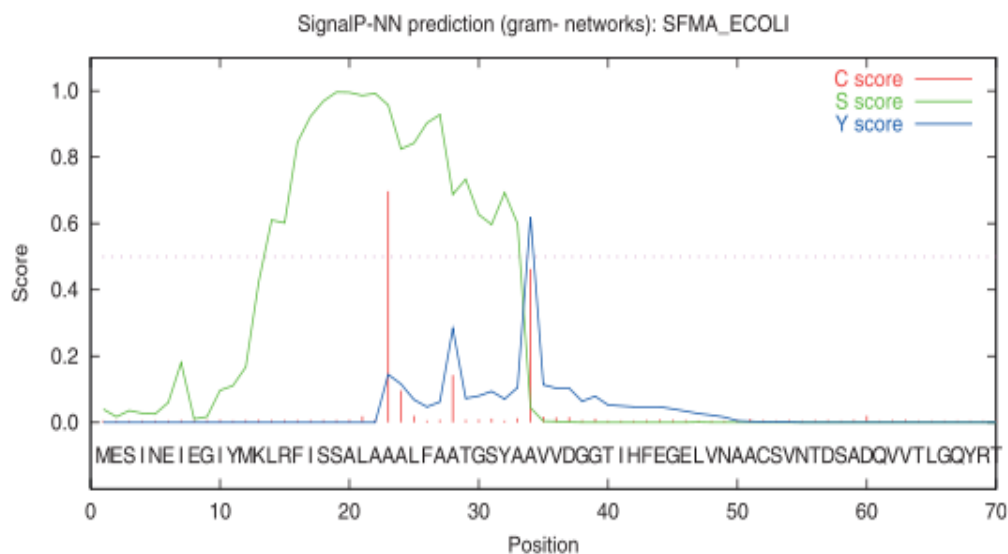


# BIOINFORMATICS

## Training Test

- 10 questions (real test: 20 questions)
  - 0.5 pts. / question (real test: 0.25 pts.)
  - 45 min. (real test: 90 min.)
  - 4 options. Only 1 option is correct.
  - Sign only the correct option.
  - No option signed = 0 pts.
  - A wrong option signed = -0.5 pts. (real test: -0.25 pts.)
1. A particular trait has a penetrance of 1 in homozygosity and 0 in heterozygosity both in males and females.
    - a) the trait is autosomal dominant
    - b) the trait is X-linked dominant
    - c) the trait is autosomal recessive
    - d) the trait is not a classical dominant / recessive trait
  2. The protein analysed in the following diagram:



- a) is probably a signal peptide: signalling region predicted is between aminoacid 34 and 70
  - b) is probably an intracellular cytosol protein
  - c) is probably a signal peptide and aminoacid 34 is potentially the cleavage site of RNA polymerase.
  - d) is probably a signal peptide and aminoacid 34 is probably the cleavage site of Signal Peptidase I.
3. Databases for consulting potential protein interactions are:
    - a) DIP, Intact, Biogrid
    - b) Biogrid, KEGG, BLAST
    - c) DIP, GEO, BLAST
    - d) ArrayExpress, GEO

4. Given the following pedigree file:

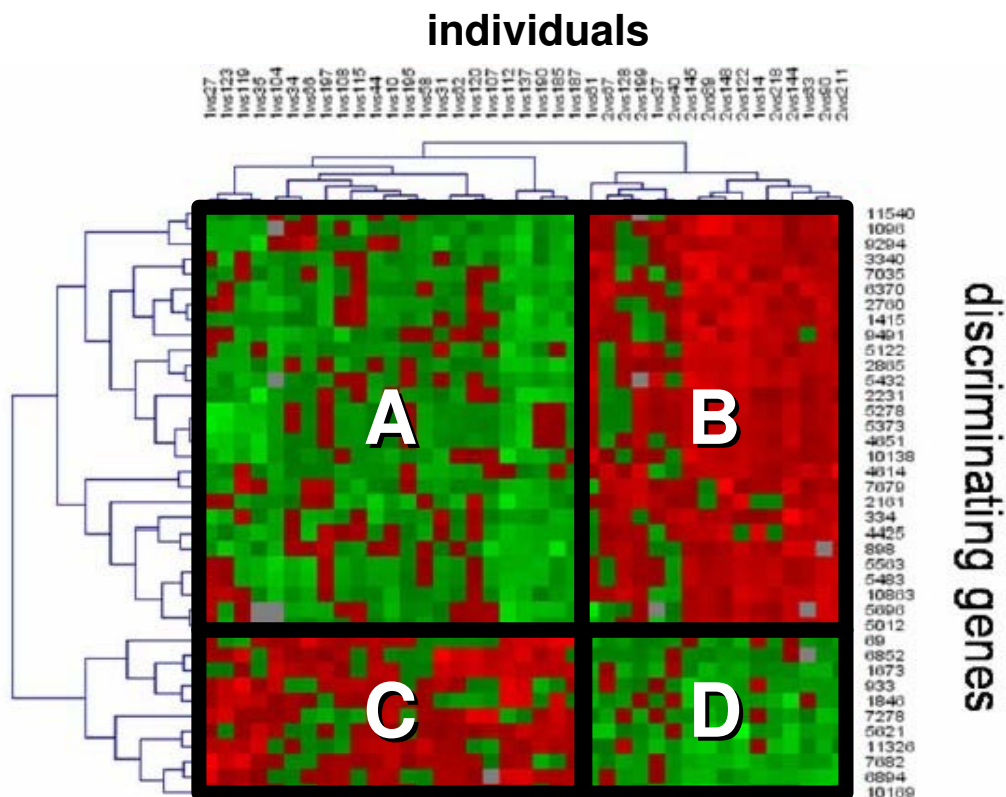
1	1	0	0	1	1	1	1	1	1	1	2
1	2	0	0	2	1	2	2	1	1	1	1
1	3	0	0	1	1	1	2	2	2	1	1
1	4	1	2	2	1	2	1	1	1	1	2
1	5	3	4	2	2	2	2	1	2	1	1
1	6	3	4	1	2	2	1	1	2	2	1
1	7	3	4	1	1	1	1	1	1	2	2

- the trait is continuous (qualitative)
- subject 2 shows the trait of interest
- the genotypes reveal that subject 3 is probably not the father of subject 7
- the software “pedstats” can be used to perform the Linkage Analysis and calculate the corresponding LOD scores.

5. A mutation affecting a regulatory site:

- has always a different effect on the phenotype than a functional mutation
- blocks the expression of a particular gene
- may alter the expression levels of a particular gene
- has always the same effect on the phenotype than a functional mutation

6. Given the following cluster analysis of gene expression profiles:

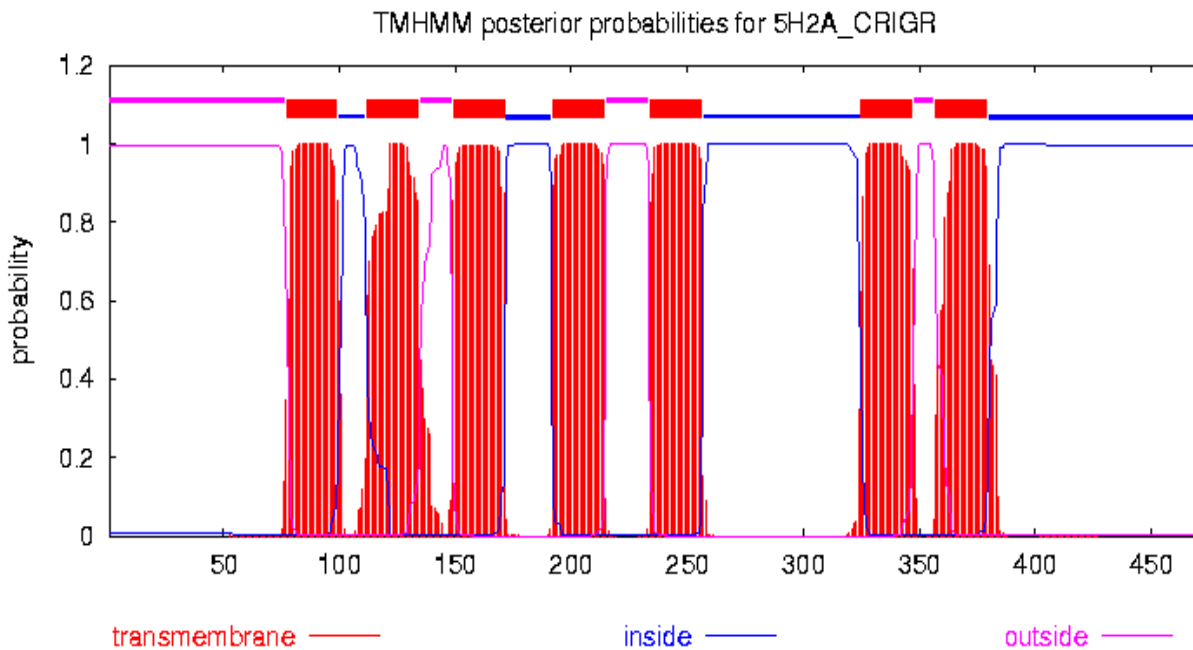


- the individuals are classified mainly in 2 large groups: AC and BD
- the genes are classified mainly in 4 large groups: A, B, C and D
- the individuals are classified mainly in 4 large groups: A, B, C and D
- the classification for this cluster analysis is not hierarchical

7. Promoters:

- a) attaches to the RNA Polymerase transcription machinery building supercoils
- b) are used to identify potential genes thanks to bioinformatic prediction *ab-initio*
- c) are the binding sites for enhancers and/or silencers
- d) attaches to the TATA-box building supercoils

8. Given the following diagram:



- a) the vertical bars show the cytoplasmic regions
  - b) the protein is most probably not a membrane protein
  - c) the vertical bars show Beta-laminar regions of the protein
  - d) the protein is most probably a membrane protein
9. We perform a comparative analysis of 10 related proteins to calculate a cladogram explaining their evolutionary relationship:
- a) using, for example, BLAST
  - b) using, for example, MERLIN
  - c) using, for example, PyMol
  - d) using, for example, ClustalX or ClustalW
10. If a BLAST search reveals the presence of two genes with a high percentage of Identity within the same organism, these two genes are most probably:
- a) syntenic
  - b) pseudogenes
  - c) paralogous
  - d) orthologous

1c.2d.3a.4c.5c.6a.7b.8d.9d.10c (uwaga: 8d !)

d