



Κέντρο Νέων Βιοτεχνολογιών και Ιατρικής Ακριβείας ΚεΝεΒΙΑ
Ερευνητική Υποδομή pMedGR

www.precisionmedicine.gr



Webinar

**1st Call for proposals for FREE Research Services:
Presentation of services and discussion of technical details**



Με τη συγχρηματοδότηση της Ελλάδας και της Ευρωπαϊκής Ένωσης

*Zoom meeting
28 Μαΐου 2020*



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pMedGR

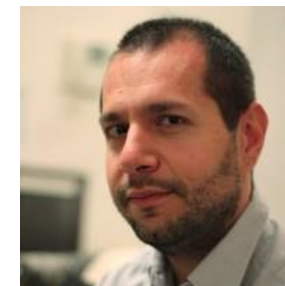
Βιοπληροφορική Ανάλυση Data Analysis & Bioinformatics Unit



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Κλειώ-Μαρία Βέρρου, MSc



Παναγιώτης Μούλος, PhD

Pipelines (Exome and Whole genome sequencing)

Input

Reads (bcl or fastq)

Output

Detailed Statistics Report

Aligned Reads

Report of the DNA alteration (exact locations, statistical significance, confidence metrics)

Genome browser visualization

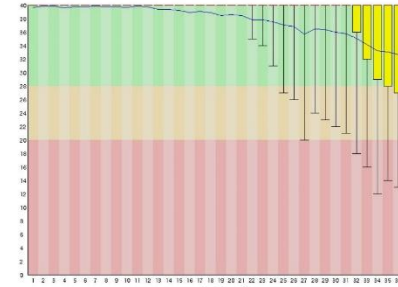
Sequencing



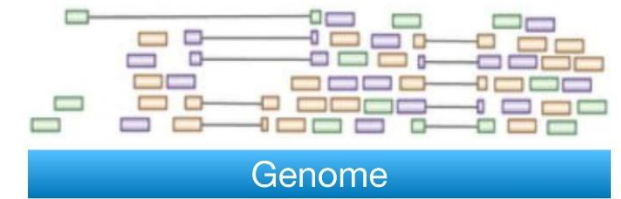
Base Calling



Quality Control



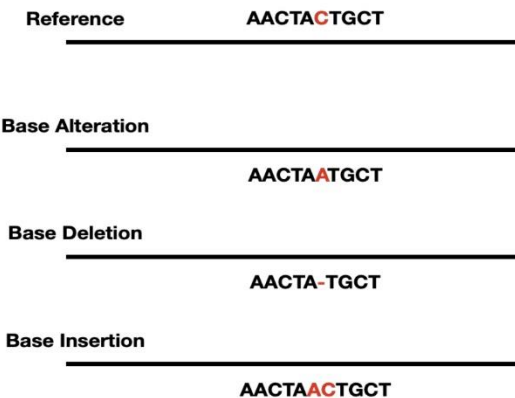
Genome Alignment



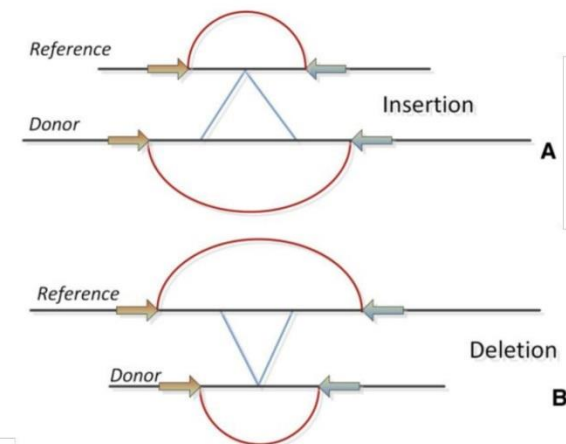
CNVs/SNPs

	Position Start	Position End	Ref	Variation	...
Chr1	183083734	183083734	G	-	
Chr3	13295559	18295559	G	A	
Chr4	25648977	25648977	-	T	
...	...	...			

Variant Calling



Copy Number Analysis



Pipelines (RNAseq)

Input

Reads (bcl or fastq)

Output

Detailed Statistics Report
(quality figures)

Transcripts' quantification file

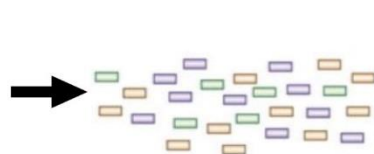
Report of gene lists
(annotations, statistical significance, confidence metrics)

De-regulated genes in a volcano plot

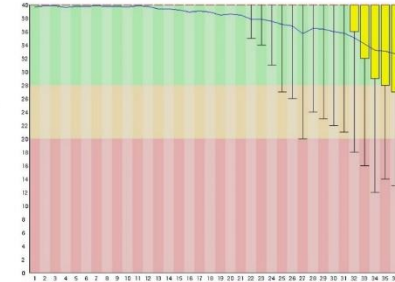
Sequencing



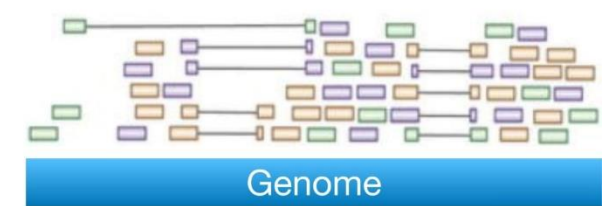
Reads



Quality Control



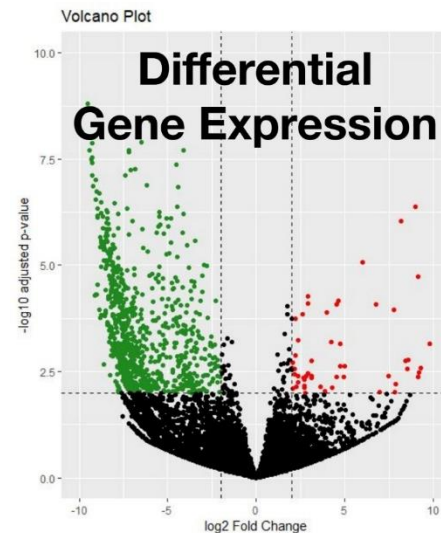
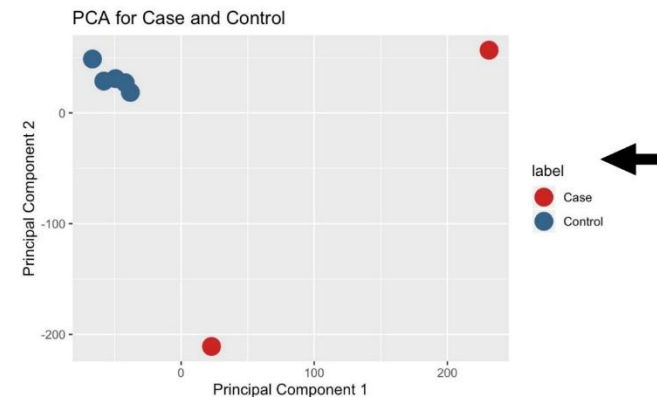
Genome Alignment



Transcript Quantification

	Exp_1	Exp_2	Ctrl_1	Ctrl_2
GeneA	10	11	56	54
GeneB	128	54	130	131
GeneC	42	4	38	35
...	...	...		

Dimensionality Reduction



Pipelines (scRNAseq)

Input

Reads (bcl or fastq)

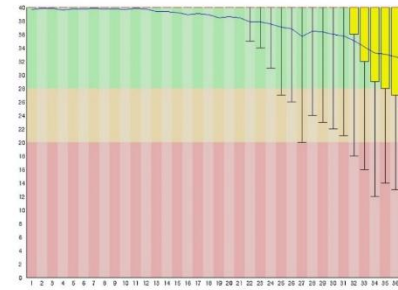
Sequencing



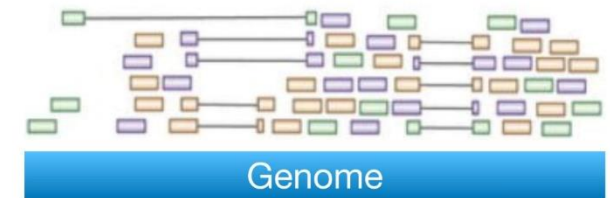
Reads



Quality Control



Genome Alignment



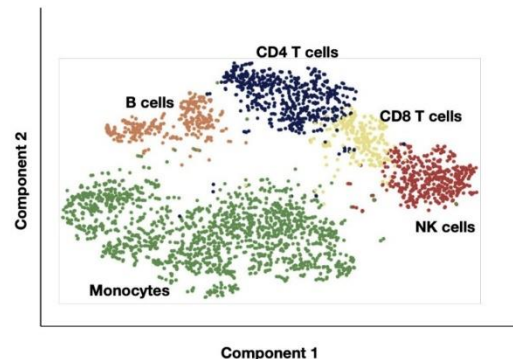
Output

Detailed Statistics Report
(quality figures)

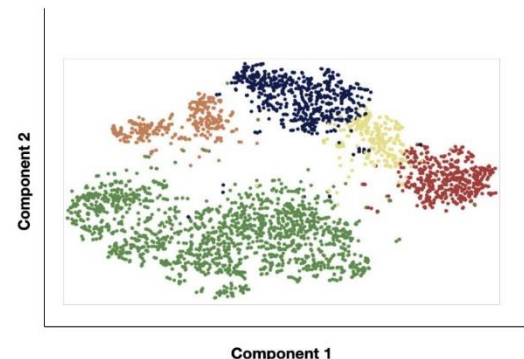
Transcripts'
quantification file

Basic visualization of the
results (dimensionality
reduction plots, cells
clustering)

Identification of different populations



Dimensionality Reduction and Clustering



Transcript Quantification

	Cell1	Cell2	Cell3	...
GeneA	10	11	56	54
GeneB	128	54	130	131
GeneC	42	4	38	35
...	...	...	...	...