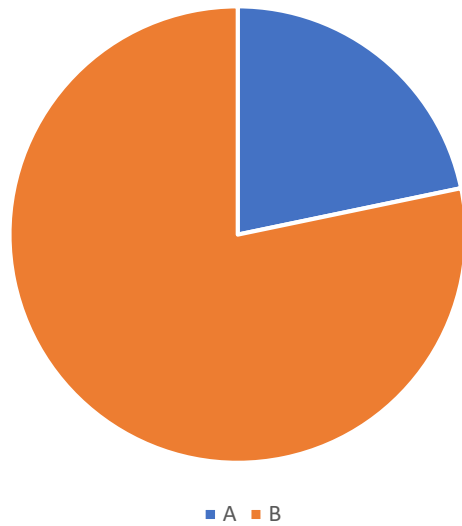


Στατιστική

Πληθυσμός - Δήγμα



Περιγραφική Στατιστική



Μέση τιμή και διακύμανση Δήγμα

Sample mean , Sample variance

Δήγμα

$$\langle x \rangle = \frac{\sum_i^n x_i}{n}$$

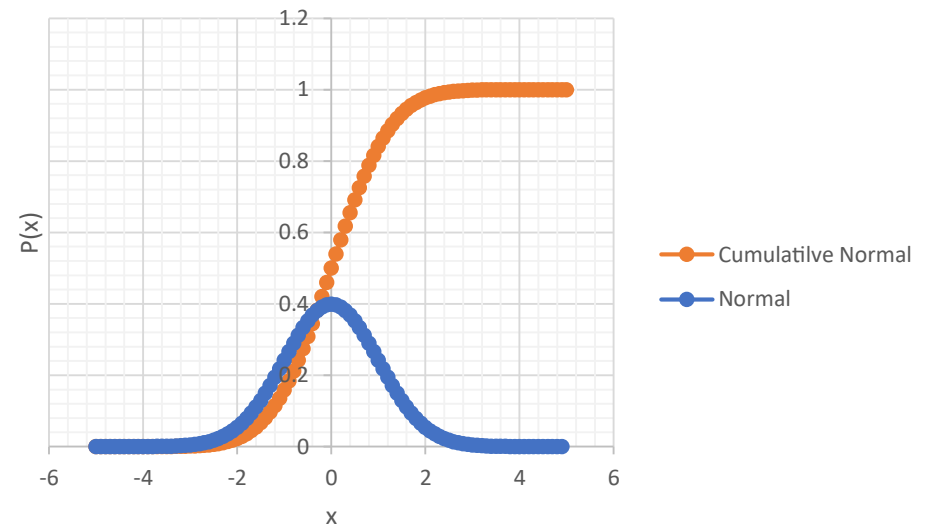
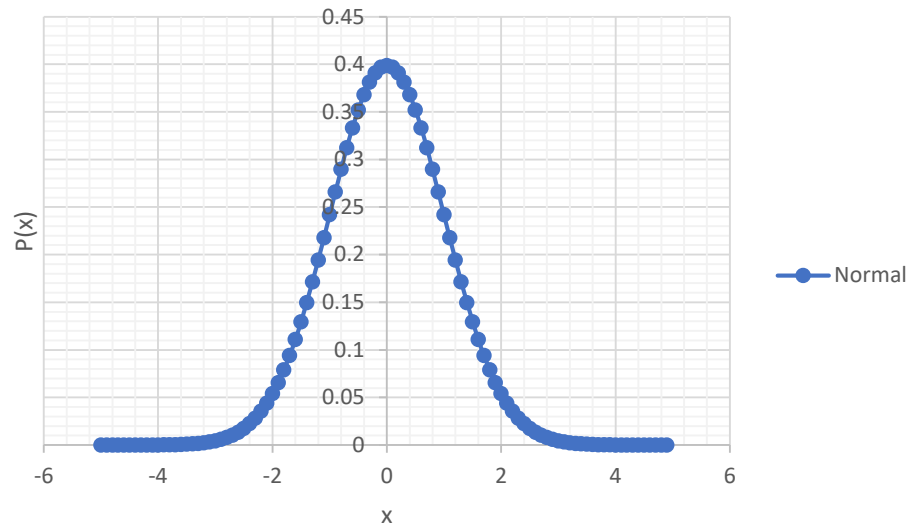
$$s^2 = \frac{\sum_i^n (x_i - \langle x \rangle)^2}{n-1}$$

Πληθυσμός

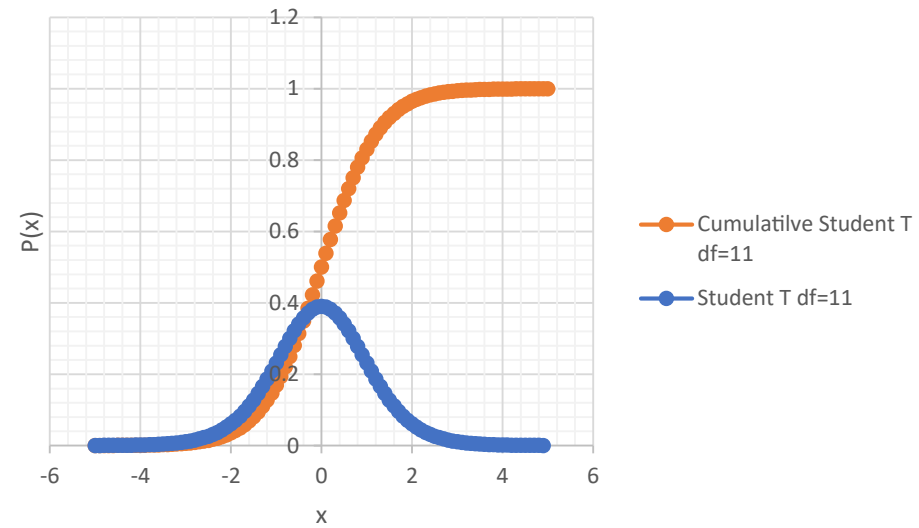
$$\mu = \frac{\sum_i^n x_i}{n}$$

$$\sigma^2 = \frac{\sum_i^n (x_i - \langle x \rangle)^2}{n}$$

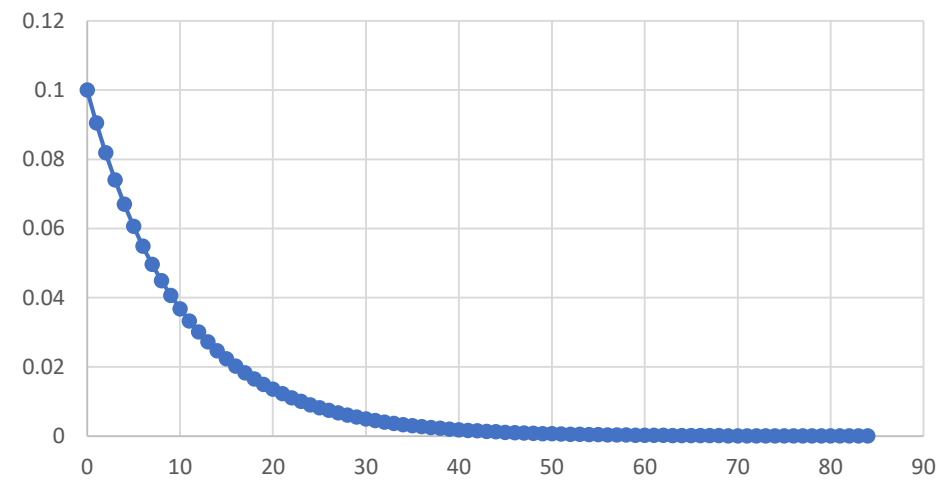
Κανονική κατανομή



Κατανομή Student T



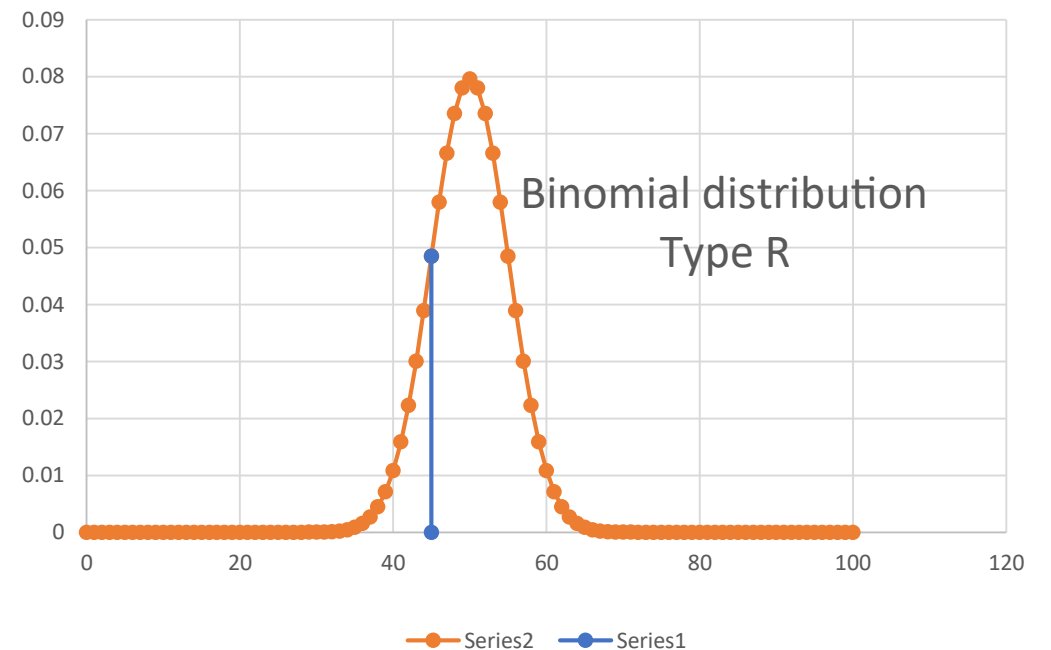
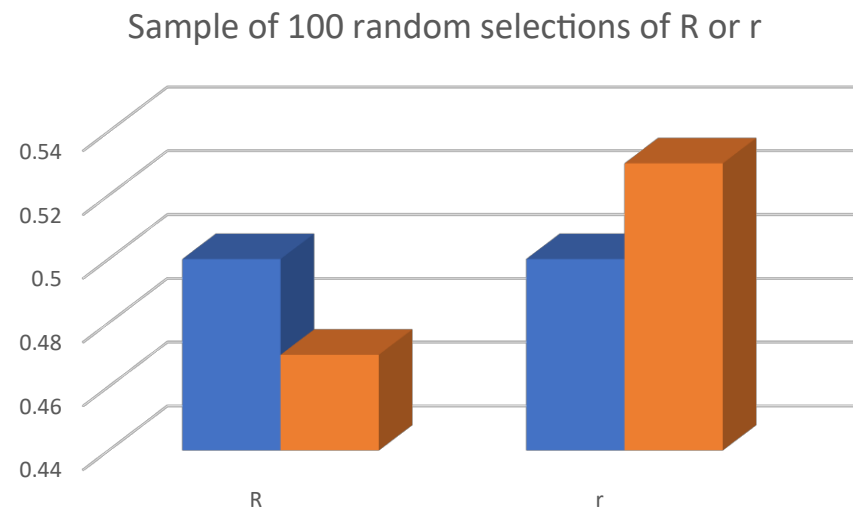
Εκθετική



Διωνυμική κατανομή

$$X_i = \begin{cases} 0 \\ 1 \end{cases}$$

$$P(X_i = Y) = \frac{N!}{Y!(N-Y)!} p^Y (1-p)^{N-Y}$$

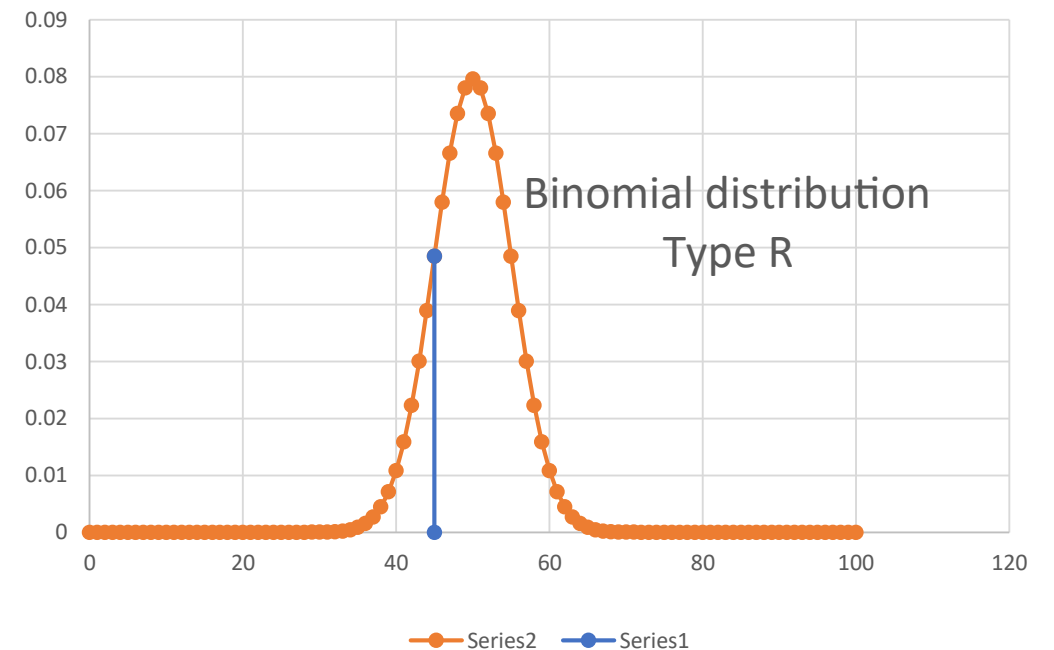


Διωνυμική κατανομή

$$\mu = \langle X \rangle = Np$$

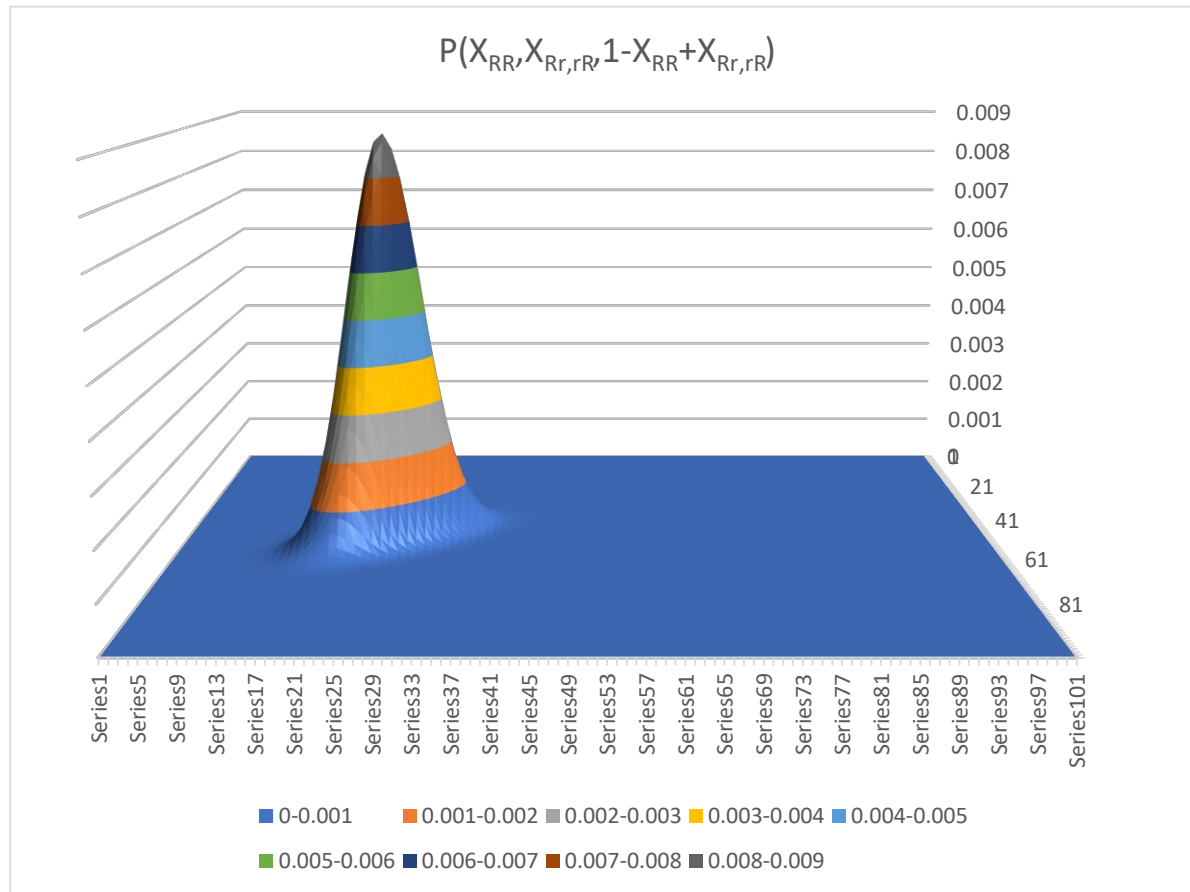
$$\sigma^2 = Np(1 - p)$$

$$\sigma = \sqrt{Np(1 - p)}$$



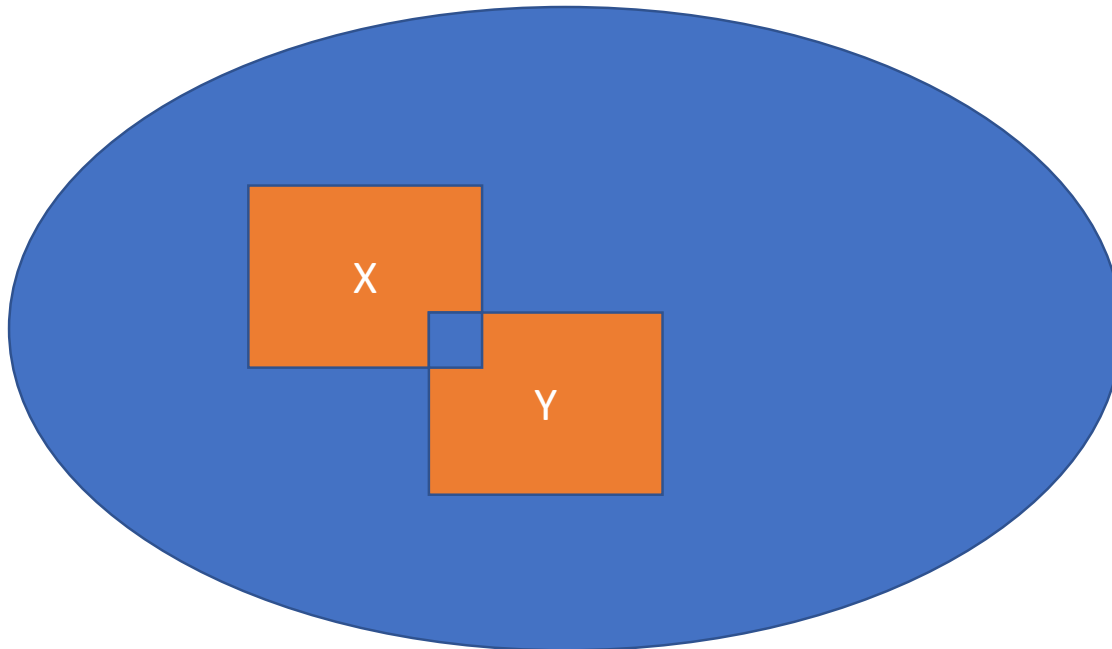
Multinomial Distribution

$$P(Y_1, \dots, Y_i, \dots, Y_k; p_1, p_i, p_k; N) = \frac{N!}{Y_1! Y_i! Y_k!} p_1^{Y_1} p_i^{Y_i} p_k^{Y_k}$$



Υπο συνθήκη πιθανότητα

$$P(X \cap Y) = P(X) P(Y|X)$$



Πληθυσμός - Δήγμα

Sample vs Population

Κεντρικό οριακό θεώρημα

Center Limit theorem

Δήγμα

Sample

$$\langle x \rangle = \frac{\sum_i^n x_i}{n}$$

$$s^2 = \frac{\sum_i^n (x_i - \langle x \rangle)^2}{n-1}$$

Πληθυσμός

Population

$$\mu = \frac{\sum_i^n x_i}{n}$$

$$\sigma^2 = \frac{\sum_i^n (x_i - \langle x \rangle)^2}{n}$$

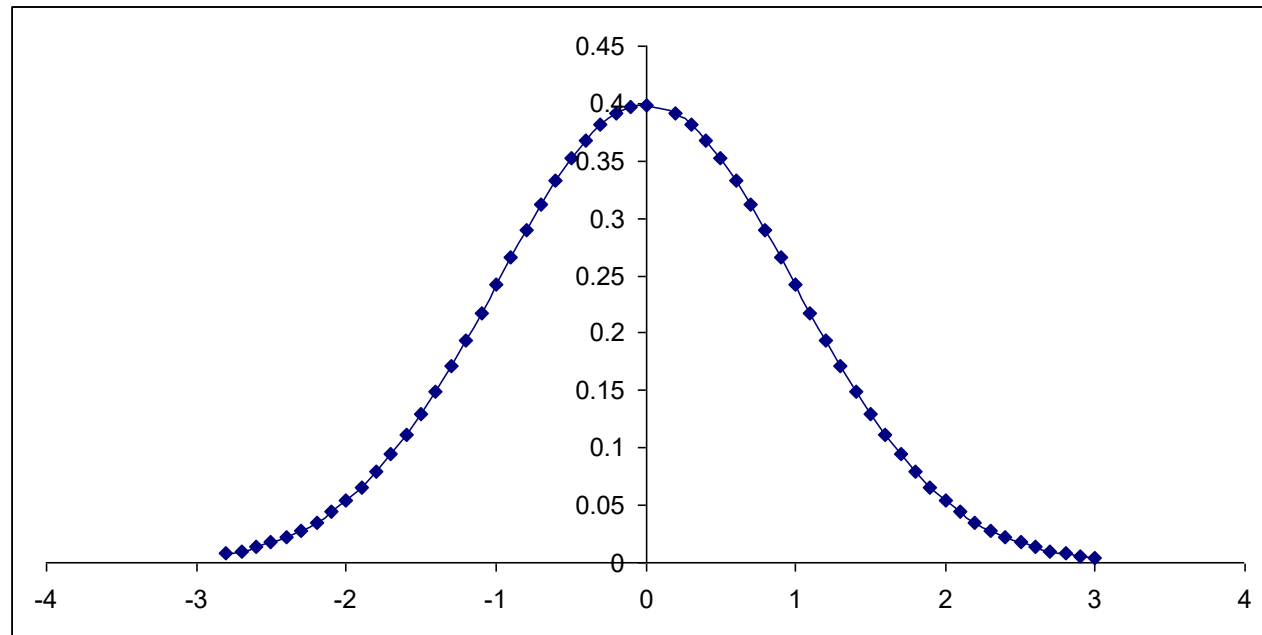
Population

Sample

$$\mu_{\langle x \rangle} = \mu$$

$$\sigma_{\langle x \rangle} = \frac{\sigma}{\sqrt{N}}$$

Κανονική κατανομή



$$f(x) = \frac{1}{\sqrt{2\pi\sigma^2}} e^{-\frac{(x-\mu)^2}{2\sigma^2}}$$

[Wikipedia](#)

Τέστ υποθέσεων H_0 Και H_1

H_0 : Μηδενική υπόθεση
null hypothesis

H_1 : Εναλλακτική υπόθεση
alternative hypothesis

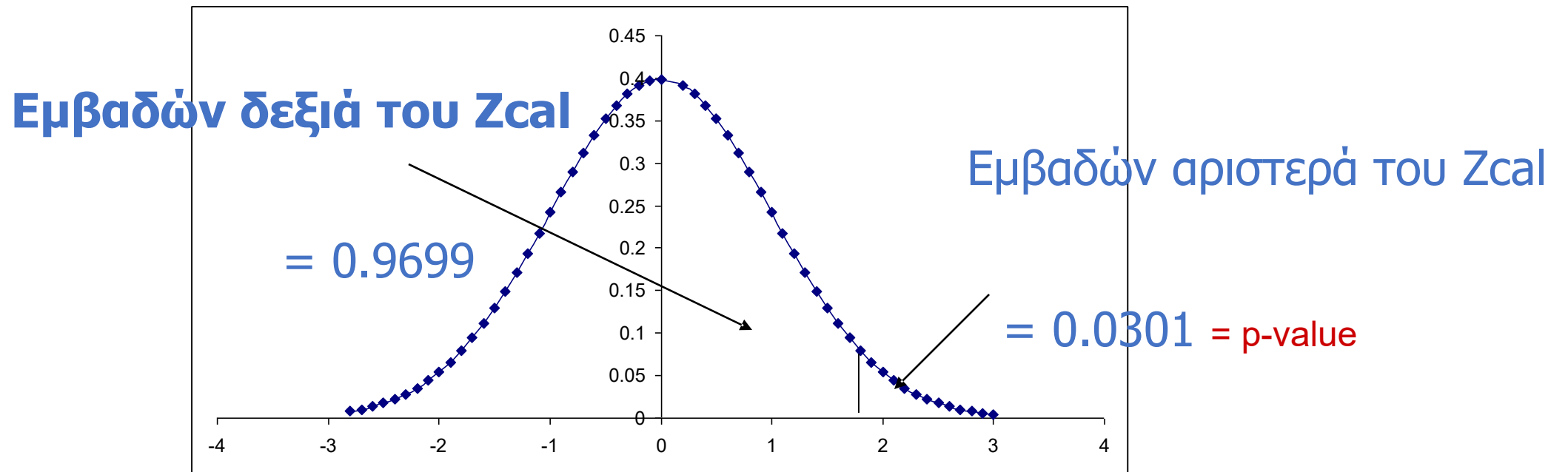
Επίπεδο σημαντικότητας Significance **Level**

[Wikipedia](#)

Balancing the two types of errors.

Κανονική κατανομή (Z τέστ)

Probability of z with a One-Tailed Test (Normal Distribution $\mu=0, \sigma=1$)



$$Z_{CAL} = 1.88$$

Determining the critical value of the test statistic, the area to the right of the critical value is either α or $\alpha/2$. It is α for a one-tail test and $\alpha/2$ for a two-tail test.

Κανονική κατανομή (Z τέστ)

Using a t-Test on sample

Sample

$$\langle x \rangle = \frac{\sum_i^n x_i}{n}$$

$$s^2 = \frac{\sum_i^n (x_i - \langle x \rangle)^2}{n-1}$$

df = n-1

Population

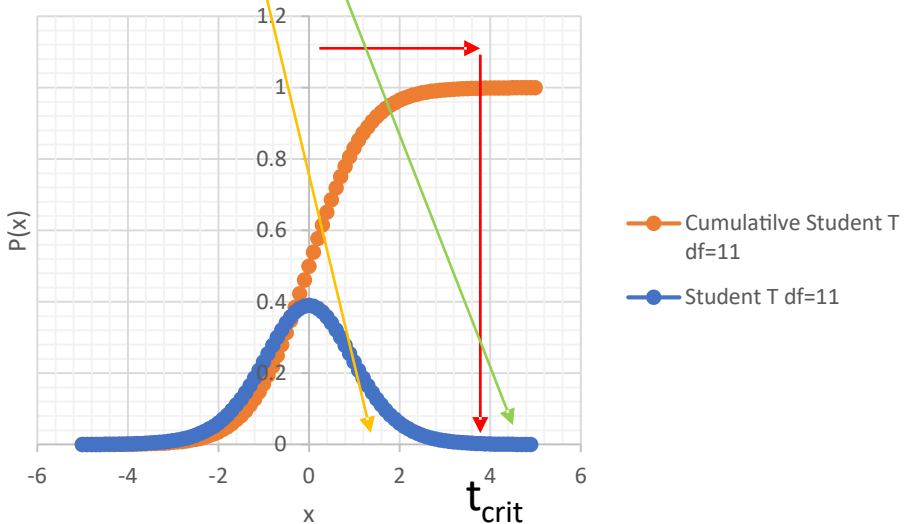
$$\mu = \frac{\sum_i^n x_i}{n}$$

$$\sigma^2 = \frac{\sum_i^n (x_i - \langle x \rangle)^2}{n}$$

$$t = \frac{(\langle x \rangle - \mu)}{\frac{s_x}{\sqrt{n}}}$$

accept

reject



$$\mu_{\langle x \rangle} = \mu$$

$$\sigma_{\langle x \rangle} = \frac{\sigma}{\sqrt{n}} \cong \frac{s}{\sqrt{n}}$$

Using a t-Test two samples (Independent)

Ίδιο μέγεθος δείγματος , αναμενόμενη ίση διακύμανση
equal size, expected equal σ

$$t = \frac{(\langle X_1 \rangle - \langle X_2 \rangle)}{s_p \sqrt{\frac{2}{n}}}$$
$$s_p = \sqrt{\frac{s_{X_1}^2 + s_{X_2}^2}{2}}$$
$$df = 2n - 2$$

Διαφορετικό μέγεθος δείγματος , αναμενόμενη ίση διακύμανση
Not equal size, expected equal σ

$$t = \frac{(\langle X_1 \rangle - \langle X_2 \rangle)}{s_p \sqrt{\frac{1}{n_1} + \frac{1}{n_2}}}$$
$$s_p = \sqrt{\frac{(n_1 - 1)s_{X_1}^2 + (n_2 - 1)s_{X_2}^2}{n_1 + n_2 - 2}}$$
$$df = 2n - 2$$

Διαφορετικό μέγεθος δείγματος , αναμενόμενη διαφορετική διακύμανση
Not equal size, expected NOT equal σ

$$t = \frac{(\langle X_1 \rangle - \langle X_2 \rangle)}{s_{\Delta}}$$
$$s_{\Delta} = \sqrt{\frac{s_{X_1}^2}{n_1} + \frac{s_{X_2}^2}{n_2}}$$
$$df = \frac{\left(\frac{s_{X_1}^2}{n_1} + \frac{s_{X_2}^2}{n_2}\right)^2}{\frac{\left(\frac{s_{X_1}^2}{n_1}\right)^2}{n_1 - 1} + \frac{\left(\frac{s_{X_2}^2}{n_2}\right)^2}{n_2 - 1}}$$

(t τέστ)

Using a t-Test two samples (dependent)

$$t = \frac{(\langle x \rangle - \mu)}{\frac{s_x}{\sqrt{n}}}$$

0 if we test if are $\mu_x=0$

$$x = X_1 - X_2$$

$$df=n-1$$

Paired Samples in the two groups

Κεντρικό οριακό θεώρημα

Center Limit theorem

Sample

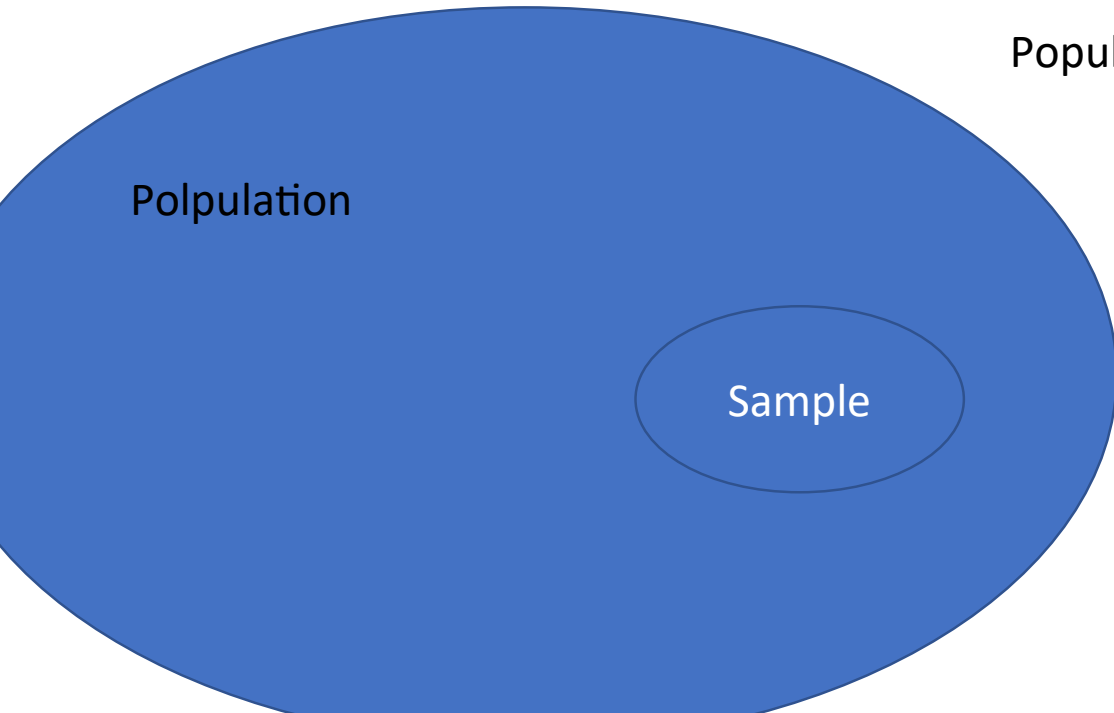
$$\langle x \rangle = \frac{\sum_{i=1}^n x_i}{n}$$

$$s^2 = \frac{\sum_{i=1}^n (x_i - \langle x \rangle)^2}{n - 1}$$

Population

$$\mu = \frac{\sum_{i=1}^N x_i}{N}$$

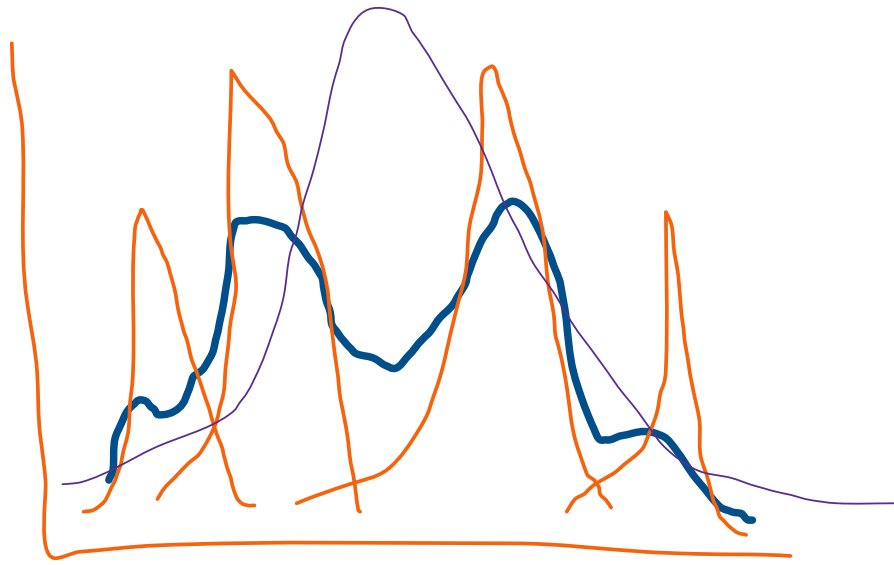
$$\sigma^2 = \frac{\sum_{i=1}^N (x_i - \mu)^2}{N}$$



$$\mu_{\langle x \rangle} = \mu$$

$$\sigma_{\langle x \rangle} = \frac{\sigma}{\sqrt{N}}$$

Anova



Nonparametric test

Γενετική ισορροπία

Hardy-Weinberg Disequilibrium

$$P_{aa} \neq p_a^2$$

$$P_{Aa} \neq 2p_A p_a$$

$$p_A + p_a = 1$$

$$D_{aa} = P_{aa} - p_a^2$$

$$D_{Aa} = P_{Aa} - 2p_A p_a$$

Is there a Correlation in Binomial distribution ?

$$P_{aa} = p_a^2 + p_a(p_A)f$$

$$P_{Aa} = 2p_A p_a(1 - f)$$

Let X_j , $j = 1, 2$ indicating whether the j th allele of a random individual is allele A or a.
Clearly, $E(X_j) = p_a$

$$\text{Var}(X_j) = p_i(1 - p_i)$$

$$\text{Cov}(X_i, X_j) = E(X_i X_j) - E(X_i)E(X_j) = P_{11} - p_1^2$$

$$\text{Corr}(X_i, X_j) = \text{Cov}(X_i, X_j) / (\sqrt{\text{Var}(X_i) \text{Var}(X_j)}) = f$$

Γενετική ισορροπία

Hardy-Weinberg Disequilibrium

$$D_{aa} = P_{aa} - p_a^2$$

$$D_{Aa} = P_{Aa} - 2p_A p_a$$

But D's are not independent :

$$p_a = P_{aa} + \frac{1}{2}P_{Aa} \quad \longrightarrow \quad p_a = P_{aa} + \frac{1}{2}(D_{Aa} + 2p_A p_a)$$
$$P_{Aa} = D_{Aa} + 2p_A p_a$$

$$\xrightarrow{D_{aa} + p_a^2 = P_{aa}} \quad p_a = D_{aa} + p_a^2 + \frac{1}{2}(D_{Aa} + 2p_A p_a)$$

$$p_a = D_{aa} + p_a^2 + \frac{D_{Aa}}{2} + p_A p_a$$

$$p_a = D_{aa} + \frac{D_{Aa}}{2} + p_a(p_a + p_A) \quad \longrightarrow \quad D_{aa} = -\frac{D_{Aa}}{2}$$

Γενετική ισορροπία

Hardy-Weinberg Disequilibrium

$$P_{Aa} = D_{Aa} + 2p_A p_a$$

$$P_{aa} = D_{aa} + p_a^2$$

$$P_{AA} = D_{AA} + p_A^2$$

$$D_{AA} = D_{aa} = -\frac{D_{Aa}}{2}$$

$$\max(-p_a^2, -p_A^2) \leq D_{AA} = D_{aa} \leq p_A p_a$$

Testing $D_{AA} = 0$ (HWE)

Γενετική ισορροπία

Hardy-Weinberg Disequilibrium

Testing $D = 0$ (HWE)

- An estimate D_{AA}
- A sampling distribution for the estimate D_{AA} .

$$N_{AA} = N(P_{AA}) = N(p_A^2 + D_{AA})$$
$$P_{aa} = D_{aa} + p_a^2$$

$$N_{Aa} = N(P_{Aa}) = N(2p_A p_a - 2D_{AA})$$
$$P_{Aa} = D_{Aa} + 2p_A p_a$$

Bailey's method:

$$\hat{p}_A = \frac{2N_{AA} + N_{Aa}}{2N}$$

$$\hat{D}_{AA} = \frac{N_{AA}}{N} - \hat{p}_A^2 = \hat{P}_{AA} - \hat{p}_A^2$$

Γενετική ισορροπία

Hardy-Weinberg Disequilibrium

Testing $D_{AA} = 0$ (HWE)

Approximating

$$E[\hat{D}_{AA}] \approx 0$$

$$\text{var}[\hat{D}_{AA}] \approx \frac{1}{N} (\hat{p}_a^2 \hat{p}_A^2)$$

$$z = \frac{\hat{D}_{AA} - E[\hat{D}_{AA}]}{\sqrt{\text{var}[\hat{D}_{AA}]}} \approx \frac{N\hat{D}_{AA}}{(\hat{p}_a \hat{p}_A)}$$

Γενετική ισορροπία

Hardy-Weinberg Disequilibrium

Likelihood for HWE

$$\hat{p}_{AA} = \frac{N_{AA}}{N} \quad \hat{p}_{Aa} = \frac{N_{Aa}}{N}$$

$$L = \left(\frac{N!}{N_{AA}! N_{Aa}! N_{aa}} \right) (\tilde{p}_{Aa})^{N_{Aa}} (\tilde{p}_{AA})^{N_{AA}} (\tilde{p}_{aa})^{N_{aa}}$$

Σχέση φαινοτύπου γονότυπου

Interface : form phenotype to genotype

Estimate allele frequencies in the ABO blood group system in humans

https://en.wikipedia.org/wiki/ABO_gene

Phenotype	A	AB	B	O
Genotype(s)	aa ao	ab	bb bo oo	
Sample	N_A	N_{AB}	N_B	N_O

Lecture Notes in Population Genetics : <http://darwin.eeb.uconn.edu/eeb348/lecture-notes/book.pdf>

Kent E. Holsinger Department of Ecology &
Evolutionary Biology, U-3043 University of
Connecticut Storrs, CT 06269-3043

Σχέση φαινοτύπου γονότυπο

Estimate allele frequencies in the ABO blood group system in humans

Phenotype	A	AB	B	O
Genotype(s)	aa ao	ab	bb bo oo	oo
Sample	N_A	N_{AB}	N_B	N_O

If the frequencies for the genotype where known :

$$N_{aa} = N_A \left(\frac{p_a^2}{p_a^2 + 2p_a p_o} \right)$$

$$N_{ao} = N_A \left(\frac{2p_a p_o}{p_a^2 + 2p_a p_o} \right)$$

$$N_{ab} = N_{AB}$$

$$N_{bb} = N_B \left(\frac{p_b^2}{p_b^2 + 2p_b p_o} \right)$$

$$N_{bo} = N_B \left(\frac{2p_b p_o}{p_b^2 + 2p_b p_o} \right)$$

$$N_o = N_o$$

Interface : form phenotype to genotype

Estimate allele frequencies in the ABO blood group system in humans

Phenotype	A	AB	B	O
Genotype(s)	aa ao	ab	bb bo	oo
Sample	N_A	N_{AB}	N_B	N_O

$$N_{aa} = N_A \left(\frac{p_a^2}{p_a^2 + 2p_a p_o} \right)$$

$$N_{ao} = N_A \left(\frac{2p_a p_o}{p_a^2 + 2p_a p_o} \right)$$

$$N_{ab} = N_{AB}$$

$$N_{bb} = N_B \left(\frac{p_b^2}{p_b^2 + 2p_b p_o} \right)$$

$$N_{bo} = N_B \left(\frac{2p_b p_o}{p_b^2 + 2p_b p_o} \right)$$

$$N_o = N_o$$

$$p_a = \left(\frac{2N_{aa} + N_{ao} + N_{ab}}{2N} \right)$$

$$p_b = \left(\frac{2N_{bb} + N_{bo} + N_{ab}}{2N} \right)$$

$$p_o = \left(\frac{N_{ao} + N_{bo} + 2N_{oo}}{2N} \right)$$

Phenotype	A	AB	B	O
Genotype(s)	aa ao	ab	bb bo	oo
Sample	NA	NAB	NB	NO
	12	44	33	22

Initial Estimates

pa	pb	po
----	----	----

0.333333 0.333333 0.333333
0.27027 0.396396 0.333333
0.267845 0.402275 0.32988
0.26786 0.403152 0.328988
0.267891 0.403323 0.328786
0.267899 0.403359 0.328742
0.267901 0.403367 0.328732
0.267901 0.403368 0.32873

Naa	Nao	Nbb	Nbo	Noo	Nab
-----	-----	-----	-----	-----	-----

4 8 11 22 22 44
3.4615388.538462 12.30508 20.69492 22 44
3.4649858.535015 12.49966 20.50034 22 44
3.4718058.528195 12.53761 20.46239 22 44
3.4736018.526399 12.54567 20.45433 22 44
3.4740098.525991 12.54742 20.45258 22 44
3.4740998.525901 12.5478 20.4522 22 44
3.4741188.525882 12.54789 20.45211 22 44

Resulting

pa	pb	po
----	----	----

0.27027 0.396396 0.333333
0.267845 0.402275 0.32988
0.26786 0.403152 0.328988
0.267891 0.403323 0.328786
0.267899 0.403359 0.328742
0.267901 0.403367 0.328732
0.267901 0.403368 0.32873

Prevalence

The number (or percentage) of people with a specific condition

Point Prevalence

The fraction of people with a specific condition at a given time

Incidence

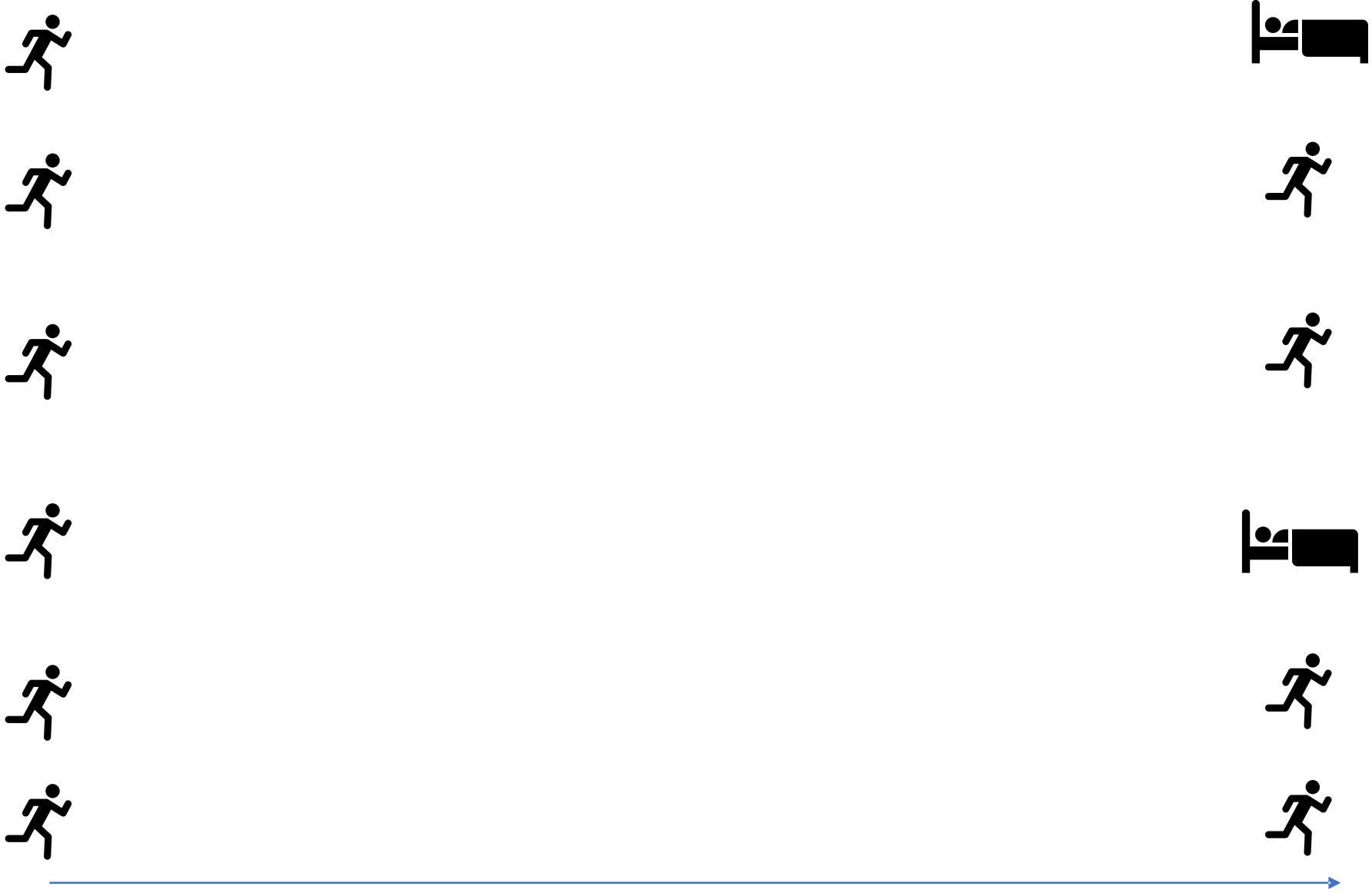
The Likelihood of developing the disease within a period of time

Cumulative Incidence : $\frac{\text{The number of people the of developing the disease within a period of time}}{\text{The number of people at risk}}$

Incidence rate : $\frac{\text{The number of people the of developing the disease within a period of time}}{\text{The sum of length of time that the persons are free of disease}}$

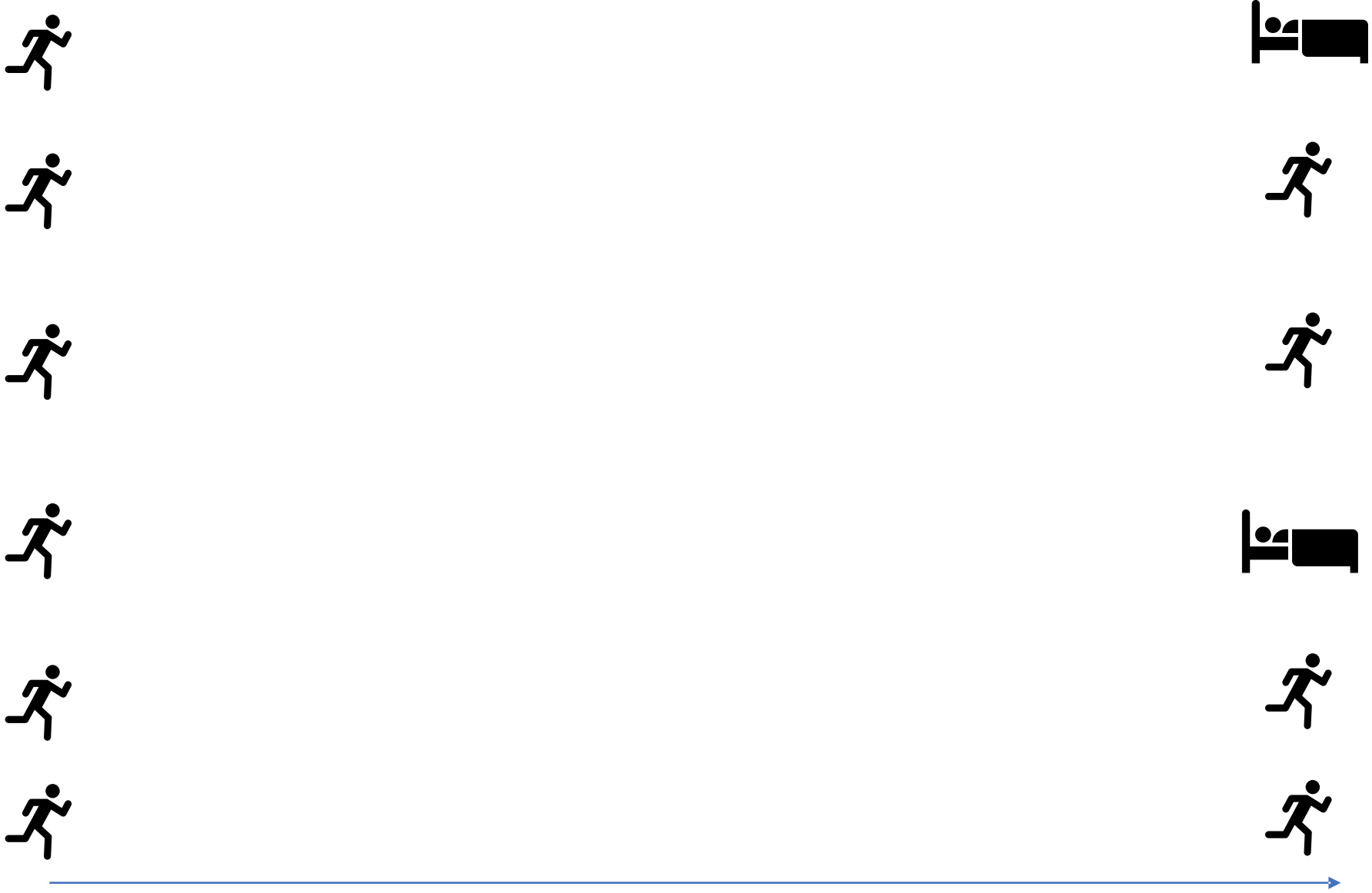
Cumulative Incidence : The number of people the of developing the disease within a period of time
The number of people at risk

=2/6



Incidence rate : The number of people the of developing the disease within a period of time
The sum of length of time that the persons are free of disease

$$=2/t$$

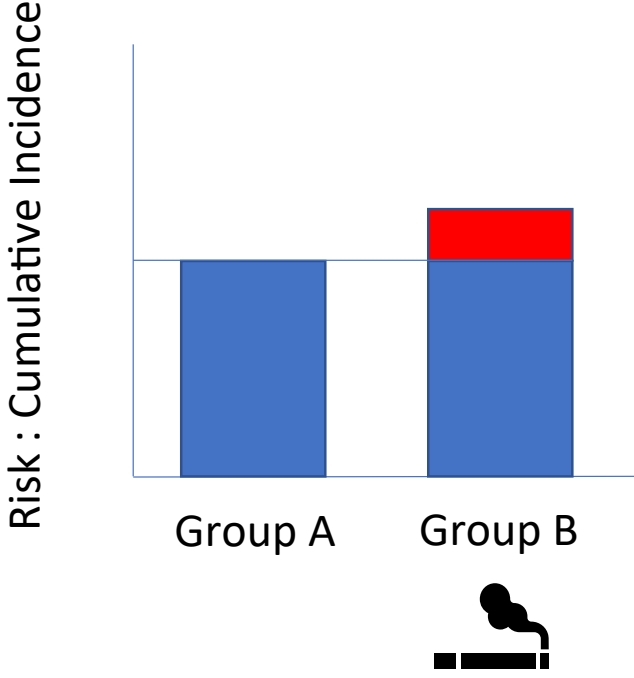


time

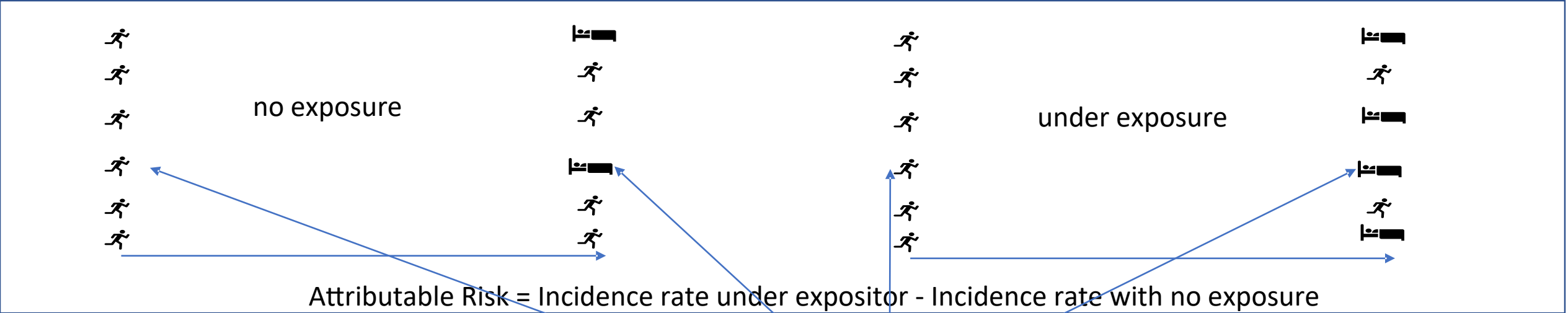
Compering Disease Risk between groups.

- Risk deference
- Relative Risk
- Odds Ratio

Risk deference
In different Groups



Risk deference under exposure



	Not exposure	exposure	Total
Incidents	5	20	25
No Incidents	123	66	189
Total	128	86	214
% Incidents	4%	23%	12%
	Attributable risk =		19%
	Risk Fraction =		83%
	Relative risk		5.95

Probability vs odds and odds Ratio

Probability $2/6$

Relative odds $2/4$

A

1

1

A

1

1

<http://uncyclopedia.wikia.com/wiki/File:Flipping-coin-animated.gif>

odds Ratio (exposed vs not exposed)

Not-exposed

A

2/4

1

1

A

1

1

exposed

A

2/3

1

1

A

1

$$\text{odds Ratio} = 2/3 / (2/4) = 4/3$$

Relative risk

	Not exposure	exposure	Total
Incidents	5	20	25
No Incidents	123	66	189
Total	128	86	214
% Incidents	4%	23%	12%
Attributable risk =		19%	
Risk Fraction =		83%	
Relative risk		5.95	

Odds Ratio

	Not exposure	exposure	Total
Disease	5	20	25
No Disease	123	66	189
Total	128	86	214
odds for Disease	0.040650407	0.30303	
odds ratio			7.454545

	Not exposure	exposure	
Incidents	5	20	25
No Incidents	123	66	189
	128	86	

3.90625 23.25581395

Attributable risk 19.34956395
 Relative risk 5.953488372
 odd ratio 7.454545455

Ανεξάρτητες μεταβλητές

$$P(Y|X) = P(Y)$$

$$P(X \cap Y) \neq P(X)P(Y|X)$$

Sensitivity Specificity of a test

- Y : positive test

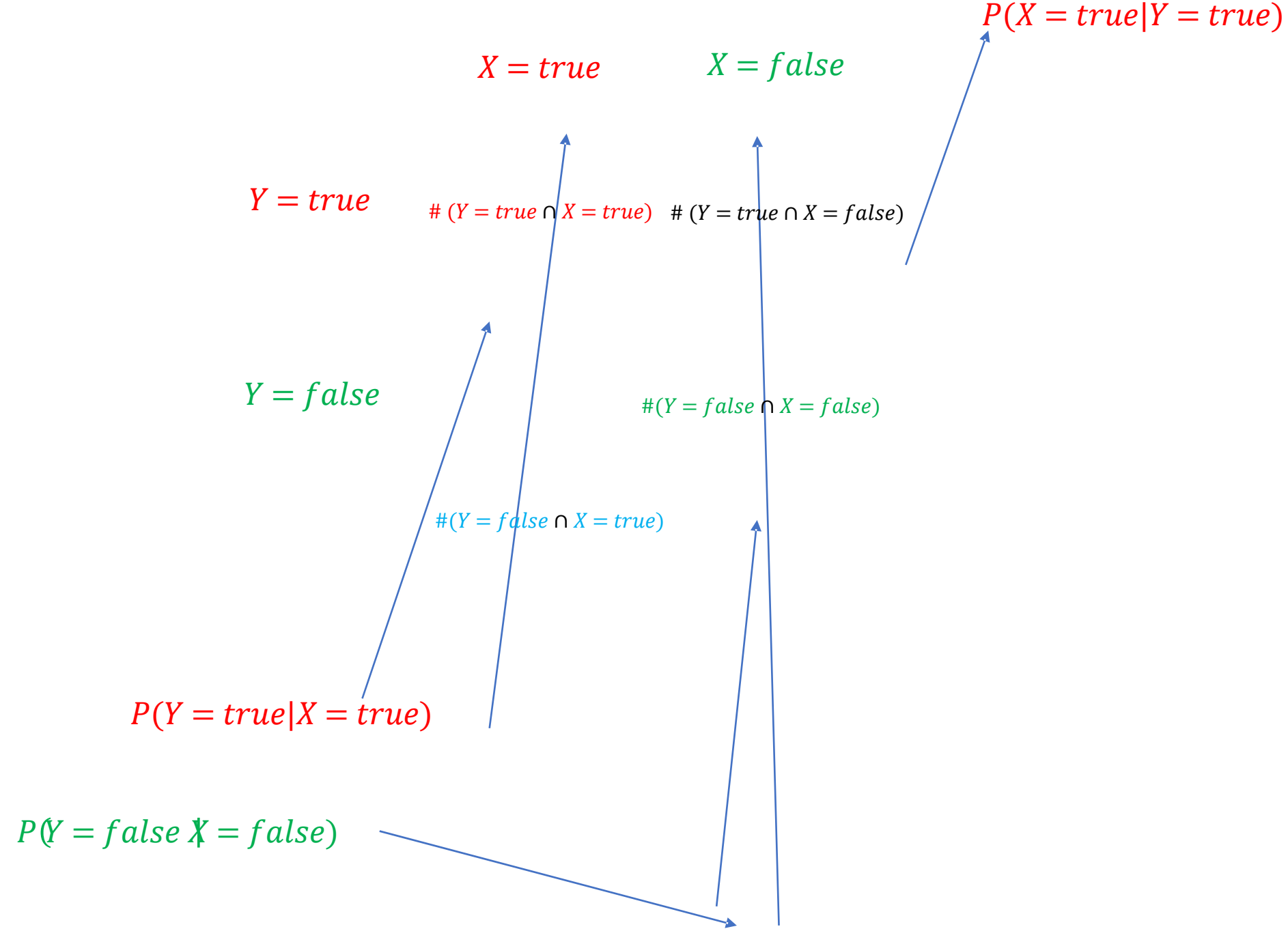
$$\text{Sensitivity } P(Y = \text{true} | X = \text{true}) = \frac{\#(Y = \text{true}) \cap (X = \text{true})}{\#(X = \text{true})}$$

- X : have the disease

$$\text{Specificity } P(Y = \text{false} | X = \text{false})$$

$$\text{False negative fraction : } P(Y = \text{false} | X = \text{true})$$

$$\text{False positive fraction : } P(Y = \text{true} | X = \text{false})$$



Bayes Theorem

$$P(X \cap Y) = P(X)P(Y|X) = P(Y)P(X|Y) \quad (1)$$

$$P(Y|X) = \frac{P(Y)}{P(X)} P(X|Y)$$