Hasso Plattner Institut
IT Systems Engineering | Universität Potsdam

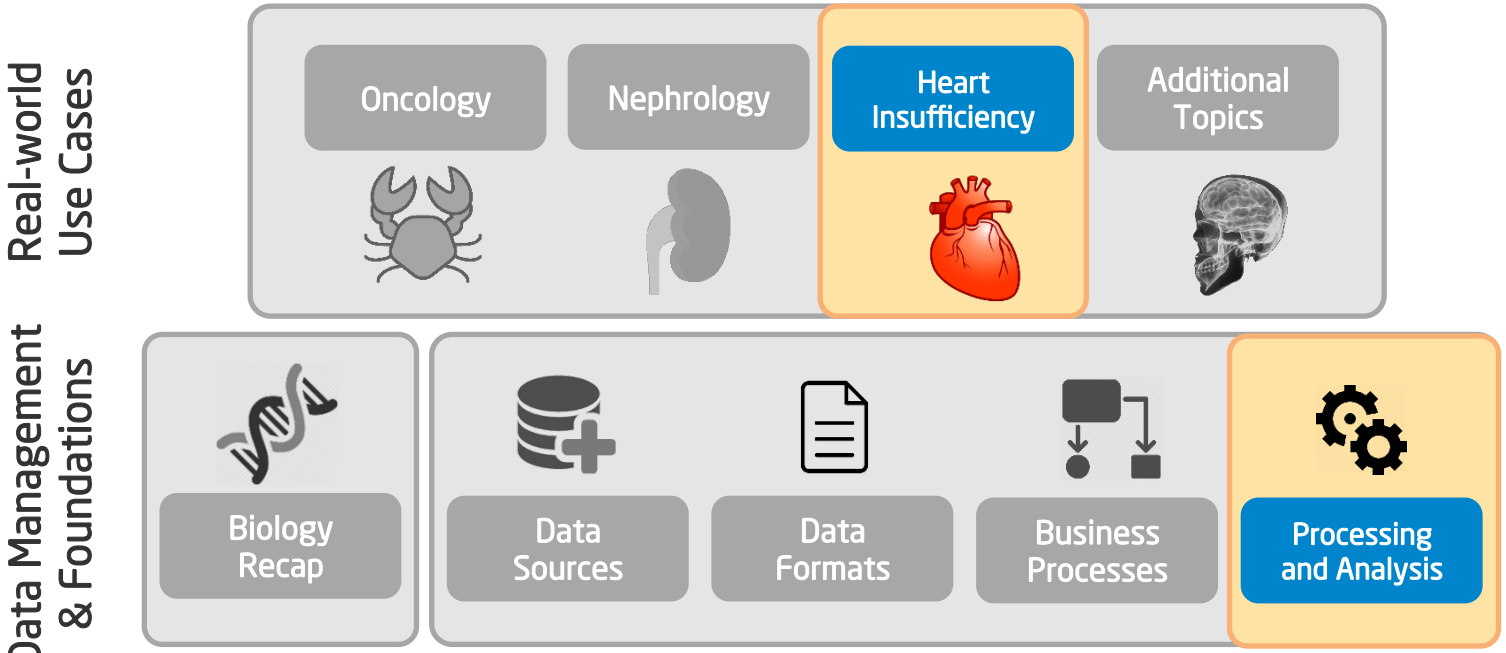
Evaluation Exercise III

Milena Kraus, Harry Cruz

Data Management for Digital Health

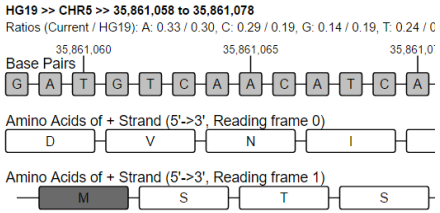
Summer 2017

Exercise III



Exercise III
Topics

- Genome Browser
- Medical Knowledge Cockpit
- Use Case Heart Failure (Systems Medicine)



AG Genome Browser

Jane Dough

female, 51 years, non-smoker

Markers

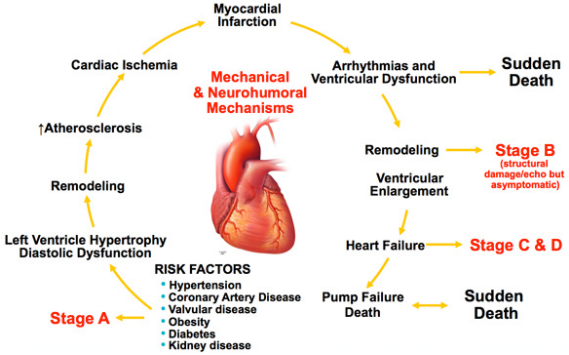
KRAS EGFR BRAF NRAS

Diagnosis

non-small cell lung cancer, stage IV

Charité

Medical Knowledge Cockpit



Evaluation Exercise III

Data Management for Digital Health, Summer 2017

Exercise III
Key Stats

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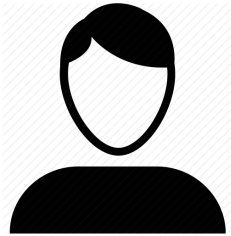
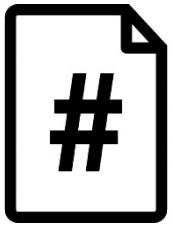
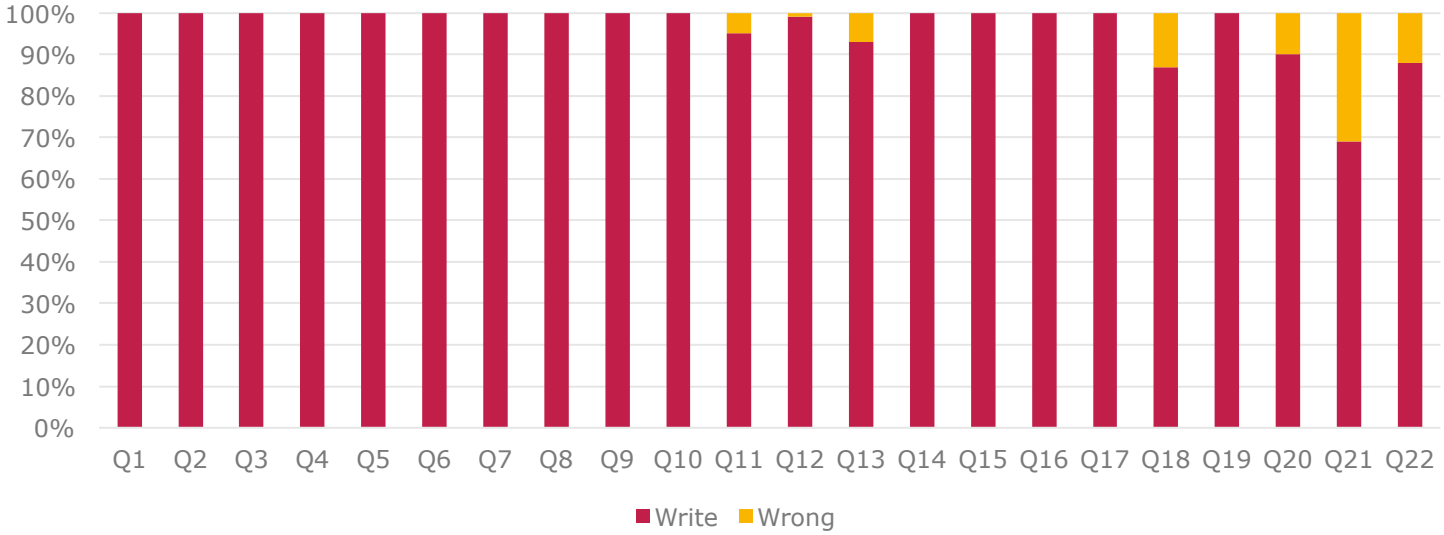
Part 1 – 5pts Part 2 – 35 pts	31 Students 29 Passed	Average score 5 / 100% 33.69 / 96%	Average time 68min





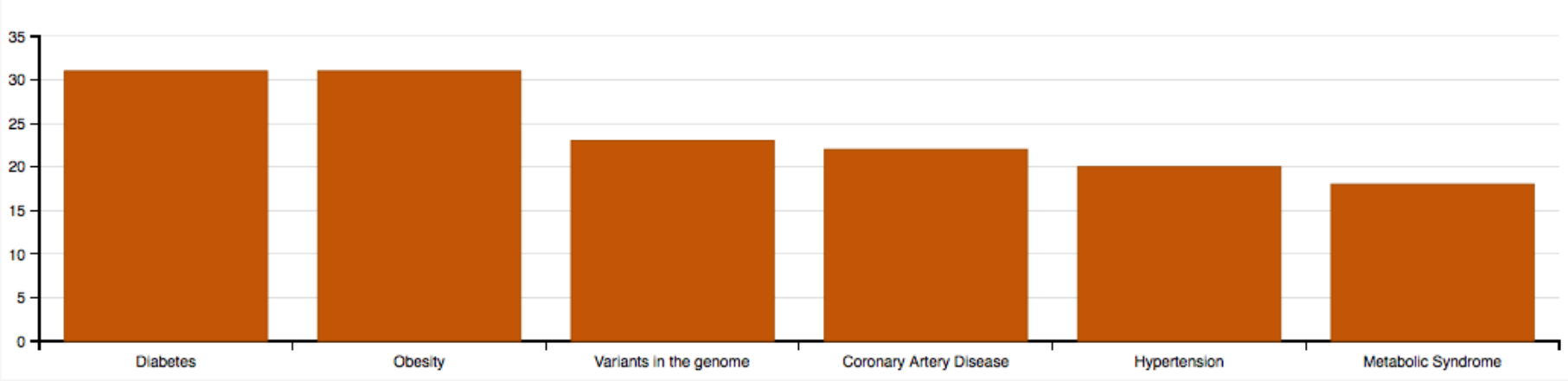
Exercise III
Key Stats



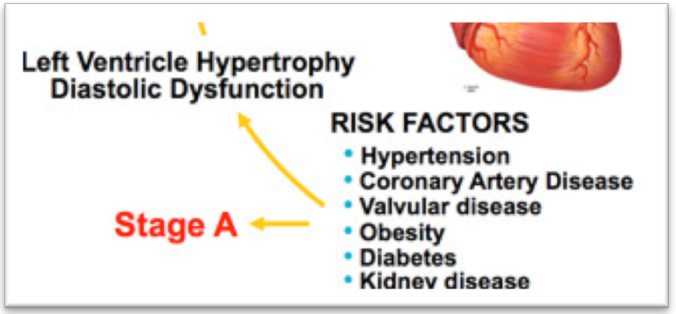
AG Genome Browser
Medical Knowledge Cockpit

Flawless!

Q 14: Which risk factors might affect the onset and progression of heart failure?



- Slide 20 (HF_SystemsMedicine): “Many genes have been found to be related to inherited forms of heart failure”
- Slide 51 (Mixed-type data analysis): “Metabolic syndrome is associated with the risk of developing cardiovascular disease and type 2 diabetes. “



Be aware of all right answers in multiple choice questions!

- In some cases, OpenHPI gave full credit if you picked at least one right answer (e.g. Q14, Q15, Q19)
- Please double check with the actual results and reconsider your answer before the exam
- In the exam, you will not get the full credit for only one correct answer!

Q 19: What are typical challenges that need consideration, when biomedical/ systems medicine data sets are analyzed, e.g., in clustering algorithms?

a) Data is of mixed-type (numerical, categorical etc.)

b) Missing values

c) Weight of variable subset within complete data set

d) Patient age

- Consider tables/variables from different origins, e.g.,:
  - 30 m variants x 200 patients
  - 20 k gene expression values x 200 patients
  - 150 clinical parameters x 200 patients
- Per default, all variables are accounted for in the same amount → variants would clearly dominate the complete analysis

Q 20: Principal Component Analysis is a technique applied to...

a) ... extract components of highest variation from a data set.

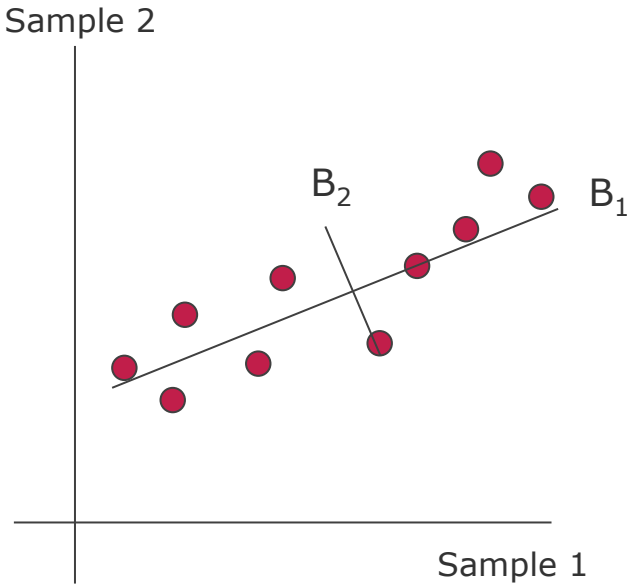
b) ... preprocess raw RNA sequencing reads.

c) ... reduce the dimensionality of large data sets.

d) ... model observed variables through a statistical distribution.

Principle Components Analysis

- Consider the mean of all points m, and a vector B going through the mean
- The vector B (PC1) is stretched along the path of most variation
- Vector B_2 (PC2) is stretched along the path of second most variation and orthogonal to B_1
- Length and orientation of the B vectors are most influenced by the outer points



Q21: Which of the following statements are correct regarding DNAseq and RNAseq?

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a) Their primary goal is to provide sequence information
b) After the preparation phase, sequencing is performed on DNA molecules
c) Sequencing output is usually a FASTQ file
d) There is no difference between algorithms used for the alignment of reads.

Information Content in RNA

- Information retrieved from RNA:
  - Quantity (primary, How many RNAs are transcribed from a specific gene?)
  - Sequence (secondary, as sequence information can be inferred more precisely from DNA)

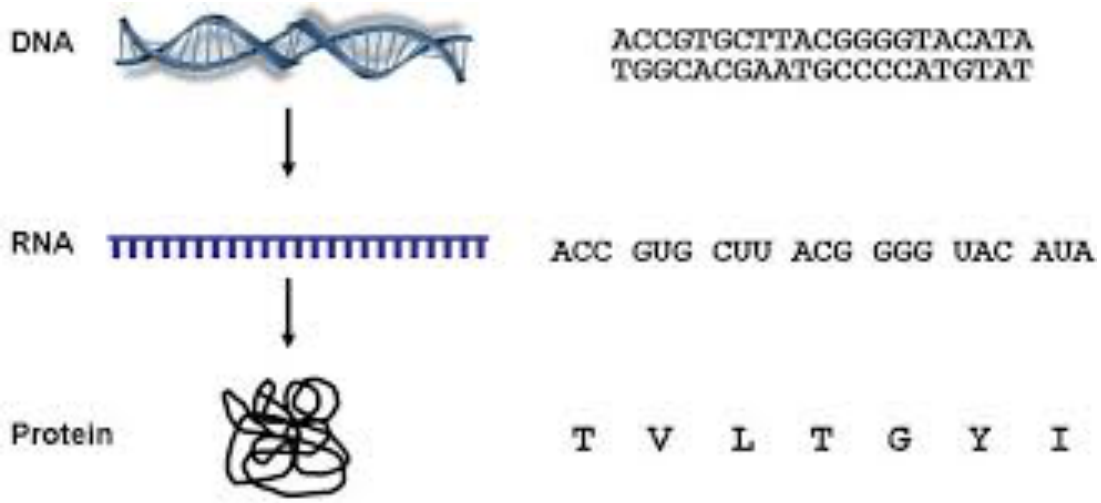


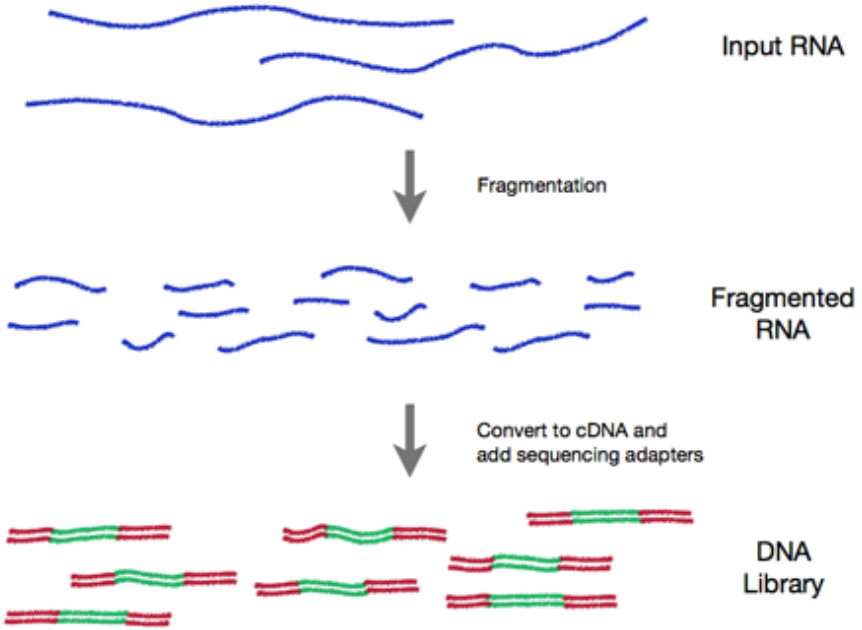
Image source: http://cureangelman.org/understanding-angelman/testing-101/

Experimental Procedure of RNAseq

- Generally similar to DNA sequencing
- Over 20.000 single stranded RNAs in variable abundance (1-k times) of 1.500-2.000 nt
- Fragmented into 30-200 nt

Differences:

- RNA is single stranded and needs to be revers-transcribed to DNA for sequencing
- Coverage is dependent on expression value of gene



Q22: Differential gene expression analysis is:

a) Used to compare different sample or patient groups
b) Performed on count data from RNAseq experiments
c) A method to find SNP's in RNAseq data
d) A model-based clustering method

- What caused the confusion to pick answer d?