Basic Stochastics and the idea of testing

Dirk Metzler

http://evol.bio.lmu.de/_statgen

October 10, 2017

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However, we can ask, for example, for

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Even these values (especially the second on) depend on our model assumptions.

Contents

We start with a simpler Example: Rolling a dice, W is the result of the next trial.

$$\mathcal{S} = \{1, 2, \ldots, 6\}$$
$$\Pr(W = 1) = \cdots = \Pr(W = 6) = \tfrac{1}{6}$$
$$(\ \Pr(W = x) = \tfrac{1}{6} \text{ for all } x \in \{1, \ldots, 6\}\ )$$

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In general, we use capitals for
random variables ($X, Y, Z, \ldots$),
and small letters ($x, y, z, \ldots$) for (possible) fixed values.

Notations for events

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We can interpret this as the set of results (elementary events) for which the event is fulfilled.
Thus, events have a lot in common with sets, and similar notations as for sets are used for events U and V:

$$U \cap V = U \text{ "and" } V$$

is the event that takes place if and only if both U and V take place.

$$U \cup V = U \text{ "or" } V$$

is the event that takes place if and only if U or V (or both) take place.

Example

Let X and Y be the results of two dice rolls, $A = \{1, 2, 3\}$, and $B = \{1, 3, 5\}$. Then:

$$\begin{aligned} \{X \in A\} \cap \{X \in B\} &= \{X \in A \cap B\} = \{X \in \{1, 3\}\} \\ &= \{X = 1\} \cup \{X = 3\} \end{aligned}$$

and

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and

$$\{X \in A\} \cap \{Y \in B\} = \{(X, Y) \in A \times B\}, \text{where}$$

$$A \times B = \{(1, 1), (1, 3), (1, 5), (2, 1), (2, 3), (2, 5), (3, 1), (3, 3), (3, 5)\}.$$

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is then the event that X takes a value that is in A and in B.

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Sometimes the curly brackets are not written:

$$\Pr(X \in A, X \in B) = \Pr(\{X \in A, X \in B\})$$

Calculation rules:

Example Rolling a dice W:

$$\begin{aligned}\Pr(W \in \{2,3\}) &= \frac{2}{6} = \frac{1}{6} + \frac{1}{6} \\ &= \Pr(W = 2) + \Pr(W = 3) \\ \Pr(W \in \{1,2\} \cup \{3,4\}) &= \frac{4}{6} = \frac{2}{6} + \frac{2}{6} \\ &= \Pr(W \in \{1,2\}) + \Pr(W \in \{3,4\})\end{aligned}$$

Caution:

$$\begin{aligned}\Pr(W \in \{2,3\}) + \Pr(W \in \{3,4\}) &= \frac{2}{6} + \frac{2}{6} = \frac{4}{6} \\ &\neq \Pr(W \in \{2,3,4\}) = \frac{3}{6}\end{aligned}$$

Example: rolling two dice (W_1, W_2):
Let W_1 and W_2 the result of dice 1 and dice 2.

$$\begin{aligned}
&\Pr(W_1 \in \{4\}, W_2 \in \{2,3,4\}) \\
&= \Pr((W_1, W_2) \in \{(4,2),(4,3),(4,4)\}) \\
&= \frac{3}{36} = \frac{1}{6} \cdot \frac{3}{6} \\
&= \Pr(W_1 \in \{4\}) \cdot \Pr(W_2 \in \{2,3,4\})
\end{aligned}$$

In general:

$$\Pr(W_1 \in A, W_2 \in B) = \Pr(W_1 \in A) \cdot \Pr(W_2 \in B)$$

for all sets $A, B \subseteq \{1, 2, \dots, 6\}$

If S is the sum of the results $S = W_1 + W_2$,
what is the probability that $S = 5$,
if dice 1 shows $W_1 = 2$?

$$\Pr(S = 5 | W_1 = 2) \stackrel{!}{=} \Pr(W_2 = 3)$$
$$= \frac{1}{6} = \frac{1/36}{1/6} = \frac{\Pr(S{=}5, W_1{=}2)}{\Pr(W_1{=}2)}$$

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What is the probability $S \in \{4, 5\}$ under the condition $W_1 \in \{1, 6\}$?

$$\begin{aligned}
&\Pr(S \in \{4,5\} | W_1 \in \{1,6\}) \\
&= \frac{\Pr(S \in \{4,5\}, W_1 \in \{1,6\})}{\Pr(W_1 \in \{1,6\})} \\
&= \frac{\Pr(W_2 \in \{3,4\}, W_1 = 1)}{\Pr(W_1 \in \{1,6\})} \\
&= \frac{2/36}{2/6} = \frac{1}{6}
\end{aligned}$$

Calculation rules:

- $0 \leq \Pr(U) \leq 1$ for each event U (in the probability space).
- $\Pr(X \in \mathcal{S}) = 1$ if X is a random variable with state space $\mathcal{S}$.
- If the events U and V exclude each other, then

$$\Pr(U \cup V) = \Pr(U) + \Pr(V)$$

- The general rule is the inclusion-exclusion formula

$$\Pr(U \cup V) = \Pr(U) + \Pr(V) - \Pr(U \cap V)$$

- Definition of conditional probabilities:
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Note: $\Pr(U, V) = \Pr(V) \cdot \Pr(U| V)$

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The probability of $\{X \in A,\ Y \in B\}$ can be computed in two steps:

- First, the event $\{X \in A\}$ must take place.
- Multiply its probability with the conditional probability of $\{Y \in B\}$, given that $\{X \in A\}$ is already known to take place.

Stochastic Independence of events

Definition (stochastic independence)

Two events U and V are (stochastically) independent, if the identity

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Note that $\Pr(U, V) = \Pr(U) \cdot \Pr(V)$ is equivalent to

$$\Pr(U|V) = \Pr(U) \text{ and also to } \Pr(V|U) = \Pr(V)$$

Stochastic Independence of random variables

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Example:

- Tossing two dice:
  $X =$ result dice 1, $Y =$ result dice 2.

$$\Pr(X = 2,\ Y = 5) = \frac{1}{36} = \frac{1}{6} \cdot \frac{1}{6} = \Pr(X = 2) \cdot \Pr(Y = 5)$$

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Bernoulli distribution

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Bernoulli random variable X:

State space $\mathcal{S} = \{0, 1\}$.
Distribution:

$$\Pr(X = 1) = p$$
$$\Pr(X = 0) = 1 - p$$

The parameter $p \in [0, 1]$ is the success probability.

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Examples:

- Tossing a coin: Possible outcomes are “head” and “tail”
- Does the Drosophila have a mutation that causes white eyes? Possible outcomes are “yes” or “no”.
- The sex of a newborn child has the values “male” or “female”.

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$$\binom{n}{k} \cdot p^k \cdot (1 - p)^{n-k}$$

Note

$\binom{n}{k} = \frac{n!}{k!\cdot(n-k)!}$ ("*n* choose *k*") is the number of possibilities to choose *k* successes in *n* trials.

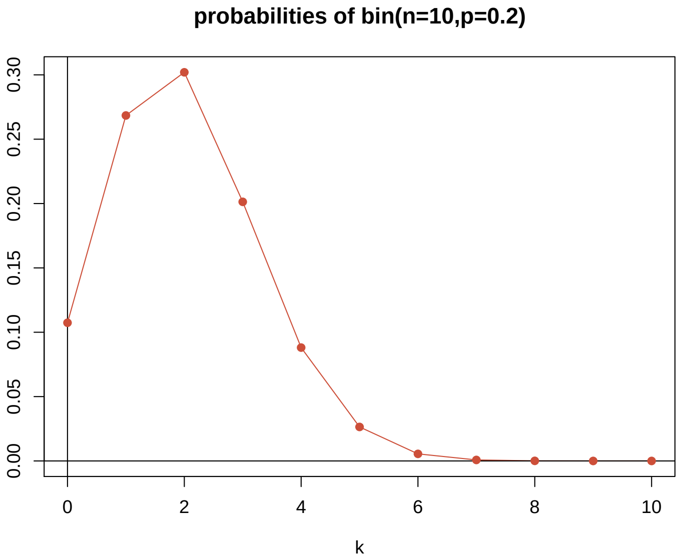
Binomial distribution

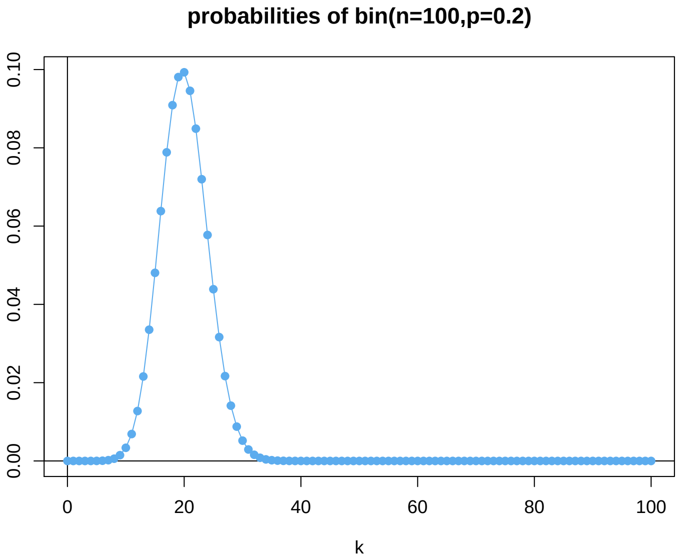
Let X be the number of successes in n independent trials with success probability of p each. Then,

$$\Pr(X = k) = \binom{n}{k} p^k \cdot (1 - p)^{n-k}$$

holds for all $k \in \{0, 1, \ldots, n\}$ and X is said to be *binomially distributed*, for short:

$$X \sim \text{bin}(n, p).$$


probabilities of bin(n=10,p=0.2)
0.00
0.05
0.10
0.15
0.20
0.25
0.30
0
2
4
6
8
10
k


probabilities of bin(n=100,p=0.2)
0.00
0.02
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20
40
60
80
100
k

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We can only answer this on the basis of a probabilistic model, and the answer will depend on how we model the population.

Modeling approach

We make a few simplifying assumptions:

- Discrete generations
- The population is haploid, that is, each individual has exactly one parent in the generation before.
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- Each individual chooses its parent purely randomly in the generation before.

“purely randomly” means *independent of all others* and *all potential parents with the same probability*.

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$$= \binom{100}{32} \cdot 0.3^{32} \cdot 0.7^{68} \approx 0.078$$

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- If we can do this, we reject H_0.
- How we measure deviation, must be clear *before* we see the data.

Statistical Testing: Imporatant terms

null hypothesis H_0 : says that what we want to substantiate is not true and anything that looks like evidence in the data is just random. We try to reject H_0.

significance level α : If H_0 is true, the probability to falsly reject it, must be $\leq \alpha$ (often $\alpha = 0.05$).

test statistic : measures how far the data deviates from what H_0 predicts into the direction of our alternative hypothesis.

p value : Probability that, if H_0 is true, a dataset leads to a test statistic value that is as least as extreme as the observed one.

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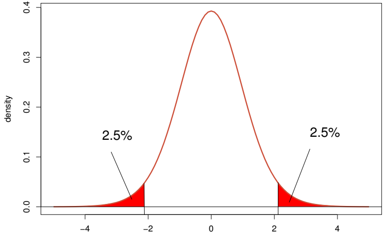
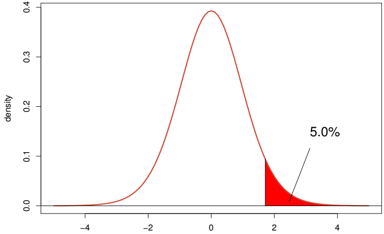
- Thus, if H_0 is true, the probability to (falsely) reject it is α (not the p value).
- This entails that a researcher who performs many tests with $\alpha = 0.05$ on complete random data (i.e. where H_0 is always true), will falsely reject H_0 in 5% of the tests.
- Therefore it is a severe violation of academic soundness to perform tests until one shows significance, and to publish only the latter.

Testing two-sided or one-sided?

We observe a value of x that is much larger than the H_0 expectation value μ.



p-value=$\Pr_{H_0}(|X - \mu| \geq |x - \mu|)$



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- AND AFTER THAT: Look at the data and check if if $\mathcal{A}$ occurs.
- Then, the probability that H_0 is rejected in the case that H_0 is actually true ("Type I error") is just α.

Violations against the pure teachings

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is as bad as:

"At first glance I saw that $\overline{x}$ is larger than μ_{H_0}. So, I immediately applied the one-sided test."

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The decision between one-sided and two-sided must not depend on the concrete data that are used in the test.

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More generally: If $\mathcal{A}$ is the event that will lead to the rejection of H_0, (if it occurs) then $\mathcal{A}$ must be defined without being influenced by the data that is used for testing.

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In some other fields the practical approach is more common: Just inform the reader about the p-values of different null-hypotheses. Let the reader decide which null-hypothesis would have been the most natural one.

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It is also common to write μ_X instead of $\mathbb{E}X$.

If we replace probabilities by relative frequencies in this definition, we get the formula for the mean value (of a sample).

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Calculating with expectatins

Theorem (Linearity of Expectation)

If X and Y are random variables with values in $\mathbb{R}$ and if $a \in \mathbb{R}$, we get:

- $\mathbb{E}(a \cdot X) = a \cdot \mathbb{E}X$
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But in general $\mathbb{E}(X \cdot Y) \neq \mathbb{E}X \cdot \mathbb{E}Y$. Example:

$$\mathbb{E}(W \cdot W) = \frac{91}{6} = 15.167 > 12.25 = 3.5 \cdot 3.5 = \mathbb{E}W \cdot \mathbb{E}W$$

Proof of Linearity: If $\mathcal{S}$ is the state space of X and Y, and if $a, b \in \mathbb{R}$, we obtain:

$$\begin{aligned}
&\mathbb{E}(a \cdot X + b \cdot Y) \\
&= \sum_{x\in\mathcal{S}} \sum_{y\in\mathcal{S}} (a \cdot x + b \cdot y) \Pr(X = x, Y = y) \\
&= a \cdot \sum_{x\in\mathcal{S}} \sum_{y\in\mathcal{S}} x \Pr(X = x, Y = y) + b \cdot \sum_{x\in\mathcal{S}} \sum_{y\in\mathcal{S}} y \Pr(X = x, Y = y) \\
&= a \cdot \sum_{x\in\mathcal{S}} x \sum_{y\in\mathcal{S}} \Pr(X = x, Y = y) + b \cdot \sum_{y\in\mathcal{S}} y \sum_{x\in\mathcal{S}} \Pr(X = x, Y = y) \\
&= a \cdot \sum_{x\in\mathcal{S}} x \Pr(X = x) + b \cdot \sum_{y\in\mathcal{S}} y \Pr(Y = y) \\
&= a \cdot \mathbb{E}(X) + b \cdot \mathbb{E}(Y)
\end{aligned}$$

Proof of the product formula: Let $\mathcal{S}$ be the state space of X and Y, and let X and Y be (stochastically) independent.

$$\begin{aligned}
&\mathbb{E}(X \cdot Y) \\
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&= \sum_{x \in \mathcal{S}} \sum_{y \in \mathcal{S}} (x \cdot y) \Pr(X = x) \Pr(Y = y) \\
&= \sum_{x \in \mathcal{S}} x \Pr(X = x) \cdot \sum_{y \in \mathcal{S}} y \Pr(Y = y) \\
&= \mathbb{E}X \cdot \mathbb{E}Y.
\end{aligned}$$

Theorem

If X is random variable with finite state space $\mathcal{S} \subset \mathbb{R}$, and if $f : \mathbb{R} \to \mathbb{R}$ is a function, we obtain

$$\mathbb{E}(f(X)) = \sum_{x \in \mathcal{S}} f(x) \cdot \Pr(X = x)$$

Exercise: proof this.

Expectation of the binomial distribution

Let $Y_1, Y_2, \ldots, Y_n$ be the indicator variables of the n independent trials, that is

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Note:

$$X \sim \text{bin}(n, p) \Rightarrow \mathbb{E}X = n \cdot p$$

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If X and Y are independent, they are also uncorrelated, that is $\mathrm{Cor}(X, Y) = 0$.

Example: rolling dice

Variance of result from rolling a dice W:

$$\begin{aligned}
\text{Var}(W) &= \mathbb{E}\big[\big(W - \mathbb{E}W\big)^2\big] \\
&= \mathbb{E}\big[\big(W - 3.5\big)^2\big] \\
&= (1 - 3.5)^2 \cdot \frac{1}{6} + (2 - 3.5)^2 \cdot \frac{1}{6} + \ldots + (6 - 3.5)^2 \cdot \frac{1}{6} \\
&= \frac{17.5}{6} \\
&= 2.91667
\end{aligned}$$

Example: Empirical Distribution

If $x_1, \ldots, x_n \in \mathbb{R}$ are data and if X is the result of a random draw from the data (such that $\Pr(X = x_i) = \frac{1}{n}$), we get:

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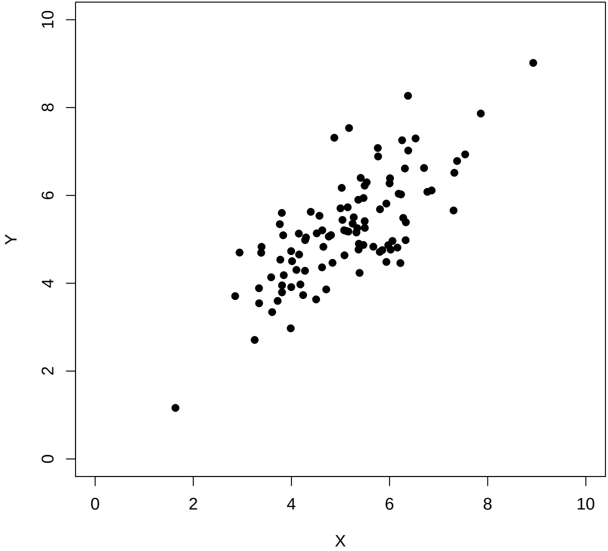
If $(x_1, y_1), \dots, (x_n, y_n) \in \mathbb{R} \times \mathbb{R}$ are data if (X, Y) are drawn from the data such that $\Pr((X, Y) = (x_i, y_i)) = \frac{1}{n}$, we get

$$\text{Cov }(X, Y) = \mathbb{E}\left[\left(X - \mathbb{E}X\right)\left(Y - \mathbb{E}Y\right)\right] = \frac{1}{n} \sum_{i=1}^{n} (x_i - \overline{x})(y_i - \overline{y})$$

Why $\text{Cov}(X, Y) = \mathbb{E}([X - \mathbb{E}X][Y - \mathbb{E}Y])$?



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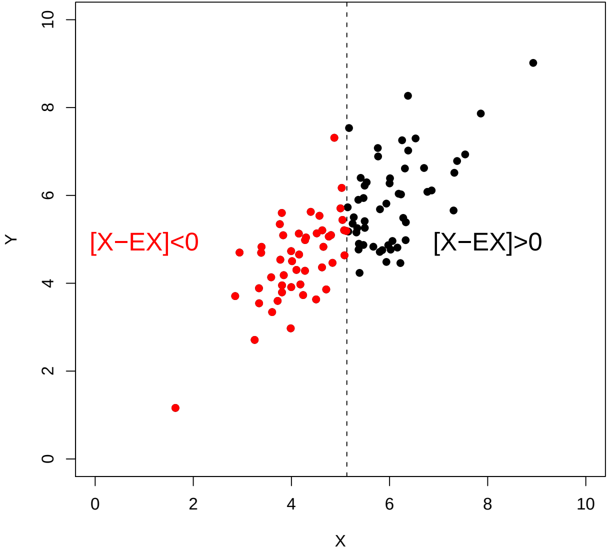

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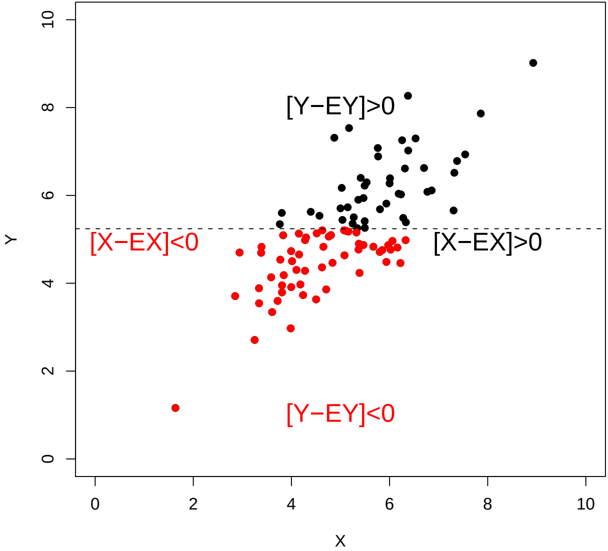

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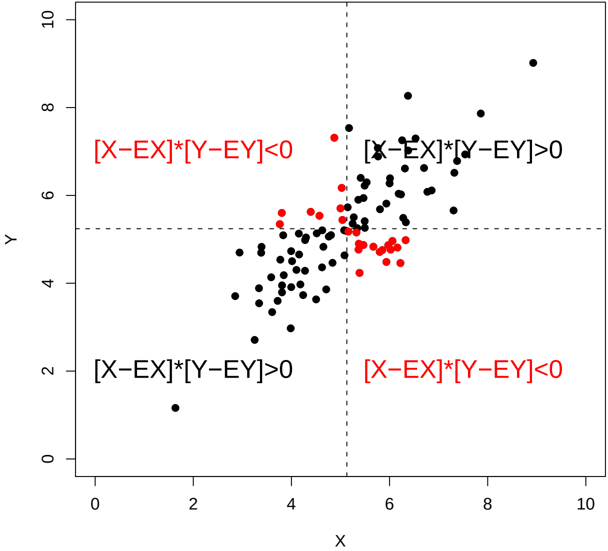

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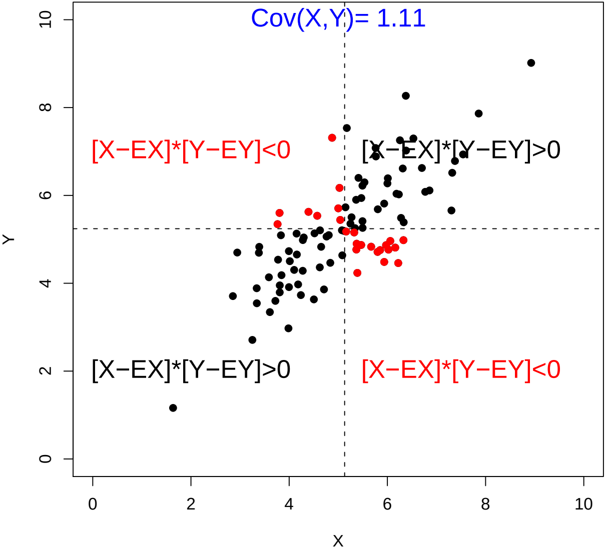

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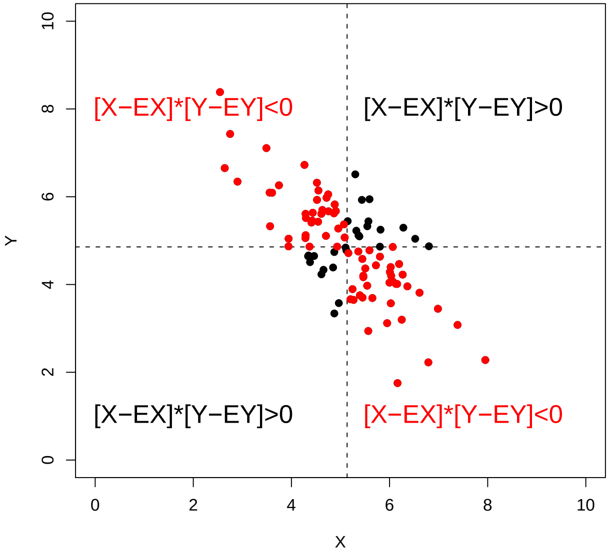

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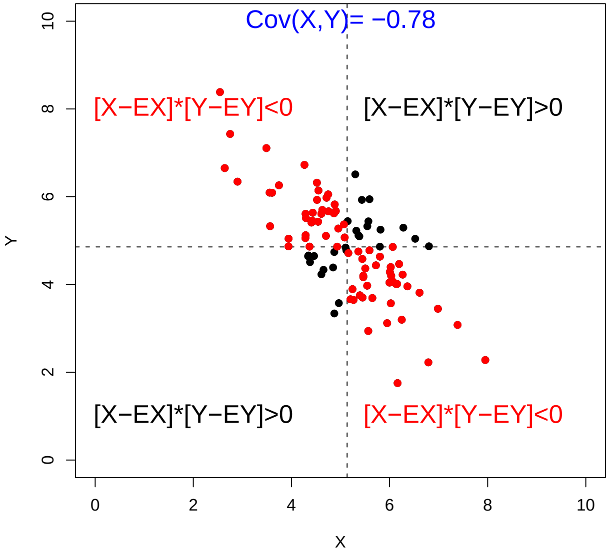
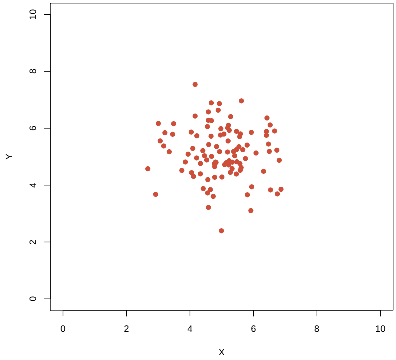
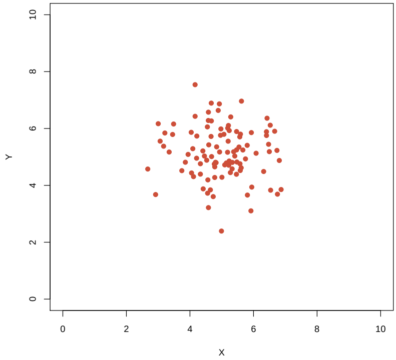


$\sigma_X = 0.95$, $\sigma_Y = 0.92$



$\sigma_X = 0.95$, $\sigma_Y = 0.92$

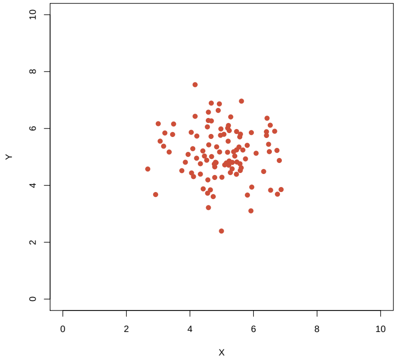
$\text{Cov}(X, Y) = -0.06$



$\sigma_X = 0.95$, $\sigma_Y = 0.92$

$\text{Cov}(X, Y) = -0.06$

$\text{Cor}(X, Y) = -0.069$



$\sigma_X = 0.95$, $\sigma_Y = 0.92$

$\text{Cov}(X, Y) = -0.06$

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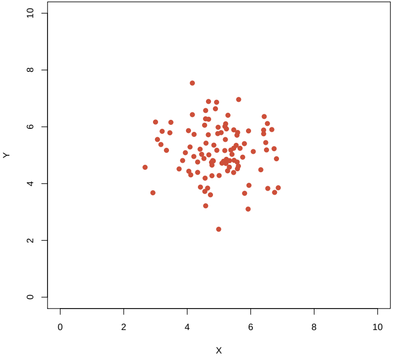

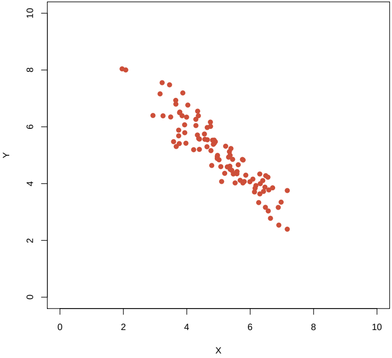

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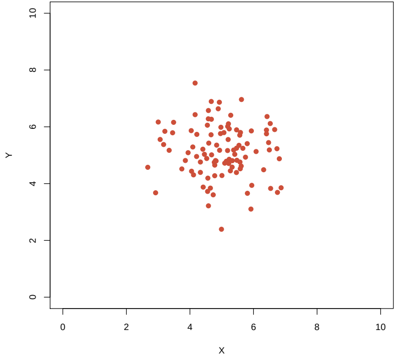
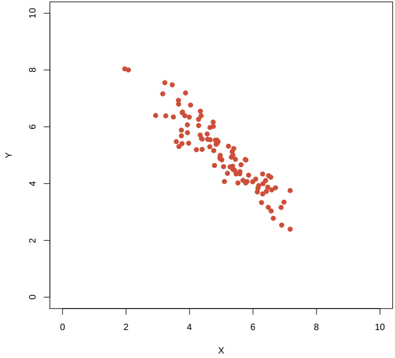
$\sigma_X = 1.13$, $\sigma_Y = 1.2$

$\text{Cov}(X, Y) = -1.26$





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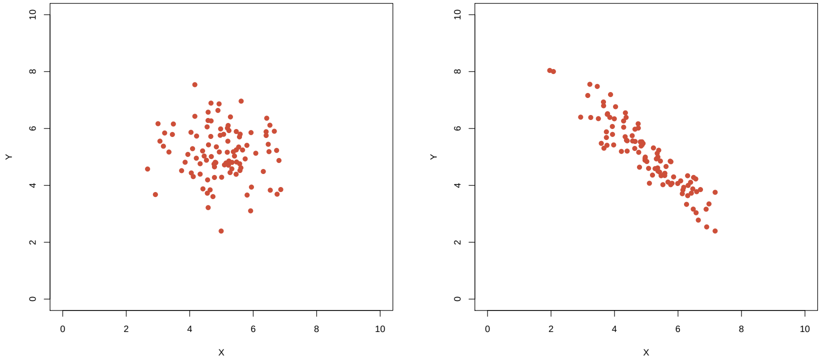

$\sigma_X = 1.14$, $\sigma_Y = 0.78$

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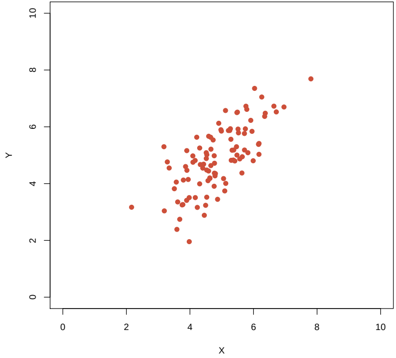
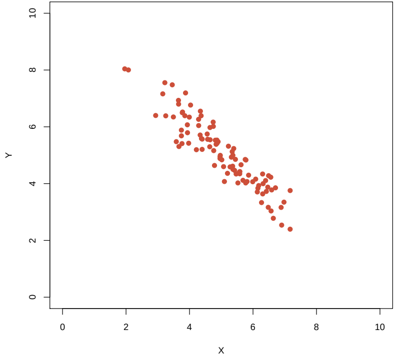
$\text{Cor}(X, Y) = -0.92$





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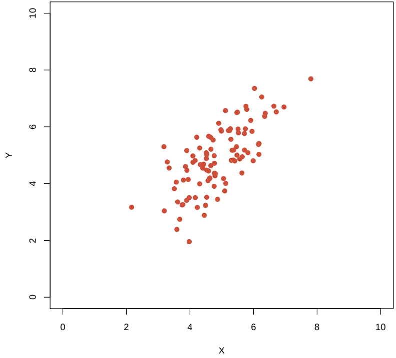

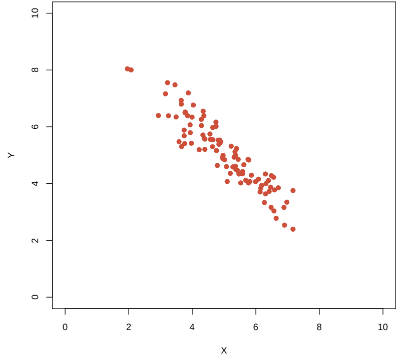

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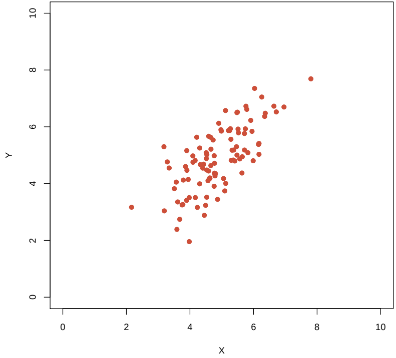

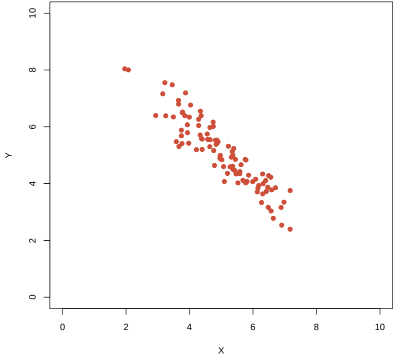

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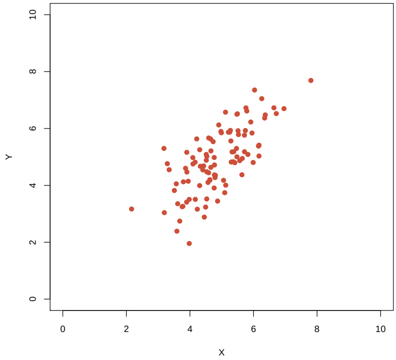
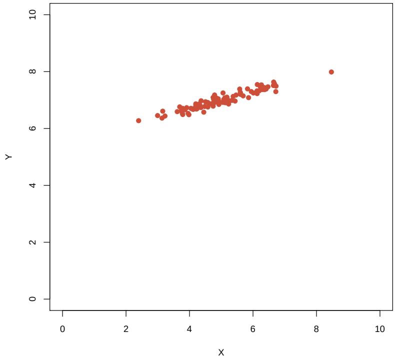
$\sigma_X = 1.03$, $\sigma_Y = 0.32$





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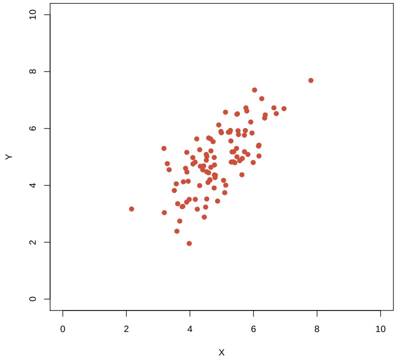
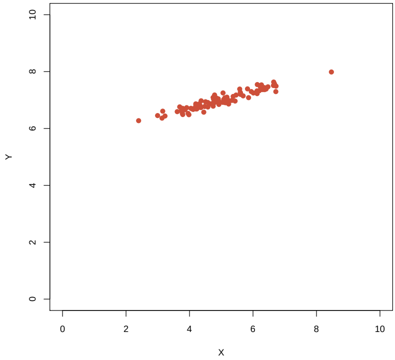


$\sigma_X = 1.03$, $\sigma_Y = 0.32$

$\text{Cov}(X, Y) = 0.32$



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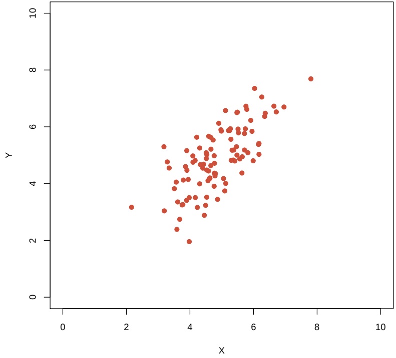

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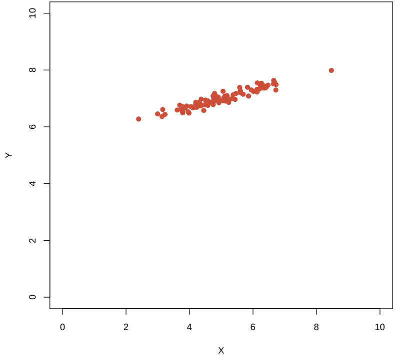

$\sigma_X = 0.91$, $\sigma_Y = 0.88$

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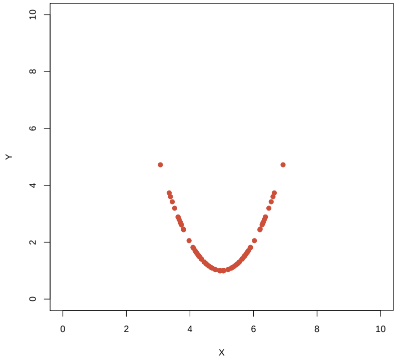
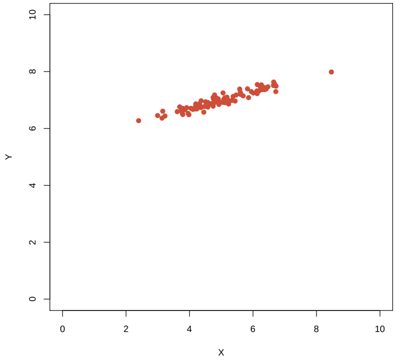
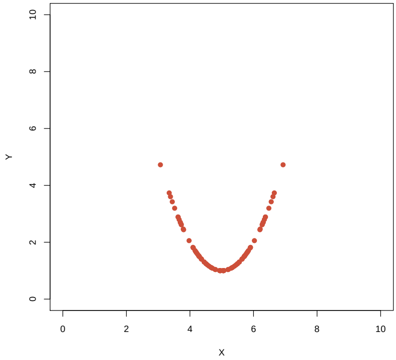




$\sigma_X = 0.91$, $\sigma_Y = 0.88$

$\text{Cov}(X, Y) = 0$



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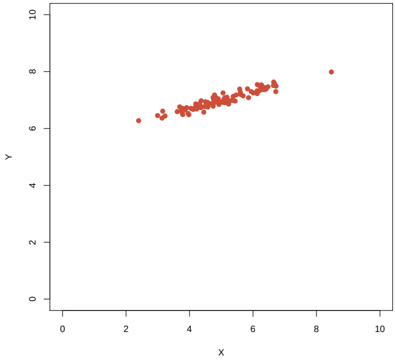

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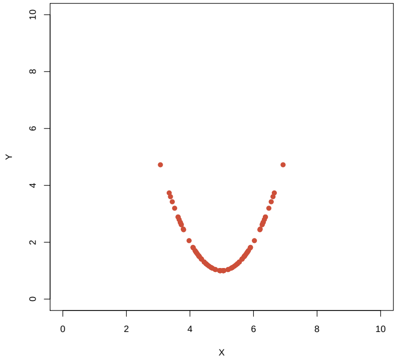

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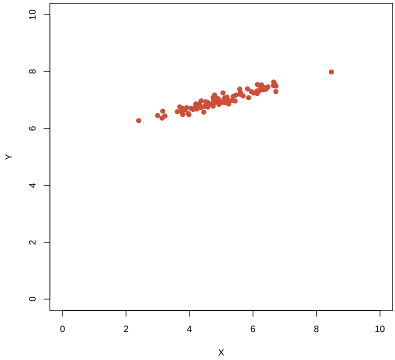

Calculation rules for variances

$$\text{Var}X = \mathbb{E}[(X - \mathbb{E}X)^2]$$

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- If (X, Y) stochastically independent we get:

$$\text{Var}(X + Y) = \text{Var}X + \text{Var}Y$$

Calculation rules for Covariances

$$\text{Cov}(X, Y) = \mathbb{E}[(X - EX) \cdot (Y - \mathbb{E}Y)]$$

- If X and Y are independent, then $\text{Cov}(X, Y) = 0$ (but not the other way around!)

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The last three rules describe the bilinearity of covariance.

Calculation rules for Correlations

$$\text{Cor}(X, Y) = \frac{\text{Cov}(X,Y)}{\sigma_X \cdot \sigma_Y}$$

- $-1 \leq \text{Cor}(X, Y) \leq 1$
- $\text{Cor}(X, Y) = \text{Cor}(Y, X)$
- $\text{Cor}(X, Y) = \text{Cov}(X/\sigma_X, Y/\sigma_Y)$

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- $\mathrm{Cor}(X, Y) = -1$ if and only if Y is an decreasing, affine-linear function of X, that is, if $Y = a \cdot X + b$ for appropriate $a < 0$ and $b \in \mathbb{R}$.

Bernoulli distribution

A Bernoulli distributed random variable Y with success probability $p \in [0, 1]$ has expectation value

$$\mathbb{E}Y = p$$

and variance

$$\text{Var } Y = p \cdot (1 - p)$$

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Proof: From $\Pr(Y = 1) = p$ and $\Pr(Y = 0) = (1-p)$ follows

$$\mathbb{E}Y = 1 \cdot p + 0 \cdot (1-p) = p.$$

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variance:

$$\begin{aligned} \text{Var } Y &= \mathbb{E}\big(Y^2\big) - \big(\mathbb{E}Y\big)^2 \\ &= 1^2 \cdot p + 0^2 \cdot (1 - p) - p^2 = p \cdot (1 - p) \end{aligned}$$

Binomial distribution

Let $Y_1, \cdots, Y_n$ be independent Bernoulli distributed with success probability p. Then follows

$$\sum_{i=1}^{n} Y_i =: X \sim \text{bin}(n, p)$$

and we get:

$$\text{Var}\, X =$$

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Binomial distribution

Theorem (Expectation value and variance of the binomial distribution)

If X is binomially distributed with parameters (n, p), we get:

$$\mathbb{E}X = n \cdot p$$

und

$$Var\,X = n \cdot p \cdot (1 - p)$$

Example: Genetic Drift

In a haploid population of n individuals, let p be the frequency of some allele A. We assume that (due to some simplifying assumptions?) the absolute frequency K of A in the next generation is (n, p)-binomially distributed.
For $X = K/n$, the relative frequency in the next generation follows:

$$\mathrm{Var}(X) = \mathrm{Var}(K/n) = \mathrm{Var}(K)/n^2$$

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Example: Genetic Drift

In a haploid population of n individuals, let p be the frequency of some allele A. We assume that (due to some simplifying assumptions?) the absolute frequency K of A in the next generation is (n, p)-binomially distributed.
For $X = K/n$, the relative frequency in the next generation follows:

$$\mathrm{Var}(X) = \mathrm{Var}(K/n) = \mathrm{Var}(K)/n^2 = n \cdot p \cdot (1-p)/n^2$$

$$= \frac{p \cdot (1-p)}{n}$$

Example: Genetic Drift

If we consider the change of allele frequencies over m generations, the variances add up. If m is a small number, such that p will not change much over m generations, the is variance of change of allele frequencies is approximately

$$m \cdot \text{Var}(X) = \frac{m \cdot p \cdot (1-p)}{n}$$

(because the changes per generation are independent of each other) and thus, the standard deviation is about

$$\sqrt{\frac{m}{n} \cdot p \cdot (1-p)}$$