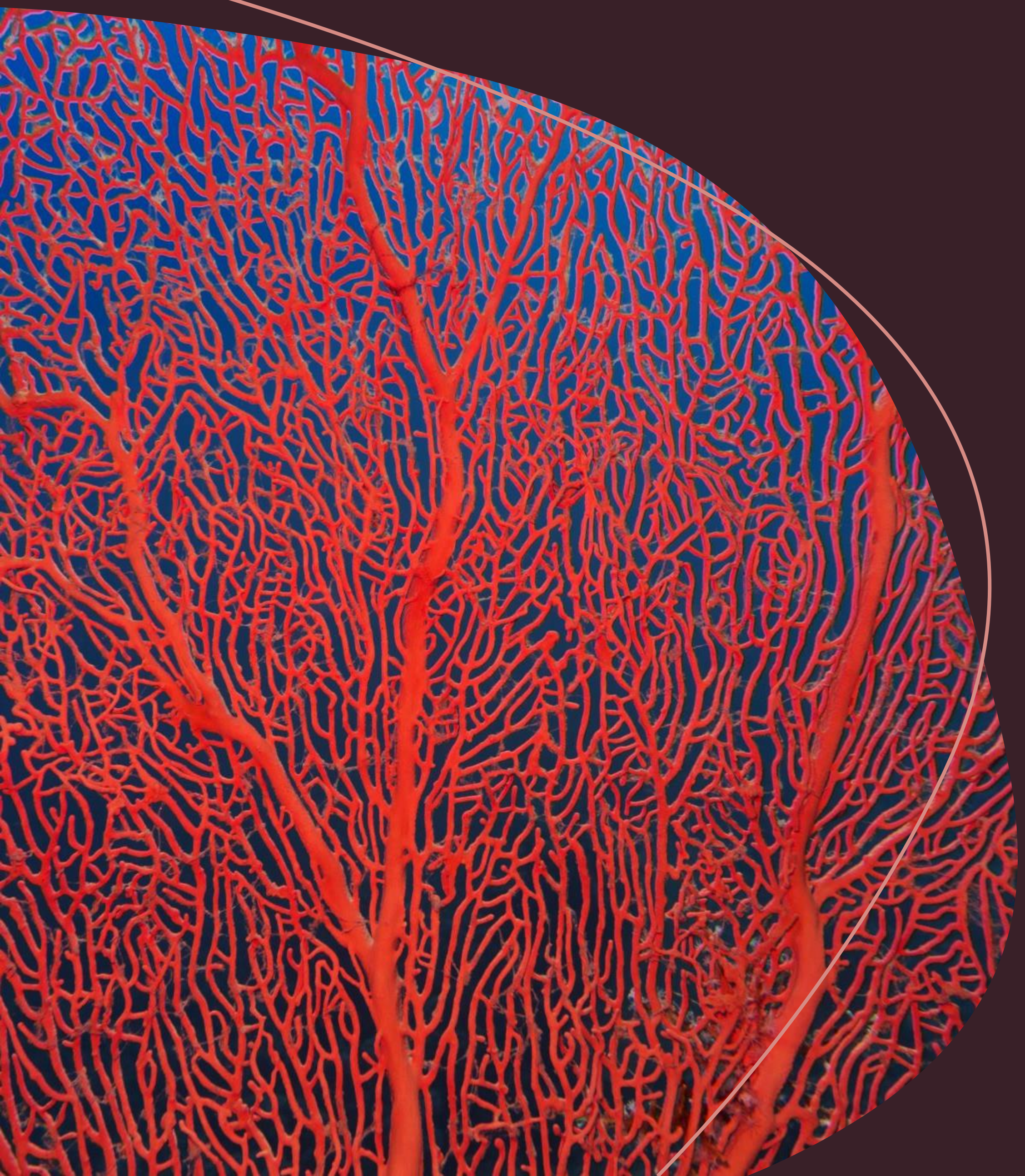


Data Mining in Bioinformatics

Fundamentals of Data Science

Project Presentation

(order of presenting)

Hosna Eltarras, Menna Mohammed,

Aisha Hagar, Yomna Jehad

What is Bioinformatics ?

- Understand Biological Data
- Biological data: Genes, RNA, Protein, EHRs and DNA Sequences
- DNA Language

ACGT

DNA Alphabet

AGGGAGAATGTTGAAACACAAGC

DNA Sentence

- English Human Language

A B C D E F ..

English Alphabet

I Love Bioinformatics

English Sentence

Reference: [1]

Explosion of Biological Data

Reference: [https://en.wikipedia.org/wiki/Genome]

Main WorkFlow

- Data Preprocessing
- Clustering
- Classification
- Deep learning

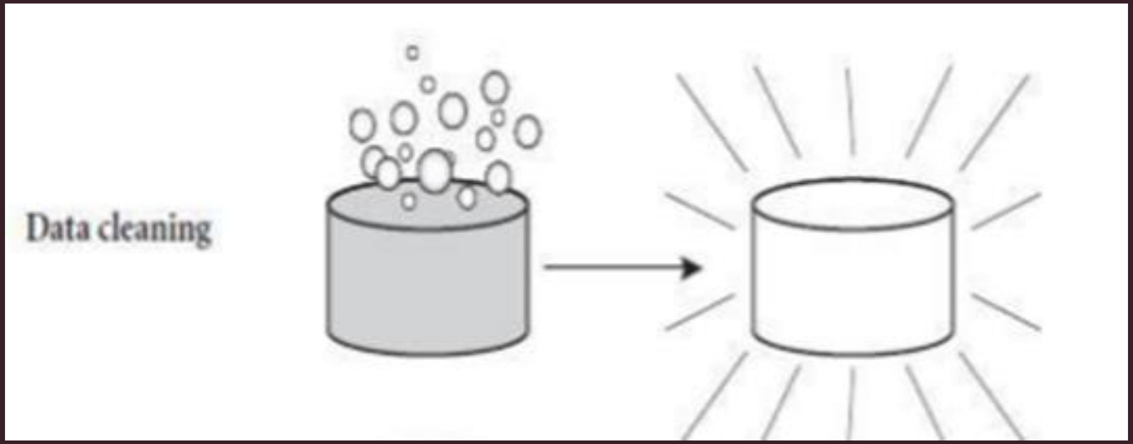
Pre-Processing

- Data cleaning
- Data transformation
- Data reduction

References: [2,3]

Data cleaning

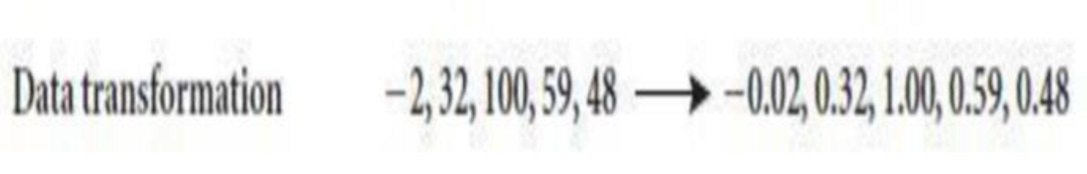
- Missing data
  - Ignore the tuples
  - Fill the missing values
- Noisy data
  - Binning
  - Regression
  - clustering



References: [2,3]

Data transformation

- Normalization
- Attribute selection
- Generalization

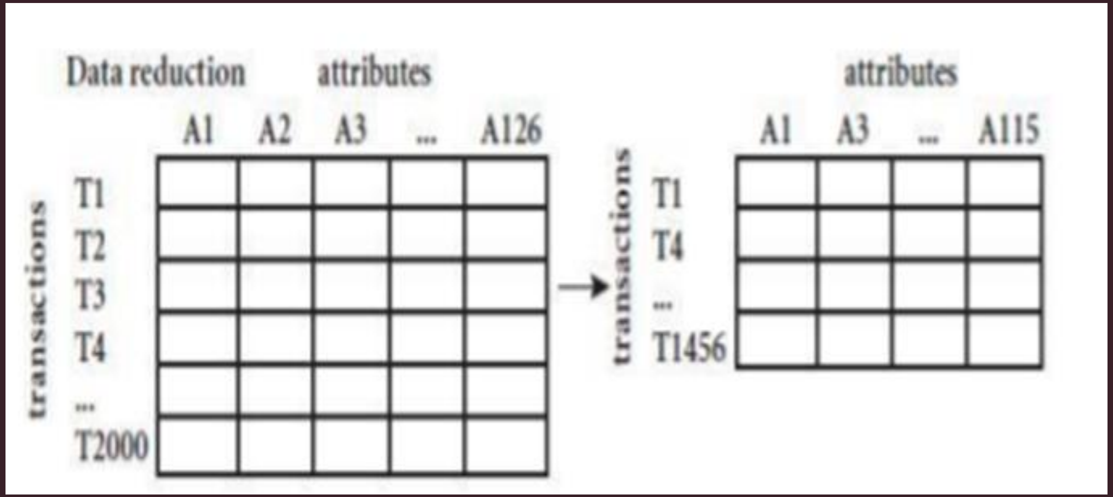


Data transformation −2, 32, 100, 59, 48 ⟶ −0.02, 0.32, 1.00, 0.59, 0.48

References: [2,3]

Data Reduction

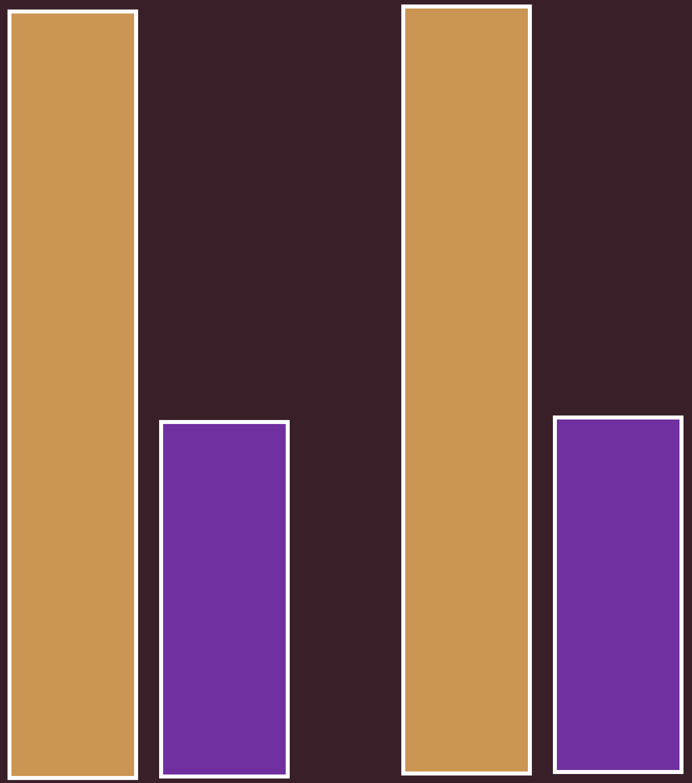
- Data cube aggregation
- Dimensionality reduction
- Numerosity reduction



References: [2,3]

Imbalanced Data

- Asymmetric Distribution
  - Hospitals
  - General Population
  - Rare Diseases

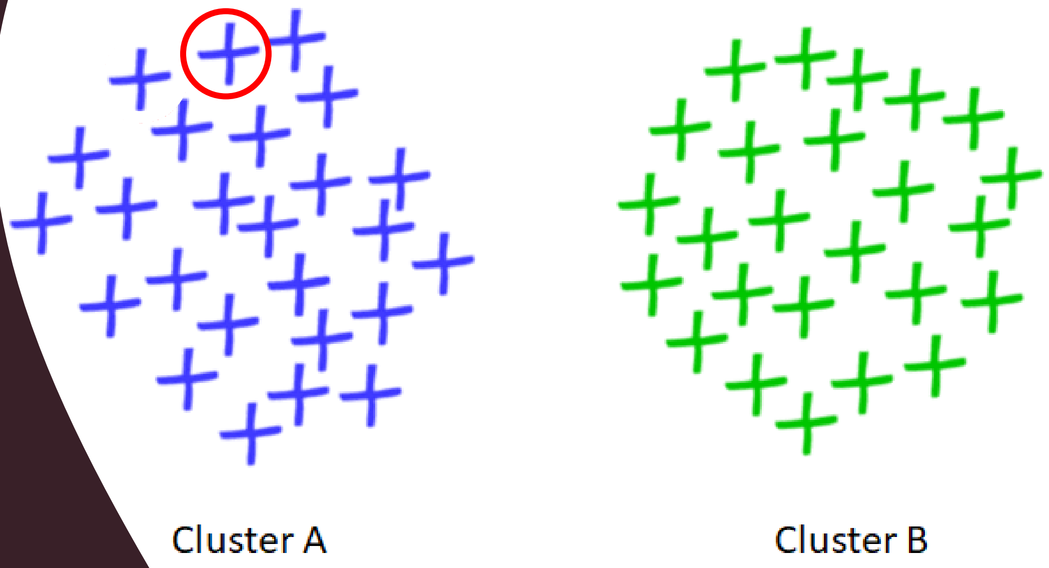


References: [4]

Clustering

Exploring hidden structure

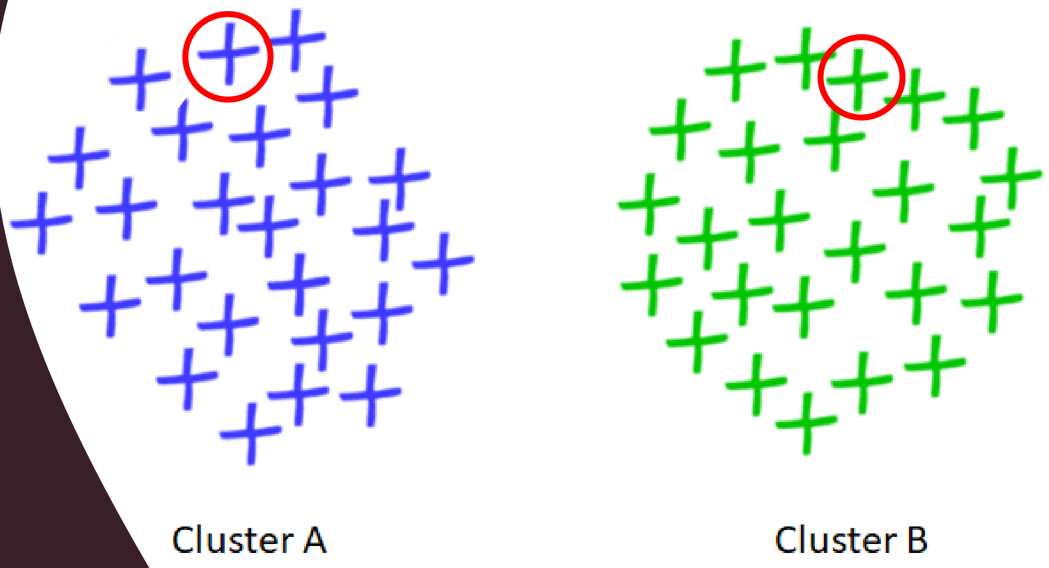
- What is the function of the Red Gene ?



References: [4,5]

Fuzzy Clustering

One gene in **Two** clusters !

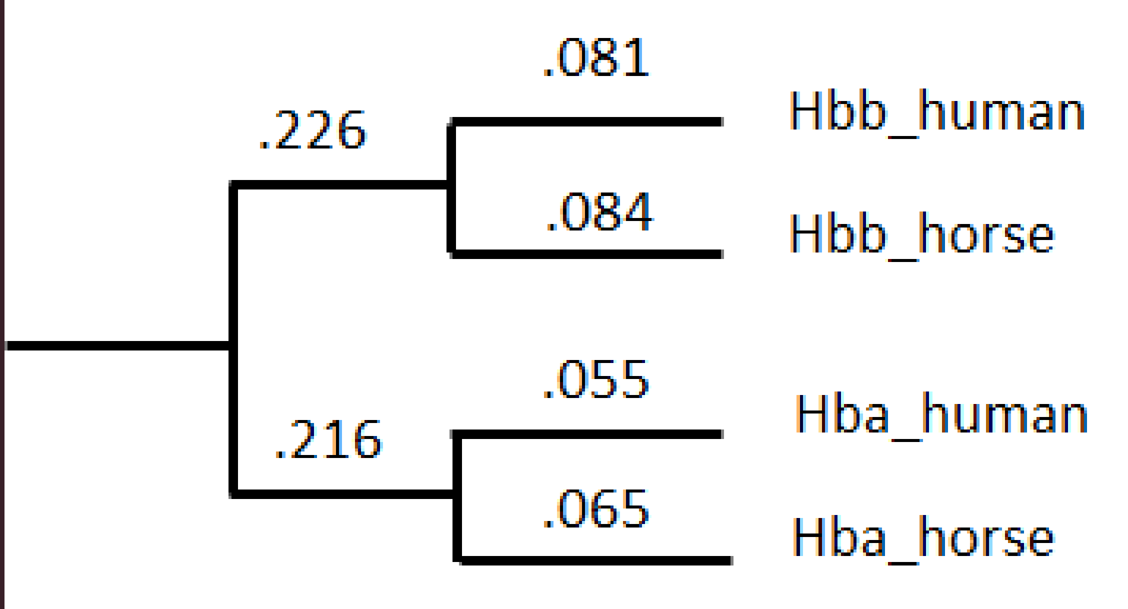


References: [4,5]

Hierarchical clustering:
phylogenetic trees

- Cure
- Clinical Trials

- Covid 19 and Bats,
  How did we know?



References: [4,5,6]

Classification in Bioinformatics

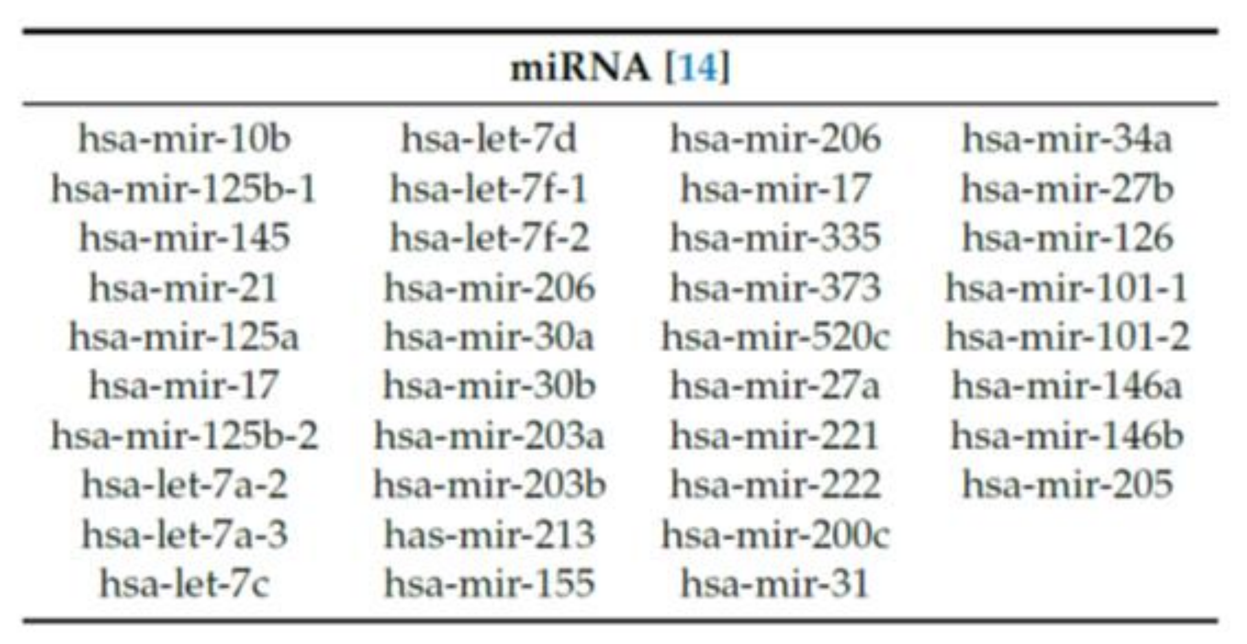
- Applications

Ex: Identify the gene signature of a disease, Classify patients' data for medical diagnosis, Evaluation of disease severity,… etc.

References: [7]

Breast Cancer Detection

- Dataset Features: MicroRNAs as biomarkers.



miRNA [14]			
hsa-mir-10b	hsa-let-7d	hsa-mir-206	hsa-mir-34a
hsa-mir-125b-1	hsa-let-7f-1	hsa-mir-17	hsa-mir-27b
hsa-mir-145	hsa-let-7f-2	hsa-mir-335	hsa-mir-126
hsa-mir-21	hsa-mir-206	hsa-mir-373	hsa-mir-101-1
hsa-mir-125a	hsa-mir-30a	hsa-mir-520c	hsa-mir-101-2
hsa-mir-17	hsa-mir-30b	hsa-mir-27a	hsa-mir-146a
hsa-mir-125b-2	hsa-mir-203a	hsa-mir-221	hsa-mir-146b
hsa-let-7a-2	hsa-mir-203b	hsa-mir-222	hsa-mir-205
hsa-let-7a-3	has-mir-213	hsa-mir-200c	
hsa-let-7c	hsa-mir-155	hsa-mir-31	

Fig: Clinically Verified MiRNAs

References: [7]

Flow of the Experiment

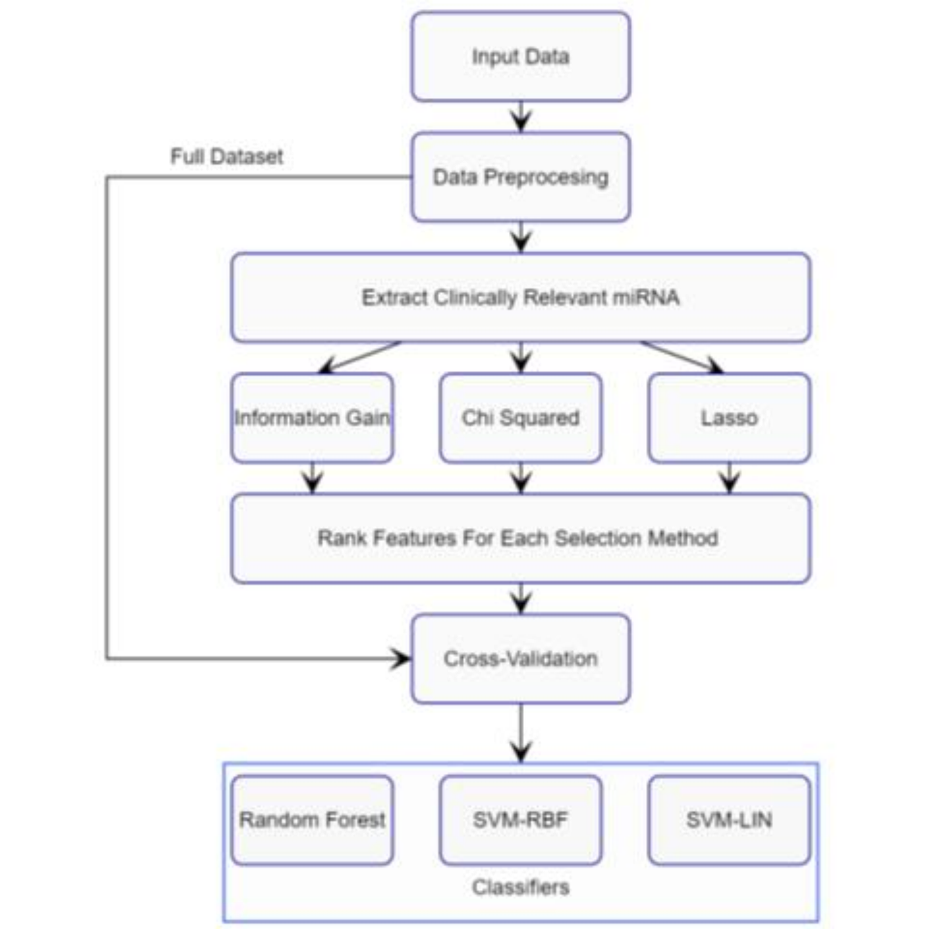
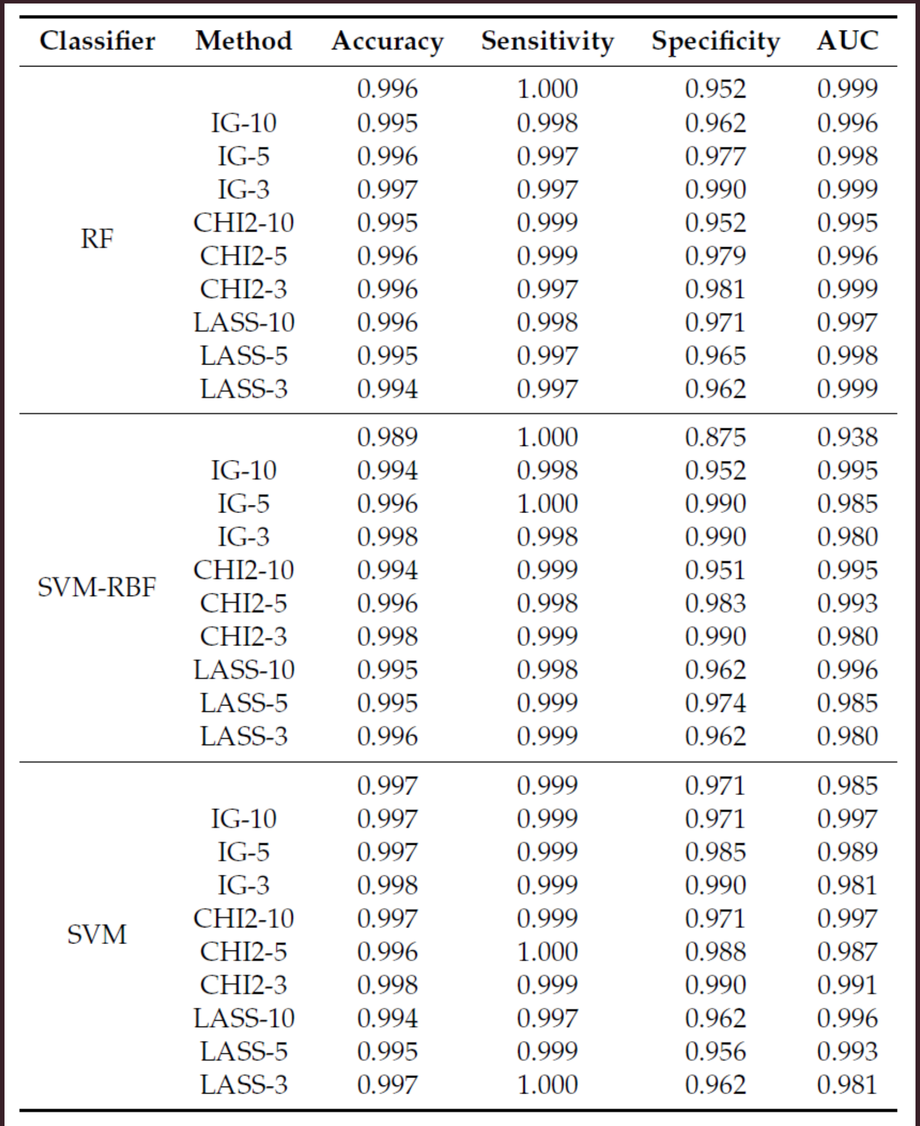


Fig: Schematics for Cancer Detection with Machine Learning

References: [7]

Evaluation

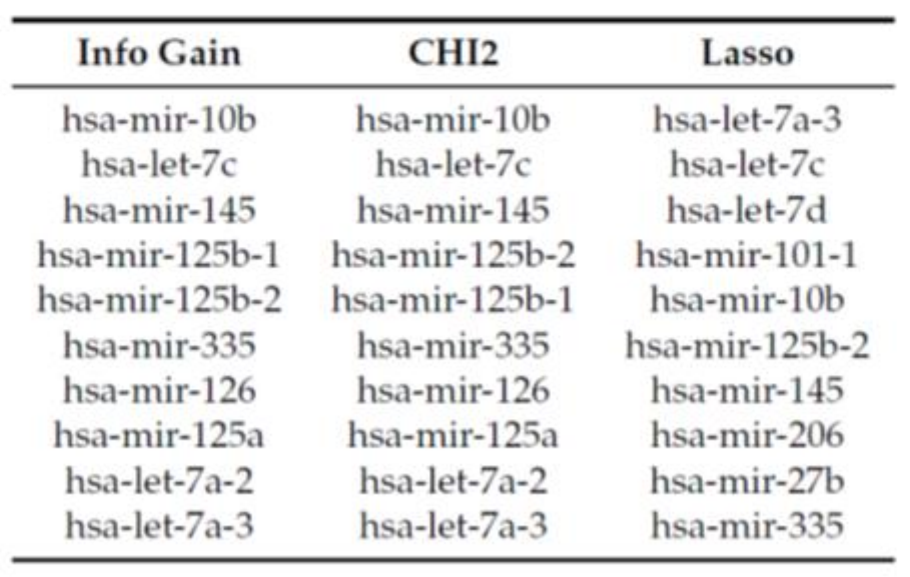


Classifier	Method	Accuracy	Sensitivity	Specificity	AUC
RF		0.996	1.000	0.952	0.999
	IG-10	0.995	0.998	0.962	0.996
	IG-5	0.996	0.997	0.977	0.998
	IG-3	0.997	0.997	0.990	0.999
	CHI2-10	0.995	0.999	0.952	0.995
	CHI2-5	0.996	0.999	0.979	0.996
	CHI2-3	0.996	0.997	0.981	0.999
	LASS-10	0.996	0.998	0.971	0.997
	LASS-5	0.995	0.997	0.965	0.998
	LASS-3	0.994	0.997	0.962	0.999
SVM-RBF		0.989	1.000	0.875	0.938
	IG-10	0.994	0.998	0.952	0.995
	IG-5	0.996	1.000	0.990	0.985
	IG-3	0.998	0.998	0.990	0.980
	CHI2-10	0.994	0.999	0.951	0.995
	CHI2-5	0.996	0.998	0.983	0.993
	CHI2-3	0.998	0.999	0.990	0.980
	LASS-10	0.995	0.998	0.962	0.996
	LASS-5	0.995	0.999	0.974	0.985
	LASS-3	0.996	0.999	0.962	0.980
SVM		0.997	0.999	0.971	0.985
	IG-10	0.997	0.999	0.971	0.997
	IG-5	0.997	0.999	0.985	0.989
	IG-3	0.998	0.999	0.990	0.981
	CHI2-10	0.997	0.999	0.971	0.997
	CHI2-5	0.996	1.000	0.988	0.987
	CHI2-3	0.998	0.999	0.990	0.991
	LASS-10	0.994	0.997	0.962	0.996
	LASS-5	0.995	0.999	0.956	0.993
	LASS-3	0.997	1.000	0.962	0.981

Fig: Performance Metrics of Classifiers with Different Feature Selection Methods Over MiRNAs Subsets(3, 5, 10)

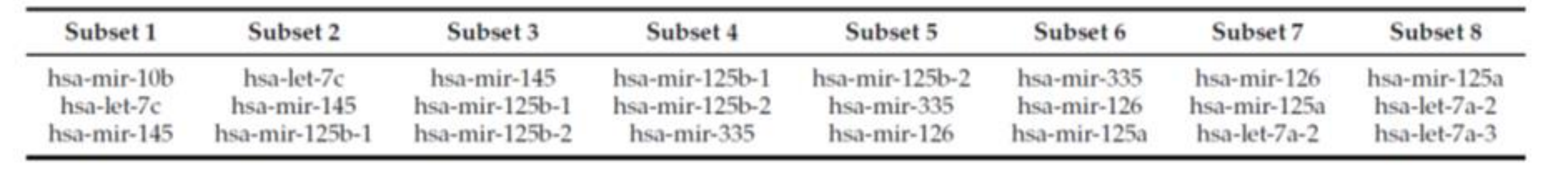
References: [7]

Selecting Fewer Features for Classification



Info Gain	CHI2	Lasso
hsa-mir-10b	hsa-mir-10b	hsa-let-7a-3
hsa-let-7c	hsa-let-7c	hsa-let-7c
hsa-mir-145	hsa-mir-145	hsa-let-7d
hsa-mir-125b-1	hsa-mir-125b-2	hsa-mir-101-1
hsa-mir-125b-2	hsa-mir-125b-1	hsa-mir-10b
hsa-mir-335	hsa-mir-335	hsa-mir-125b-2
hsa-mir-126	hsa-mir-126	hsa-mir-145
hsa-mir-125a	hsa-mir-125a	hsa-mir-206
hsa-let-7a-2	hsa-let-7a-2	hsa-mir-27b
hsa-let-7a-3	hsa-let-7a-3	hsa-mir-335

Fig: Top Ranked Features Under Different Feature Selection Techniques



Subset 1	Subset 2	Subset 3	Subset 4	Subset 5	Subset 6	Subset 7	Subset 8
hsa-mir-10b	hsa-let-7c	hsa-mir-145	hsa-mir-125b-1	hsa-mir-125b-2	hsa-mir-335	hsa-mir-126	hsa-mir-125a
hsa-let-7c	hsa-mir-145	hsa-mir-125b-1	hsa-mir-125b-2	hsa-mir-335	hsa-mir-126	hsa-mir-125a	hsa-let-7a-2
hsa-mir-145	hsa-mir-125b-1	hsa-mir-125b-2	hsa-mir-335	hsa-mir-126	hsa-mir-125a	hsa-let-7a-2	hsa-let-7a-3

Fig: Subsets of Ranked miRNAs

Performance Evaluation Over Different Subsets

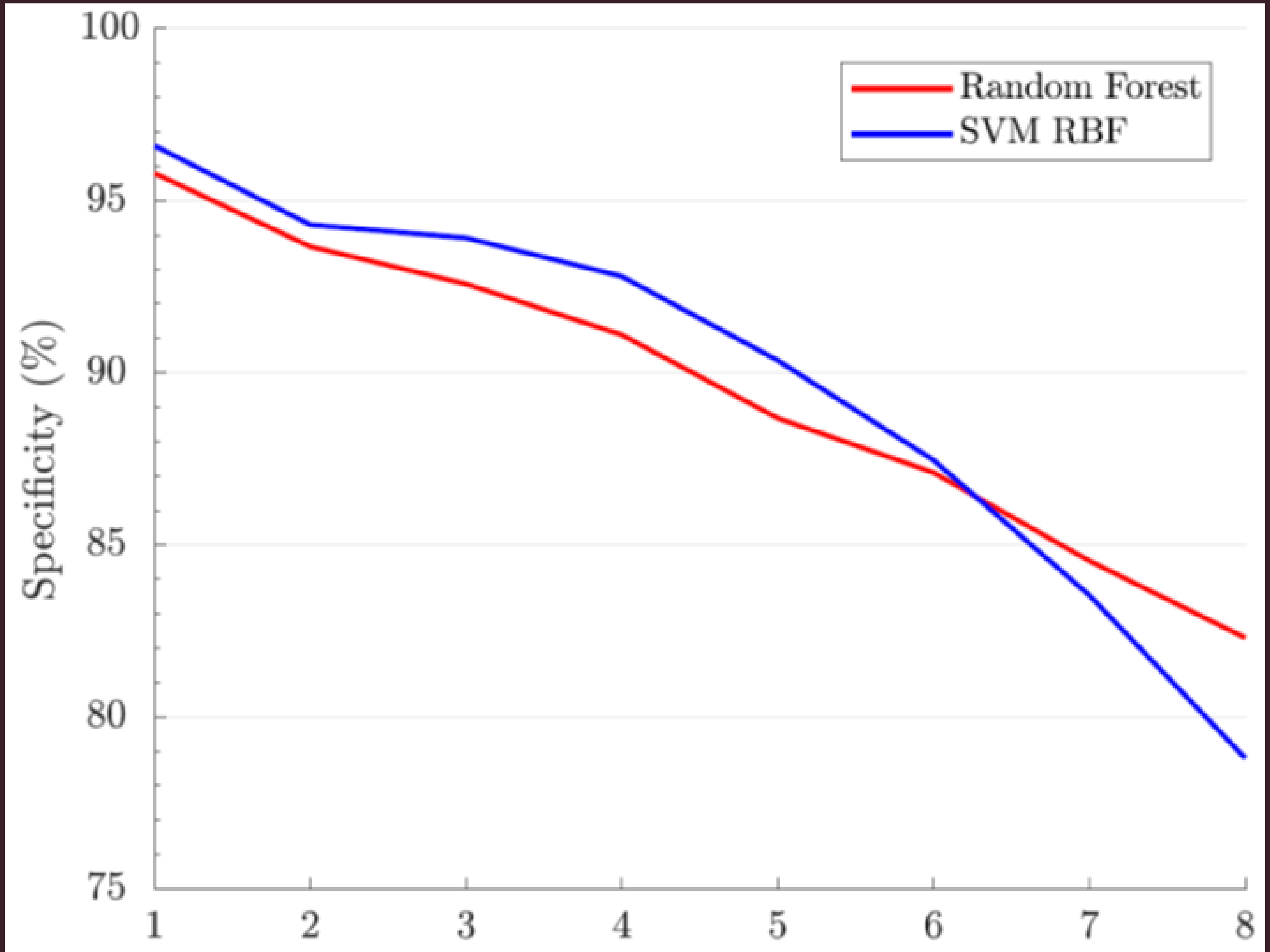
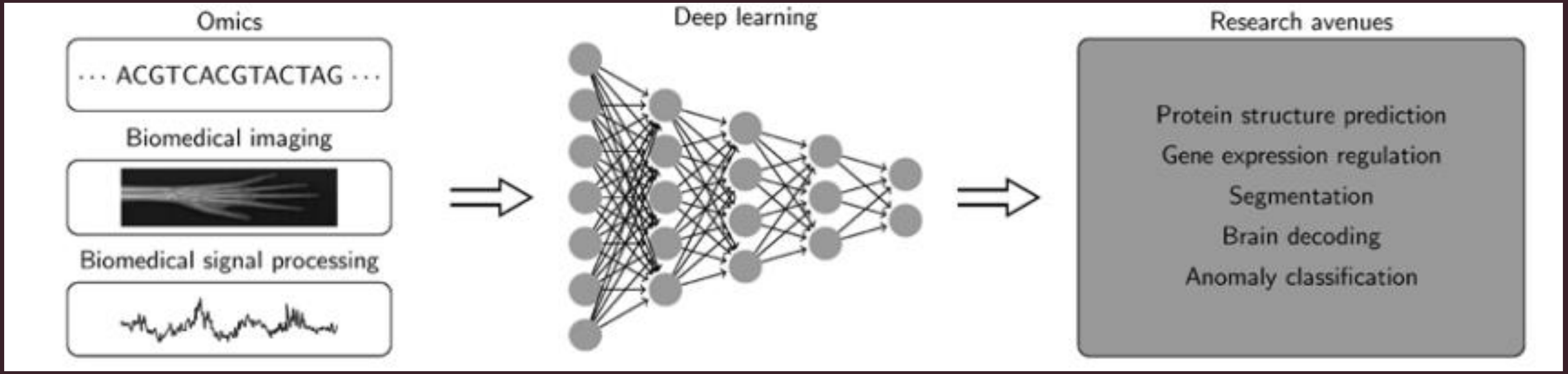


Fig: Specificity Across Different Clinical miRNA Subsets

References: [7]

Deep learning in bioinformatics

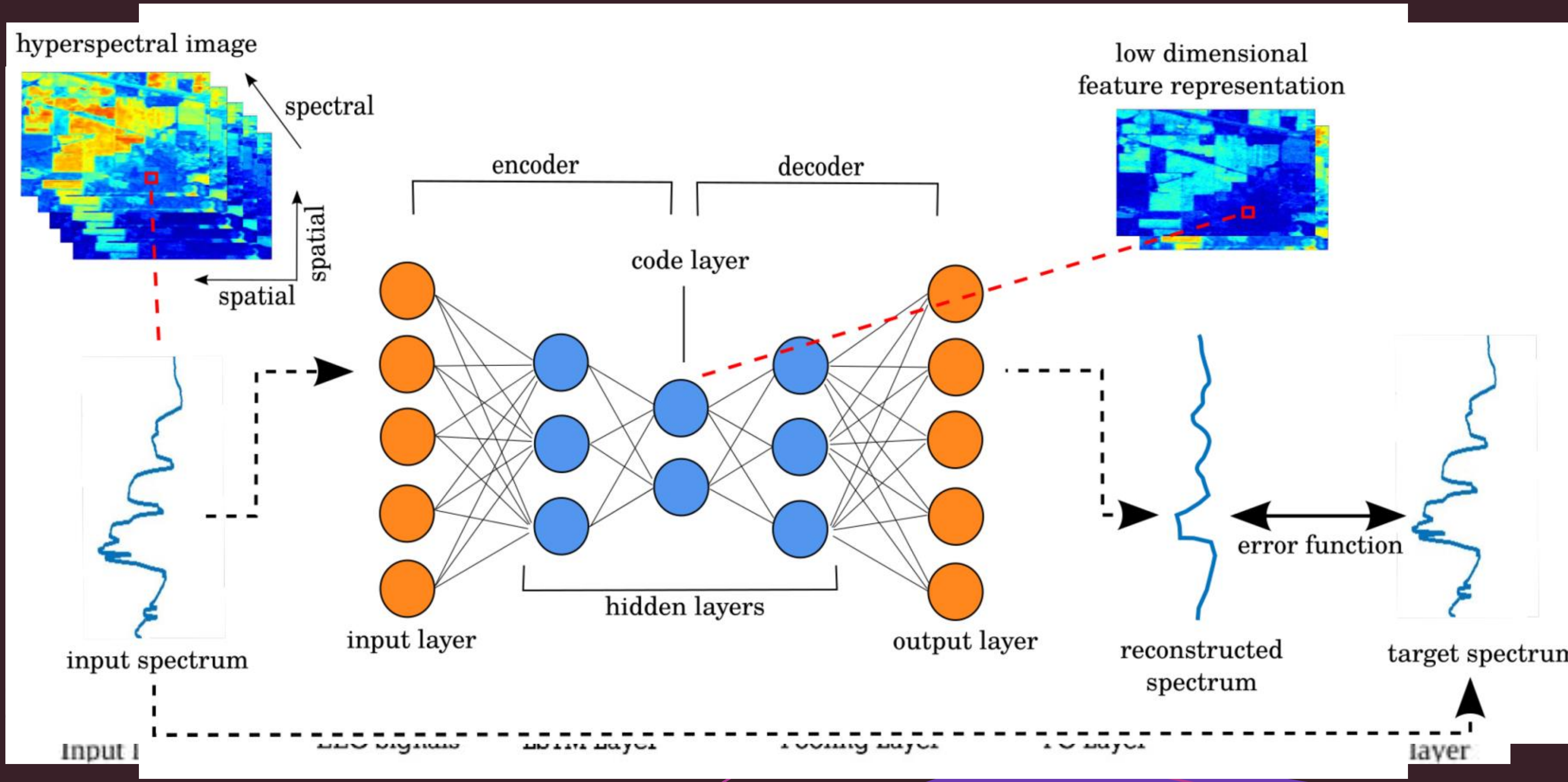


- Increasing computational capacity and the improved algorithms.
- Massive amount of data.
- Automatic feature extraction.

References: [8,9]

Deep learning Architectures & Applications

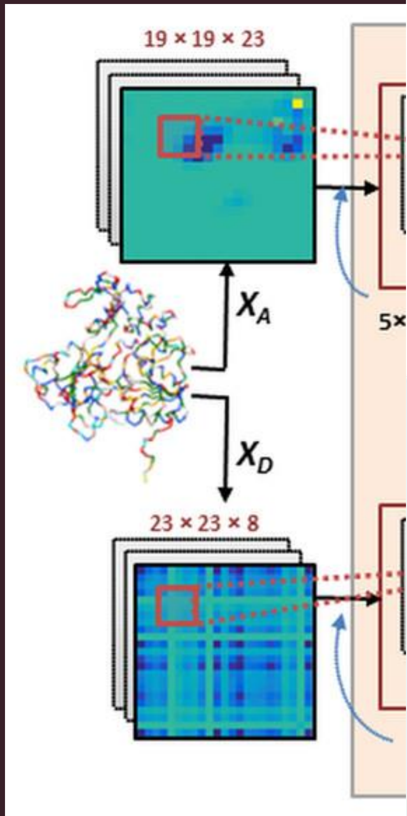
- Convolutional neural networks.
- Recurrent neural networks.
- Autoencoder.

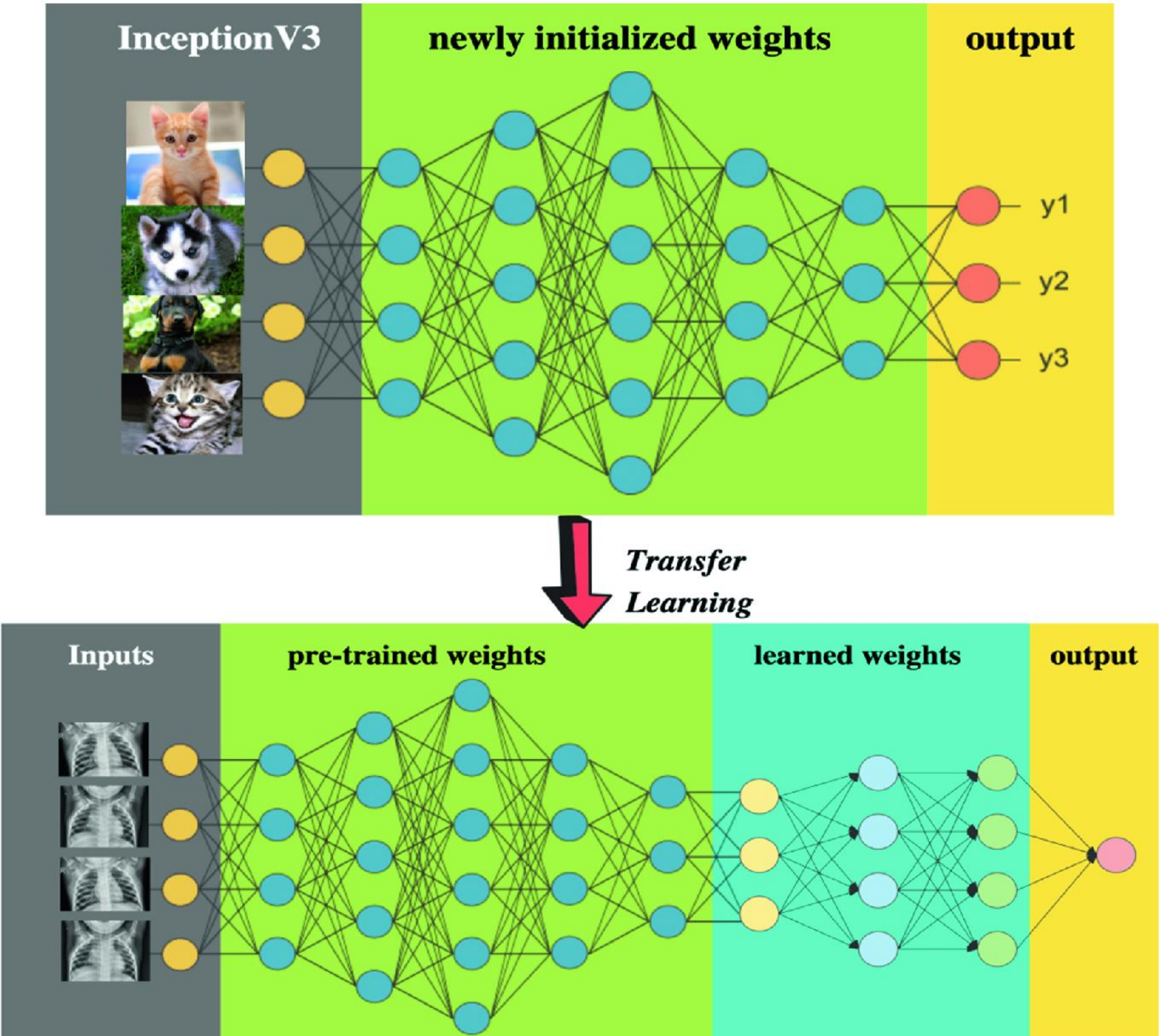


References: [8,9]

Deep learning Architectures & Applications

- Ensemble deep learning in bioinformatics.
- Transfer Learning Deep Learning.





References: [8,9]

Challenges

- Heterogeneous data
- No standard schema
- Imbalanced data
- Interpretability of models
- Computational challenges

CURSE of Dimentionality

References: [4,10]

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