

## Administrativa Week 12

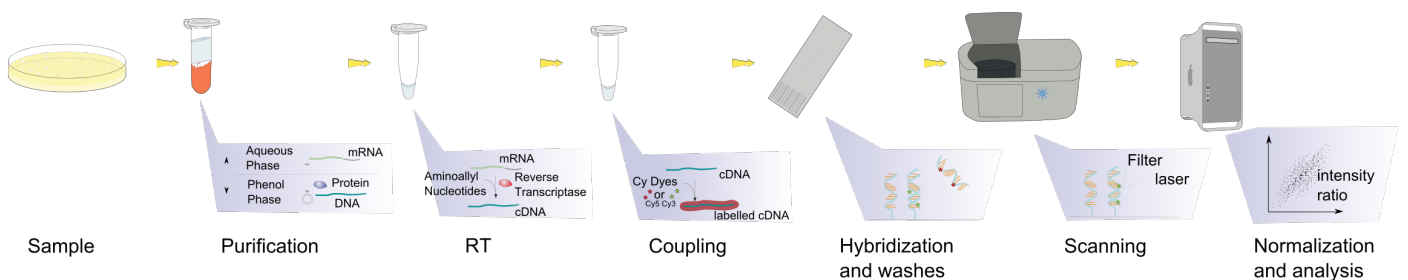
- Assignment 1 marks have been published -> Blackboard
  - check your filled-out marking rubric
- Assignment 2 – currently marked
- **Self-Reflection Survey**
  - This is an integral part of the assignment submission  
=> if you have not done so, please enter those today
  - We reserve the right to not mark a submission without self-reflection survey..
- Today's tutorials will concentrate **Assignment 2 demo**
- Assignment 3 has been published since Tuesday in Blackboard

## Assignment 3 Submission

- groupwork; worth 15%
- Use either Spark or Flink, depending on how assigned by Bilal (cf. announcement in Piazza)
- Deadline: **Tuesday, 13 June**
  - strictly speaking: Friday, 9 June, but without late penalty till 13 June
- Submission details
  - source code
  - documentation
  - measured execution times on small and large data set
  - *no demo this time*

## Assignment 3: Data Mining with Apache Flink/Spark

- Scenario: **Gene** Expression Study
  - cell samples taken from **cohort** of patients suffering different diseases
  - analysed with regard to which genes are 'active' in these cells
  - method: gene profiling using DNA microarrays



[Source: Wikipedia]

## Assignment 3 Data

- Source Data
  - GEO.txt
    - pre-processed gene expression data for each sample
    - patientid, geneid, expression value**
  - PatientMetaData.txt
    - meta-data about each patient
    - patientid, age, gender, postcode, diseases, drug\_response**
  - GeneMedatData.txt
    - meta-data about each gene, such as location on **genome** or which other gene is targeted by the produced **protein**; not used in this assignment

Note: patients can have multiple diseases

## Assignment 3 Tasks

- **Task 1: Explorative Data Analysis**  
“Number of cancer patients with certain active genes per cancer type”
- **Task 2: Frequent Itemset Mining** (Apriori Algorithm)  
Iterative algorithm to identify gene combinations which are often active together (active gene -> for the purpose of this assignment, simply genes with an expression value above a given threshold)
- **Task 3: Association Rule Generation**  
Based on the frequent itemset, create rules *some genes => other genes* which hold in the given data set with some confidence

## Assignment 3: Apriori Algorithm

- Apriori finds frequent itemset among **transactional** data set
  - “Which items are frequently bought together?”
- In this scenario:
  - transactions = gene expressions
  - items = (strongly) expressed genes
  - itemset = set of strongly expressed genes

patient	gene 1	gene 2	gene 3	gene 4	...	gene N
patient1	300	-800	2000	1300		2500
patient2	1650	0	1850	1550		500

- What is frequent?
  - any itemset with a minimum *support* (= count of occurrences)

## Example

### GEO.txt:

patient1, 1, 300  
patient1, 2, -800  
patient1, 3, 2000

...

patient2, 1, 1650  
patient2, 2, 0  
patient2, 3, 1850

...

patient 3, 1, 110

...

Note: We are only interested  
in results for cancer patients

represents a gene expression table:

patient	gene 1	gene 2	gene 3	gene 4	...	gene N
patient1	300	-800	2000	1300		80
patient2	1650	0	1850	1550		500
patient3	110	50	60	1900		100



Filtering for strongly expressed genes  
(eg. expression value above 1500):

'transaction' =>

patient	gene 1	gene 2	gene 3	gene 4	...	gene N
patient1	0	0	1	1		0
patient2	1	0	1	1		0
patient3	0	0	0	1		0

{gene3}  
{gene4}  
{gene3, gene4}

frequent itemsets  
(eg. support >= 2)

{gene1}  
{gene3}  
{gene4}  
{gene3, gene4}  
{gene1, gene3, gene4}

Itemsets  
(with at least  
one occurrence)

## Assignment 3: Apriori Algorithm (cont'd)

- Apriori is an efficient algorithm to find frequent itemset without the need to brute-force generation of all possible itemsets
- Input:
  - 'transactions' = set of strongly expressed genes per patient sample
  - support threshold (eg. if support threshold of 30%, then minimum support count must be  $> 0.3 * \text{number of transactions}$ )
- Step 1: Identify frequent 1-itemsets
  - Single genes which have a support count greater than threshold
- Step 2: Iterate
  - in each iteration, try extend current itemsets of size  $k$  to extend with any of the frequent 1-itemsets; if support  $>$  threshold, keep as  $k+1$ -itemset
- Stop iterating if either no new  $k+1$ -itemset found,  
or after max number of iterations

## Assignment 3: Task 3 – Association Rule Generation

- Input:
  - Frequent itemset from Task 2
  - confidence threshold (eg. At least 60% confidence in rules)
- Step 1: Generate all possible subsets of each frequent itemsets
- Step 2: For each subset  $R$  of a frequent itemset  $S$  check:
  - Calculate confidence of rule  $R \Rightarrow (S - R)$ :  
 $\text{confidence}(R \Rightarrow (S - R)) = \text{support}(S) / \text{support}(R)$
  - If confidence value is above given threshold, then keep in result

### Example:

- Input:
  - Frequent  $k$ -itemset from Task 2:
 

$\{\text{gene3}\}$   
 $\{\text{gene4}\}$   
 $\{\text{gene3, gene4}\}$
  - confidence threshold: 80%
- Step 1: Generate all possible subsets of each frequent  $k$ -itemsets
 

$\{\text{gene3}\}$   
 $\{\text{gene4}\}$   
 $\{\text{gene3, gene4}\}$

$\longrightarrow$

$\{\text{gene3}\}$   
 $\{\text{gene4}\}$   
 $\{\text{gene3} \ \{\text{gene4}\}$
- Step 2: For each subset  $R$  of a frequent itemset  $S$  check:
  - We can ignore the frequent 1-itemsets
  - Only rules to check hence:
 

$\{\text{gene3}\} \Rightarrow \{\text{gene4}\}$

confidence:  $2/2 = 100\%$

$\{\text{gene4}\} \Rightarrow \{\text{gene3}\}$

confidence:  $2/3 = 66\%$

$\longleftarrow$  Above confidence threshold of 80%
  - Calculate confidence of rules:  
 $\text{support}(\{\text{gene3}\}) = 2$     $\text{support}(\{\text{gene4}\}) = 3$     $\text{support}(\{\text{gene3, gene4}\}) = 2$