



# NEGEDIA Bioinformatics Services

NEGEDIA delivers advanced bioinformatics solutions for sequencing data analysis, helping clients extract meaningful insights. Our experts support every stage, from experimental design to interpretation, ensuring top analytical precision.

Our optimized pipelines follow industry gold standards, while our cloud-based infrastructure enables scalable, efficient, and cost-effective analyses with fast turnaround times.

We provide microbiome-wide ecosystem insights for health, biotech, agriculture, and environmental research, offering an in-depth view of microbial diversity, function, and community dynamics with unmatched accuracy.

## OUR SERVICE INCLUDES

- Experimental design
- Raw reads quality check included in all our services
- Advanced analysis (II and III level)
- Custom analysis
- Data interpretation Support
- Long-term data storage options

## DATA ACCESS & STORAGE OPTIONS


- **Included in the service cost**
  - FastQ quality check
  - Free cloud transfer (Google Cloud)
  - Direct download via signed link – free for the first time
- On requests**
- Additional sequencing data download after the first week (NGD1040R)
  - Sequencing data storage, up to 1TB/year (NGD1050R)


## CUSTOM ANALYSIS

- **Dedicated analyses for RNA and DNA sequencing** (e.g., analysis of RNA from non-reference genomes, differentially expressed gene cluster analysis, submission to public databases such as GEO/SRA).
- **Experimental design consulting** and customized solutions tailored to the specific needs of our clients' projects.
- **Pipeline development:** all our solutions are cloud-based, GDPR-compliant, and ensure reproducible results.



<b>RNA II LEVEL ANALYSIS</b>	<b>NGD0900R</b> Count Analysis standard depth (up to 20M)	<b>NGD0901R</b> Count Analysis medium dept (up to 60M)	<b>NGD0902R</b> Count Analysis high depth (> 60M)	<b>NGD0920R</b> Single Cell Gene Expression
	Cleaning, Mapping on the genome, Count per gene. <b>Deliverables:</b> BAM files and Raw Count Matrix per gene. <b>TAT:</b> working days			Cell ranger count output <b>Deliverables:</b> BAM files, Count matrices (csv file), Cluster counts (csv file), t-SNE Projection plots <b>TAT:</b> 15 working days
<b>RNA III LEVEL ANALYSIS</b>	<b>NGD0910R</b> DEGs (for 8 samples)		<b>NGD0921R</b> RNA Single Cell Gene Expression	<b>NGD0950R</b> RNA Custom
	Normalization: Quality check, DEGs calling, Pathways enrichment. <b>Deliverables:</b> Normalized count matrix, DEGs expression, Pathways results. <b>TAT:</b> 5 working days		Cluster Annotation, Cell cycle status, Cell Type, Proportion Analysis. <b>Deliverables:</b> Annotated umap. <b>TAT:</b> 5 working days	Upon request
<b>RNA FULL ANALYSIS</b>	<b>NGD0930R</b> RNA Fusion	<b>NGD0940R</b> RNA miRNA	<b>NGD0925R</b> STOmics Stereo-seq	<b>NGD0922R</b> RNA Single Cell TCR
	Cleaning, Mapping on the genome, Fusion Calling <b>Deliverables:</b> BAM files and Fusions report. <b>TAT:</b> 10 working days	Cleaning, Mapping on the genome, Count per gene, Normalization, Quality check, miRNA DE calling, Pathways enrichment. <b>Deliverables:</b> BAM files and Raw Count Matrix per miRNA, Normalized count matrix, miRNA DE expression, Pathways enrichment results. <b>TAT:</b> 10 working days	Spatial gene expression matrix generation, Tissue segmentation and preliminary clustering. <b>Deliverables:</b> Processed spatial gene expression matrix, Plots, Clustering Annotation, Quality control report. <b>TAT:</b> 20 working days	Cleaning, Mapping. Counting per gene. Counting per VDJ, Cluster Annotation, <b>Deliverables:</b> BAM files, Count matrices, Cluster counts, t-SNE Projection plots, Annotated umap. <b>TAT:</b> 15 working days

					
<b>DNA II LEVEL ANALYSIS</b>	<b>NGD096R</b> Exome Variants Calling	<b>NGD0961R</b> Exome 200X Variants Calling	<b>NGD0970R</b> Genome Variants Calling	<b>NGD0971R</b> Genome 50X Variants Calling	<b>NGD0990R</b> Nanopore WGS Mapping
	Cleaning, Mapping on the genome, Deduplication, Alignment refinement, Variants calling <b>Deliverables:</b> BAM files, raw and annotated VCF files <b>TAT:</b> 10 working days				Mapping to the Reference Genome, Calculating Read Depth <b>Deliverables:</b> Alignment report for all samples, Reads Alignment Stats, Aligned BAM and BAI file <b>TAT:</b> 15 working days
<b>DNA III LEVEL ANALYSIS</b>	<b>NGD0980R</b> Variants Classification Germinal	<b>NGD0981R</b> RNA Variants Classification Somatic		<b>NGD0991R</b> Nanopore WGS Methylation Peak Calling	
	Variants Classification <b>Deliverables:</b> Table with variants classified by Varsome (ACGM guidelines) <b>TAT:</b> 5 working days	Variants Classification <b>Deliverables:</b> Table with variants classified by Varsome (ACGM guidelines) <b>TAT:</b> 5 working days		Sample Probability Computation, Base Modification Calling, Haplotype-Specific Analysis (optional), Validation <b>Deliverables:</b> Report of the alignment statistics, Modified bases BEDMethyl, Alignment file and file index <b>TAT:</b> 7 working days	
<b>OTHER ANALYSIS</b>	<b>NGD1000R</b> DNA Cut&Run/Cut&Tag	<b>NGD1001R</b> DNA ATAC-seq	<b>NGD1002R</b> DNA ChipSeq	<b>NGD1010R</b> DNA Custom (price per hour)	
	Mapping on genome, Differentially peaks calling, Differentially binding sites, Functional annotation <b>Deliverables:</b> Interactive plots and reports. <b>TAT:</b> 10 working days	Map chromatin accessibility across the genome, Identify potential regulatory elements and transcription factor binding sites, Differentially peaks calling. <b>Deliverables:</b> Interactive plots and reports. <b>TAT:</b> 10 working days	Mapping on genome, Differentially peaks calling, Differentially binding sites, Functional annotation <b>Deliverables:</b> Interactive plots and reports. <b>TAT:</b> 10 working days	Upon requests	

				
<b>METAGENOMIC ANALYSIS</b>	<b>NGD1020R</b> Metagenomics Shotgun Abundance II Level	<b>NGD1022R</b> Metagenomics Shotgun Abundance III Level	<b>NGD1030R</b> Whole Genome Shotgun without pathogens	<b>NGD1031R</b> Whole Genome Shotgun including pathogens
	Cleaning, Taxonomic, assignation and Relative Abundance. <b>Deliverables:</b> Taxonomic report (html and tsv file). <b>TAT:</b> 5 working days	Diversity within and between samples, Alpha-Beta diversity, Comparison among samples. <b>Deliverables:</b> Interactive plots and reports. <b>TAT:</b> 5 working days	Key microbiome health status pattern and functionality. <b>Deliverables:</b> Diet and Probiotic suggestion, comprehensive report. <b>TAT:</b> 5 working days	

NEGEDIA® is a cutting-edge NGS laboratory built on the Telethon Foundation's 30 years of expertise in rare genetic disease research. We are committed to making genomics accessible across the Life Sciences, providing high-precision sequencing and bioinformatics solutions for both research and clinical applications.

» **Clinical Services:** Supporting laboratories, clinics, and hospitals in leveraging NGS for clinical research and diagnostic advancements.

» **Research Services:** Supporting research institutes and universities at every stage, from experimental planning to advanced bioinformatics analysis.

For more information or to discuss a project, contact us at [info@negedia.com](mailto:info@negedia.com)

# GENOMICS AND BEYOND

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