



Κέντρο Νέων Βιοτεχνολογιών και Ιατρικής Ακριβείας ΚεΝεΒΙΑ  
Ερευνητική Υποδομή pMedGR



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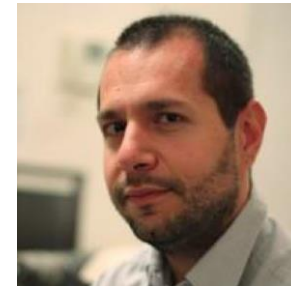
# Βιοπληροφορική Ανάλυση Data Analysis & Bioinformatics Unit



Γιώργος Παυλόπουλος, PhD



Κλειώ-Μαρία Βέρρου, MSc



Παναγιώτης Μούλος, PhD

# Whole Exome Sequencing: Ποή εργασίας

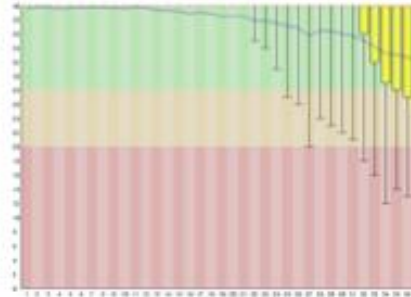
## Sequencing



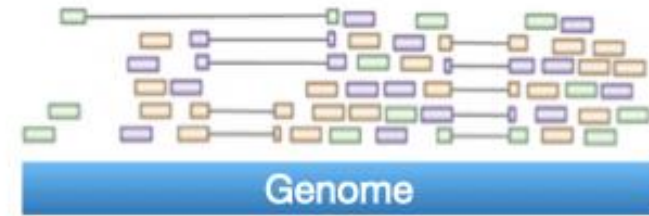
## Base Calling



## Quality Control



## Genome Alignment



## Variant Calling

Reference AACTACTGCT

Base Alteration

AACTAATGCT

Base Deletion

AACTA-TGCT

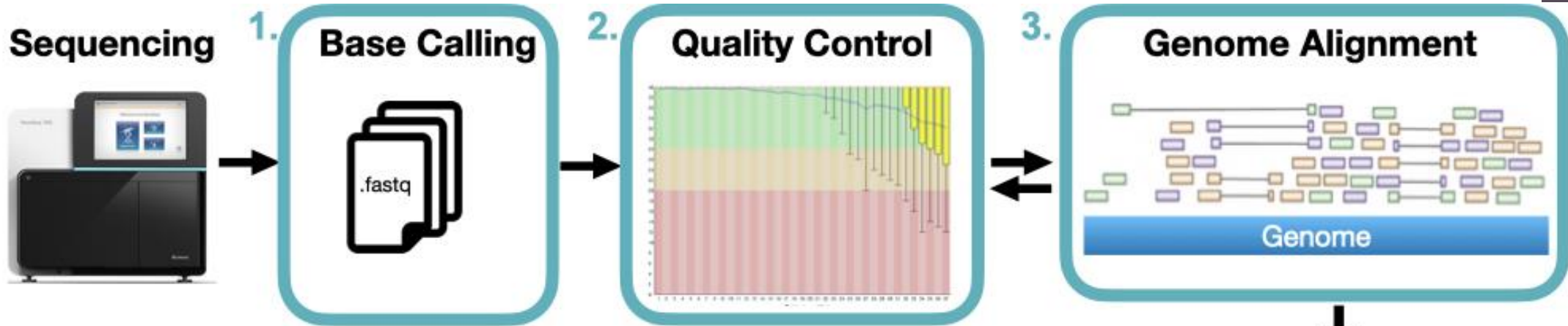
Base Insertion

AACTAACTGCT

## SNPs

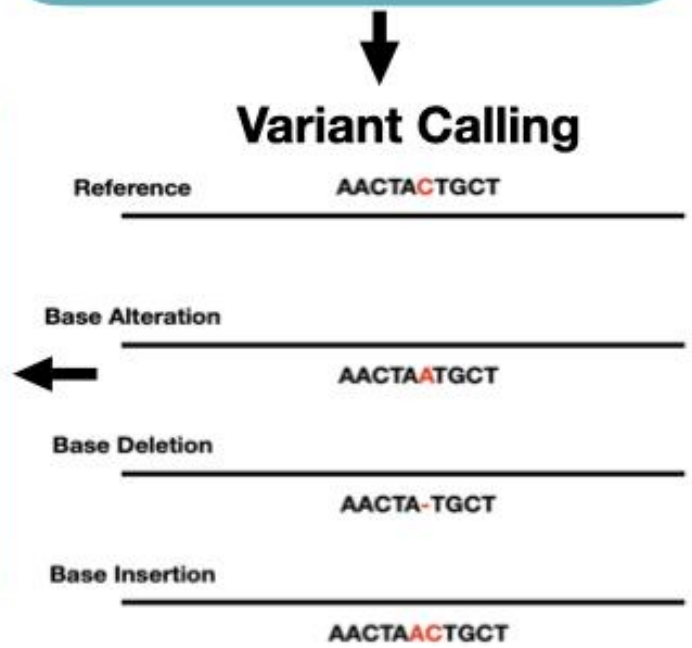
	Position Start	Position End	Ref	Variation	...
Chr1	183083734	183083734	G	-	
Chr3	13295559	18295559	G	A	
Chr4	25648977	25648977	-	T	
...	...	...			
Chr5	23477	23477	C	T	
ChrX	2019477	2019477	A	-	

# Whole Exome Sequencing: Παραδοτέα



Deliverables
1. FASTQ files (raw data)
2. Quality Metrics
3. BAM files (alignment data)
4. Variant Calling Files (VCF)

4. SNPs					
	Position Start	Position End	Ref	Variation	...
Chr1	183083734	183083734	G	-	
Chr3	13295559	18295559	G	A	
Chr4	25648977	25648977	-	T	
...	...	...			
Chr5	23477	23477	C	T	
ChrX	2019477	2019477	A	-	



# RNA Sequencing (QuantSeq & TruSeq): Ποή εργασίας

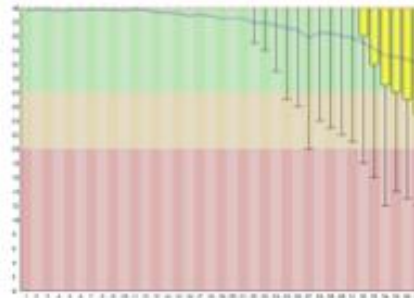
**Sequencing**



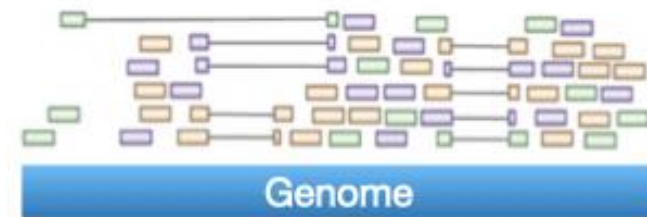
**Base Calling**



**Quality Control**



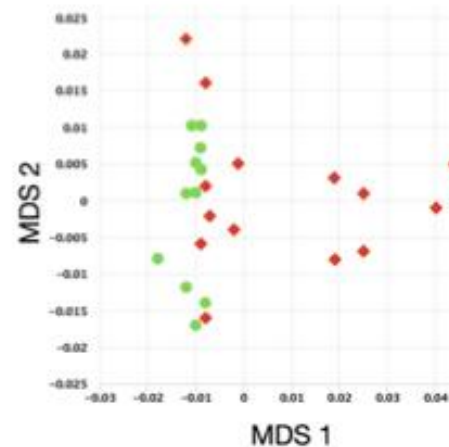
**Genome Alignment**



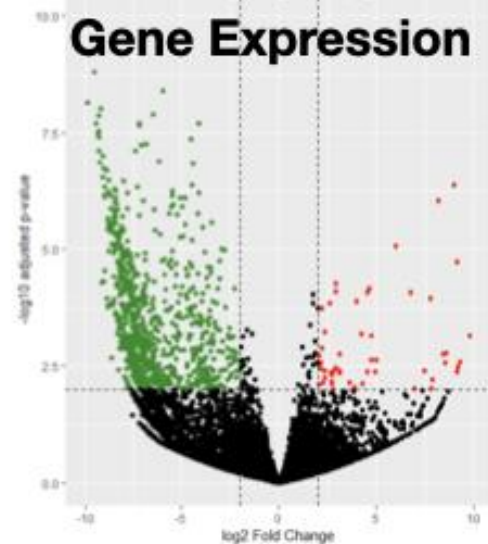
**Transcripts' Quantification**

	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**



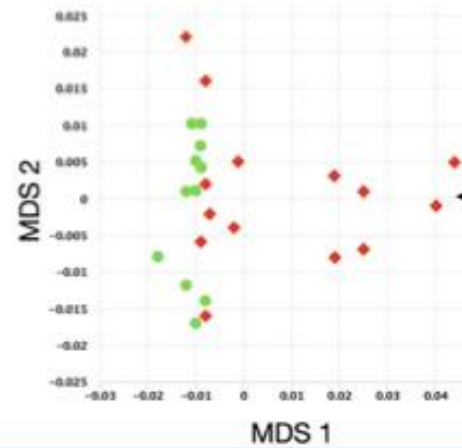
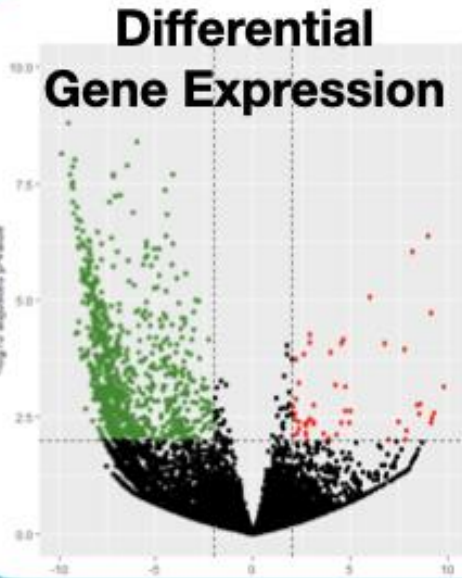
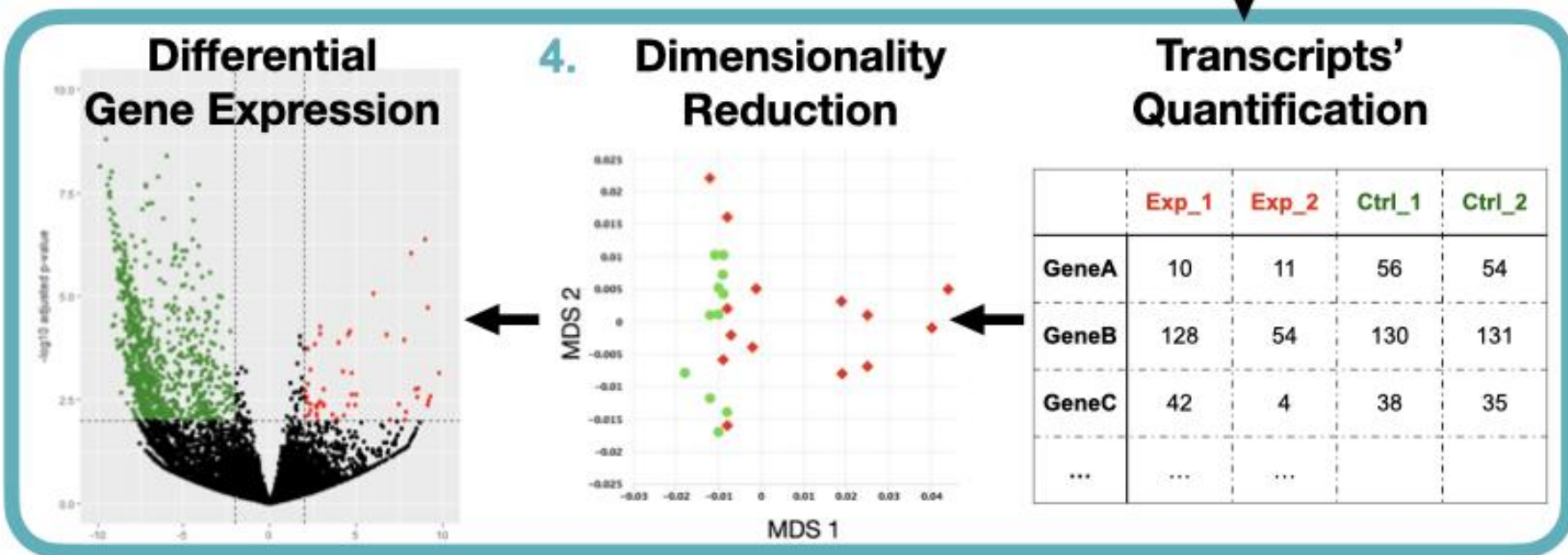
**Differential Gene Expression**



# RNA Sequencing (QuantSeq & TruSeq): Παραδοτέα



Deliverables
1. FASTQ files (raw data)
2. Quality Metrics
3. BAM files (alignment data)
4. MetaseqR2 Report



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

# Επιπλέον παρεχόμενες υπηρεσίες

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pMedGR

- Γονιδιακή έκφραση σε επίπεδο μοναδιαίων κυττάρων (Single Cell RNAseq)
- Εκ νέου σύσταση ιικού γονιδιώματος (*De novo* assembly of SARS-CoV2 genome)

## Υπό ανάπτυξη

- Ανάλυση διαφορικής μεθυλίωσης (Differential DNA Methylation Analysis)
- Ανάλυση ανοσοκατακρημνισμένης χρωματίνης (ChIPSeq Analysis)