

# How to present results of genomic studies in an intelligible form

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## Purpose of a scientific talk at a conference



- Convey a message to the audience
- The audience should be able to <u>understand</u> your reasoning
- Provide evidence that your empirical results are sound and reliable
- The audience should be <u>convinced</u> by your arguments

- Description of your experiment should ideally enable others to repeat the experiment
- Due to restrictions (mainly in time) often not fully achievable
- But still try to be as precise and comprehensive as possible





Step 1: State your research question

Ideally as formal hypothesis



If you want to test whether a certain factor F (a marker, a gene, a breed) has an effect on a variable y (growth, health):

- H<sub>o</sub>: F does not affect y
- H<sub>1</sub>: F does affect y (one-sided or two sided)

## Step 2: Describe your experiment

#### "Material" and methods



Provide all relevant information on the substrate you worked with (animals, tissues, technologies) in your experiment

Describe the actual experiment (what was done, how often, when were samples taken etc.)

Describe editing and quality control of the data (outlier detection, filtering criteria, imputation etc.)

Step 2: Describe your experiment

"Material" and methods



Describe the statistical approaches you use

Just naming a software (we used SAS/R ...) is not sufficient.

Provide the complete models you have used (including the relevant assumptions)

Describe in such detail that a knowledgeable person can follow

#### Direct model⇔ Direct-Social model



Peeters et al., EAAP 2013

For quality of life

Step 3: Present your results

Graphs are much more intuitive than tables – the human brain is analog







# Experiment I: which breed x treatment combination has the lowest performance? 3 seconds

	Treatment A	Treatment B	Treatment C
Breed 1			
Breed 2			
Breed 3			
Breed 4			



	Treatment A	Treatment B	Treatment C
Breed 1	4,3 ± 0,2	$\textbf{2,4} \pm \textbf{0,1}$	2,5 ± 0,1
Breed 2	2,5 ± 0,1	4,4 ± 0,2	3,3 ± 0,2
Breed 3	3,5 ± 0,3	$\textbf{2,1} \pm \textbf{0,4}$	2,3 ± 0,2
Breed 4	4,5 ± 0,2	2,8 ± 0,2	4,7 ± 0,3



# **Experiment I:** which breed x treatment combination has the lowest performance?





## Experiment II: which breeds react similar to the three treatments? 6 seconds

	Treatment A	Treatment B	Treatment C
Breed 1			
Breed 2			
Breed 3			
Breed 4			



## Experiment II: which breeds react similar to the three treatments? 6 seconds

	Treatment A	Treatment B	Treatment C
Breed 1	4,3 ± 0,2	$\textbf{2,4} \pm \textbf{0,1}$	$\textbf{2,5} \pm \textbf{0,1}$
Breed 2	2,5 ± 0,1	4,4 ± 0,2	$\textbf{3,3}\pm\textbf{0,2}$
Breed 3	3,5 ± 0,3	$\textbf{2,1} \pm \textbf{0,4}$	2,3 ± 0,2
Breed 4	4,5 ± 0,2	2,8 ± 0,2	4,7 ± 0,3







## **Experiment II:** which breeds react similar to the three treatments?





# Experiment II: which breeds react similar to the three treatments?



Step 3: Present your results



- standard errors
- confidence intervals
- posterior distributions









Average weight

Average weight

## Box plots are a good way of characterising the entire distribution



You can also show the 'confidence band' around an estimated (non-linear) regression



## Bayesian statistics provide posterior distributions of estimated quantities





Step 3: Present your results

Report the statistical significance of your results

- You have stated earlier which hypotheses were tested and which statistical tests were used
- Report p-values or ,usual' error levels (\* = 0.05; \*\* = 0.01; \*\*\* = 0.001)





Endogenous Cortisol by Sex Class

## Account for multiple testing



Nominal error level  $\alpha = 0.05$  (\*) means that under the H<sub>0</sub> (no effect) 1 out of 20 tests finds an effect

N = 1000 tests  $\rightarrow$  50 ,significant' effects even under the H<sub>0</sub>

- Bonferroni (1935) correction: use in each test the test level α /N; the global error probability is kept but testing is very (too) conservative – improved versions e.g. by Holm (1979) and Hochberg (1988)
- False Discovery Rate (Benjamini and Hochberg, 1995): Test such, that a proportion α of the significant results are false positive
- Permutation test (Doerge and Churchill, 1996): generate the distribution of the test statistic under H<sub>0</sub> through permutation of the data.

## Genome-wide vs. chromosome-wise testing

With multiple testing the power decreases with the number of tests

- If you can a priori restrict the location of a QTL to a subregion (e.g. a chromosome), then chromosome-wise testing is legitimate
- Otherwise you always have to account for <u>all</u> tests you are doing → only genome-wide results should be communicated



Step 3: Present your results - some basics

- state clearly what is shown on a slide
- put titles on axes in sufficiently large and readable fonts
- add a legend, if necessary
- Use colours and symbols in a systematic way
- Assign colours, symbols, and line types consistently across slides to the same object







Figures – which type?





Figures – which type?





Step 3: Present your results



Be creative!

- Avoid default settings of standard software
- Make use of the great opportunities of modern statistics and graphics programs
- Be inspired by what the leaders in the field are doing
- But: Fanciness should not be at the expense of clarity



Chromosome





Carbone et al., 2014 Nature





#### Groenen et al., 2014 Nature





Human (118) ECM-receptor interaction (4) Small cell lung cancer (3)

Mouse (84)

Dog (120) Hypertrophic cardiomyopathy (4) Dilated cardiomyopathy (4) Tight junction (4) Adherens junction (4) Focal adhesion (5) Regulation of actin cytoskeleton (4)

> Horse (311) Glycerolipid metabolism (4) Retinol metabolism (4) Endocytosis (8)

Cow (147) Fatty-acid metabolism (3) Regulation of actin cytoskeleton (6) Lysine degradation (3)

Pig (331) ECM-receptor interaction (7) ABC transporters (5) Focal adhesion (11) RNA degradation (5) Epithelial cell signalling in HPI (5) Spliceosome (7)

### **Final remarks**





It is worth spending some thought and time on preparing the perfect presentation of your research

Learn the rules so you know how to break them properly

Thank you!





# Experiment II: which breeds react similar to the three treatments?

