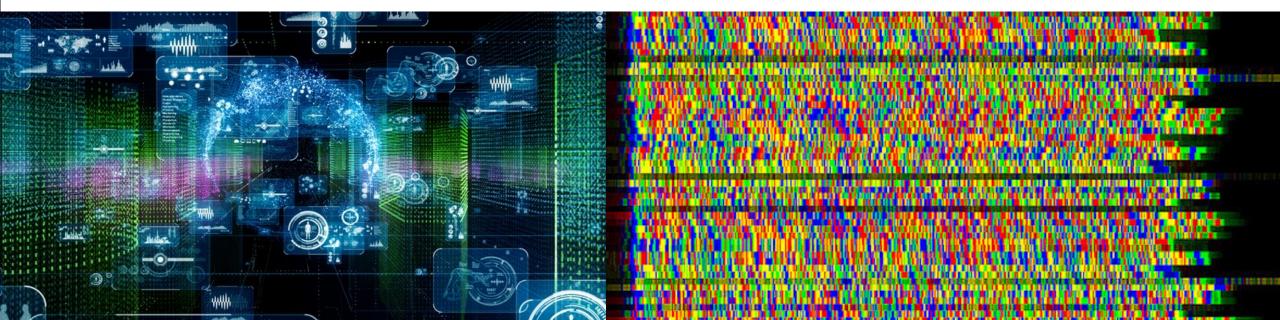




THE FUTURE IN HEALTH THROUGH INNOVATION





THE VALUES OF THE UMPHIULIU HATIEGANU GENOMICS CENTER

Creating an environment for a better quality of life through our values
Originate improvement in genomic medicine and bring them into the clinic
Commitment to offer high-quality genomic services for all the patients in need
Dedication to work for the improvement of genomic profiling to improve the standard of care

VISION

- Discoveries and innovation in genomic medicine to improve people's health in our region
- Extend the frontiers of science, continuously reinventing methods for disease understanding and exploring every source of innovation

MISSION

- Genomics innovative environment, through integrated research services, from the initial design to the final product
- Development of new products in the field of genomic medicine to discover new diagnostic strategies and innovative therapies
- Support for researchers, doctors, PhD students, undergraduate and master's students discovering and investigating the foundation of genomics and its applicability in human medicine







- 1. Processing of biological samples
- 2. Extraction and qualitative and quantitative quantification of nucleic acids (blood, serum, plasma, urine, tumor tissue, CSF, cells)
- 3. Discovery of new therapeutic compounds/testing therapeutic compounds in vitro models
- 4. Fluorescence and confocal microscopy/laser capture microdissection /cell painting
- 5. Flow cytometry
- 6. Microarray gene expression: complete coding, non-coding transcriptome analysis
- 7. Next-generation sequencing whole genome, exome sequencing, RNAseq, miRNAseq, lncRNAseq, commercial panels, custom panels
- 8. Quantitative analysis by real-time amplification
- 9. Bioinformatics
- 10. Gene therapy CRISPR
- 11. Pathological anatomy / immunohistochemistry / digital pathology / convolutional neural networks
- 12. Determination of water compounds / Screening mycotoxins in cereals/ Environmental metal species





PEER REVIEW PUBLICATIONS

PUBLICATION **2014**



TOTAL ISI PUBLICATIONS

TOTAL IF 237.458 AVERAGE IF 7.91

PUBLICATION 2015



TOTAL

TOTAL IF 109 AVERAGE IF 2.94

PUBLICATION **2016**



IOIAL

TOTAL IF 98.957 AVERAGE IF 4.12

PUBLICATION **2017**



TOTAL ISI PUBLICATIONS

TOTAL IF 153.477 AVERAGE IF 5.11

PUBLICATION 2018



TOTAL ISI PUBLICATIONS

TOTAL IF 203.809 AVERAGE IF 4.24

PUBLICATION 2019



IOIAL ISI PUBLICATIONS

TOTAL IF 234.19 AVERAGE IF 4.33

PUBLICATION 2020



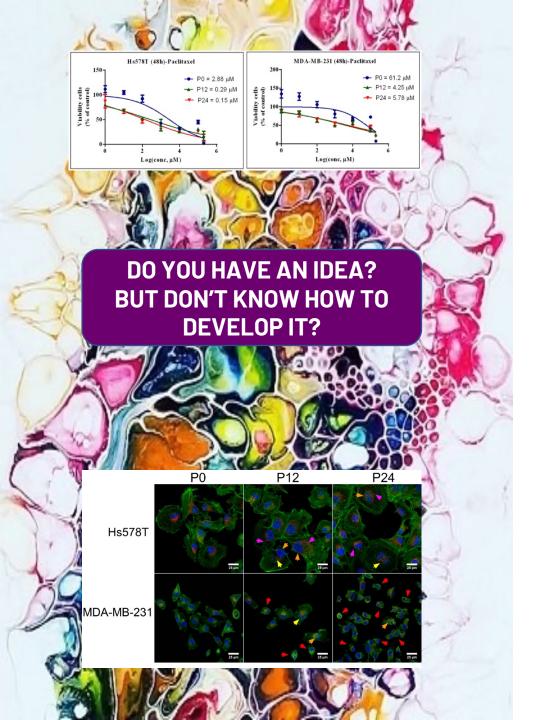
62

PUBLICATION 2021



TOTAL ISI PUBLICATIONS

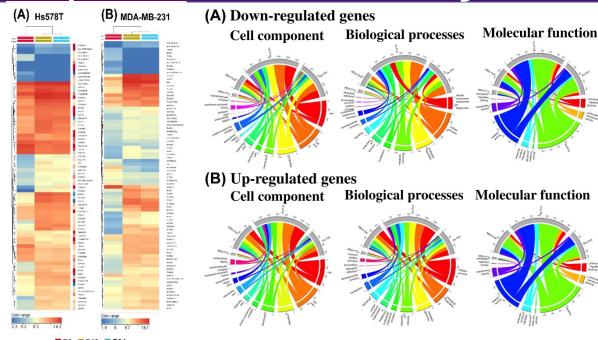
TOTAL IF 326.015 AVERAGE IF 6.27

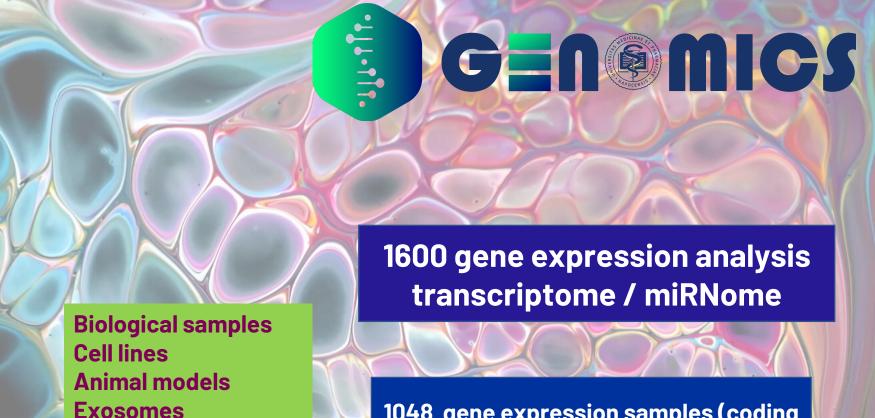




WHY US?

From concept - design - data analysis - interpretation of results - final product (patents, scientific articles, grants)





Animal models

Cell lines

1048 gene expression samples (coding genes and IncRNA)

552 gene expression samples (noncoding genes and miRNome)

Tumor tissue Normal tissue

- Bladder cancer
- Oral cancer
- Prostate cancer
- Glioblastoma
- Hematologic diseases
- Endometriosis
- Melanoma
- Breast cancer
- Lung cancer

Plasma samples from patients and normal subjects

- Colorectal cancer
- Bladder cancer
- Thyroid cancer



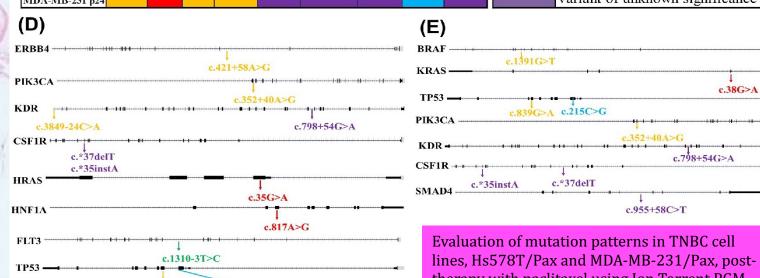


(A)

c.4732_4734delGTG

()												
	Mutation											
	c.421+58	c.352 40 A>G	c.3849-24	c.798+54	c.*37defT	c.*35instA	c.35 G>A	e.817 A>G	c.1310-3	c.469 G>T	e.215 C>G	c.4732 4734
Mutation	A>G ERBB4	PIK3CA	C>A KDR	G>A KDR	CSF1A	CSF1A	HRAS	HNF1A	T>C FLT3	TP53	TP53	delGTG NOTCH1
HS578T p0												
HS578T p12												
HS578T p24												
(B)	(C)											
	Mutation						drug response					

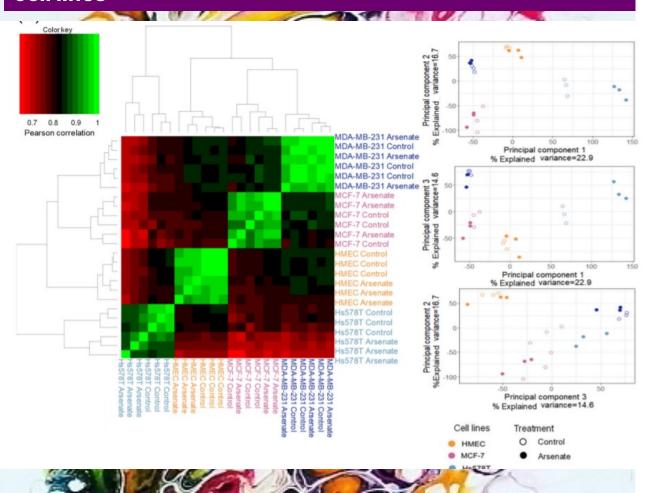
c.*37deIT c.*35inst benign Sample BRAF KRAS TP53 PIK3CA KDR TP53 SMAD4 CSF1A A CSF1A likely pathogenic MDA-MB-231 p0 pathogenic MDA-MB-231 p12 variant of unknown significance MDΛ-MB-231 p24



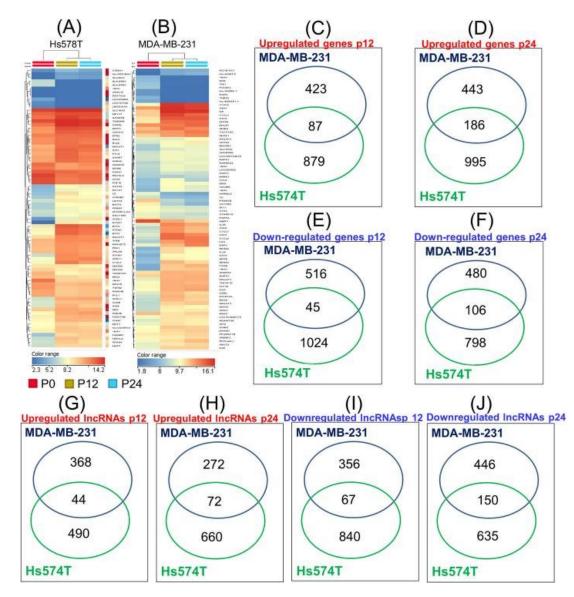
lines, Hs578T/Pax and MDA-MB-231/Pax, posttherapy with paclitaxel using Ion Torrent PGM Machine and Ion AmpliSeq Cancer Hotspot Panel Pool

E CENENT MICS

Arsenic activates specific mechanisms in breast cancer cell lines



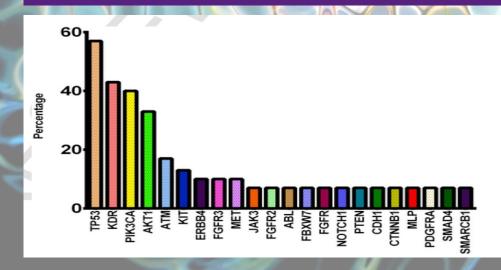
Activation of doxorubicin resistance mechanisms in triple-negative breast lines microarray study





Next generation sequencing

600 Cancer panel analysis
Patients
Cell lines
Animal models



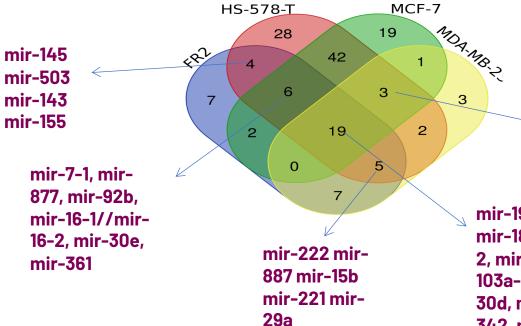
411 BRCA1/2

- 4			Committee of the Commit			
	Locus BRCA1	HGVS	Amino-acid	Mutation	Nb.	Percentage
1	chr17:41209079	c.5266dupC	p.Gln1777fs	Frameshift Insertion	11	36.67%
2	chr17:41234559	c.4218delG	p.Lys1406fs	Frameshift Deletion	1	3.33%
3	chr17:41276044	c.68_69delAG	p.Glu23fs	Frameshift Deletion	1	3.33%
4	chr17:41243941	c.3607C>T	p.Arg1203Ter	Nonsense	9	30%
5	chr17:41245861	c.1687C>T	p.Gln563Ter	Nonsense	2	6.67%
6	chr17:41258504	c.181T>G	p.Cys61Gly	Missense	4	13.34%
7	chr17:41226539	c.4485-1G>T		Splicesite	1	3.33%
8	chr17:41258472	c.212+1G>T		Splicesite	1	3.33%
				Total	30	100%



CENTRAL MICES

29 miRNA seq

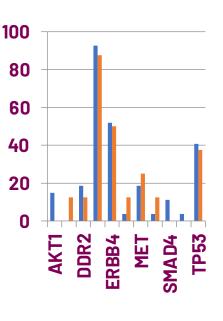


mir-23b mir-423 mir-24-2

mir-191, mir-301a, mir-181b-1//mir-181b-2, mir-103a-2//mir-103a-1, let-7b, mir-30d, mir-652, mir-342, mir-30a, mir-671, mir-425, mir-181b-2, mir-320a, mir-182 mir-181a-2//mir-181a-1, mir-30c-2//mir-30c-1, mir-454, mir-23a, mir-21

35

Lung and Colon Panel



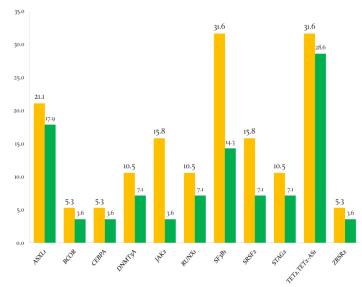
for dog samples

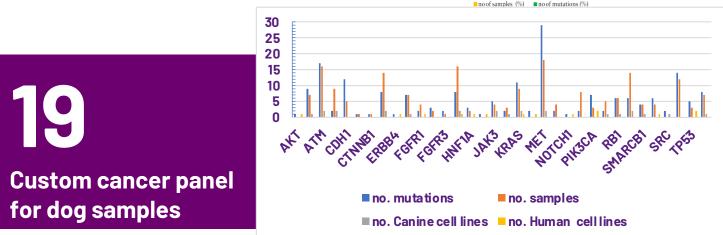
■ % cancer samples (n=27)

% pneumonia samples (n=8)

19

Oncomine Myeloid panel











HID NGS Panel GENE chr snp rs1490413 rs7520386 rs4847034 rs560681 rs10495407 rs891700 CHRM3-AS2 rs1413212 rs876724 rs1109037 LOC105373422 rs993934 CERKL rs12997453 rs907100 rs1357617 rs4364205 rs1872575 **QTRT2** rs1355366 rs6444724 **ATP13A4** rs2046361 PALLD rs6811238 rs1979255 rs717302 rs159606 rs7704770 PWWP2A rs251934 **ADAMTS2** rs338882 rs13218440 HIVEP1 rs214955 SYNE1

Comprehensive cancer panel 400 genes

Locus	Genotype	Ref	Genes	% Frequency Strand	Exon	Transcript	Coding
chr1:144866643	G/A	G	PDE4DIP	22.96 -		NM_0011988 3434.3	c.5599C>T
chr1:144879090	T/C	т	PDE4DIP	38 -		NM_0011988 2734.3	c.4360A>G
chr1:144879264	G/G	A	PDE4DIP	100 -		NM_0011988 2734.3	c.4186T>C
chr1:144909956	G/A	G	PDE4DIP	63.64 -		NM_0011988 1734.3	c.2232C>T
chr1:144918957	T/A	т	PDE4DIP	42.18 -		NM_0011988 1034.3	c.1229A>T
chr1:144931087	T/C	т	PDE4DIP	36.36 -	intron 5	NM_0011988 34.3	c.637- 7266A>G
chr1:144931392	G/A	G	PDE4DIP	43.94 -	intron 8	NM_0011988 34.3	c.637- 7571C>T
chr1:144994658	C/A	С	PDE4DIP	22.39 -		NM_0011988 134.3	c.74G>T

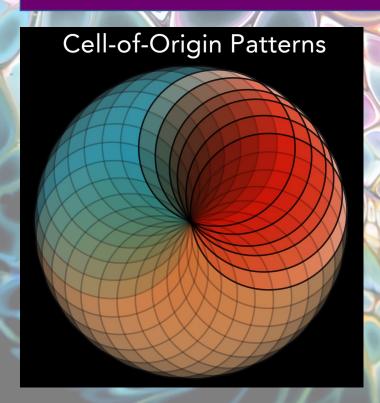
Custom NGS Panels for different diseases (COVID, Lymphoma, Hereditary Telangiectasia Type 2, etc)

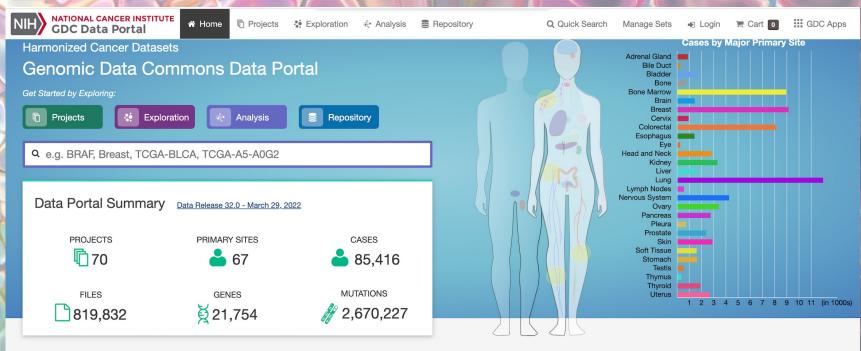
Locus Hg 19	Genes	Coding
chr1:214524509	PTPN14	c.*6756T>C
chr1:214524613	PTPN14	c.*6652A>G
chr1:214524620	PTPN14	c.*6645insG
chr1:214524622	PTPN14	c.*6643T>C
chr1:214524754	PTPN14	c.*6511G>A
chr1:214525316	PTPN14	c.*5949T>G
chr1:214525603	PTPN14	c.*5662T>C
chr1:214526332	PTPN14	c.*4933T>C
chr1:214526363	PTPN14	c.*4902A>T
chr1:214526612	PTPN14	c.*4653A>C
chr1:214526964	PTPN14	c.*4301A>G
chr1:214527252	PTPN14	c.*4013C>T
chr1:214529147	PTPN14	c.*2118C>G



Bioinformatic analysis 20.000

TCGA, MIRNA, GENESPRING,GENE EXPRESSION, FEATURE EXTRACTION, STRING, Ingenuity Pathway Analysis, MIRBASE, MRNA-MIRNA NETWORK MIRTARGETLINK, VARIANT CALLER

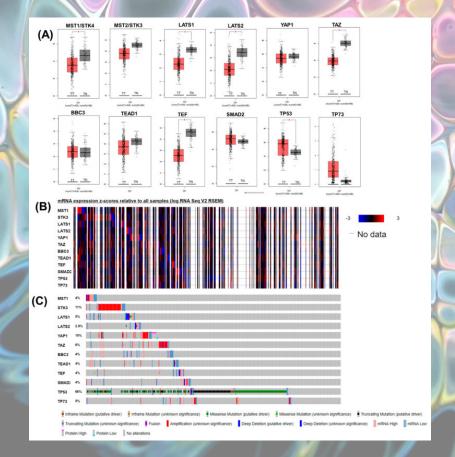


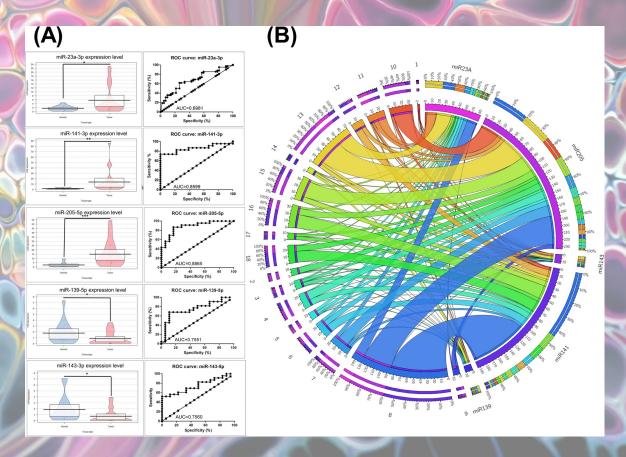


E GENERALES

qRT-PCR

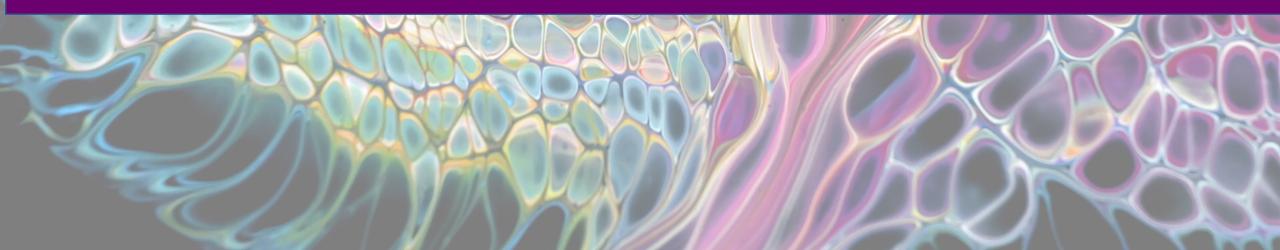
24.000 gene expression tests, miRNA, IncRNA Cells, exosomes, tissues, serum, plasma, blood, urine samples







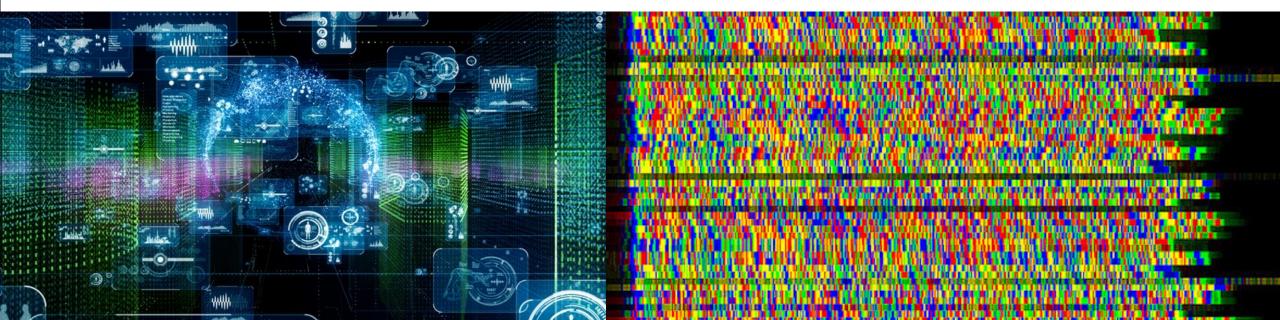
qRT-PCR 60.000 tests done for SARS-CoV-2 diagnosis

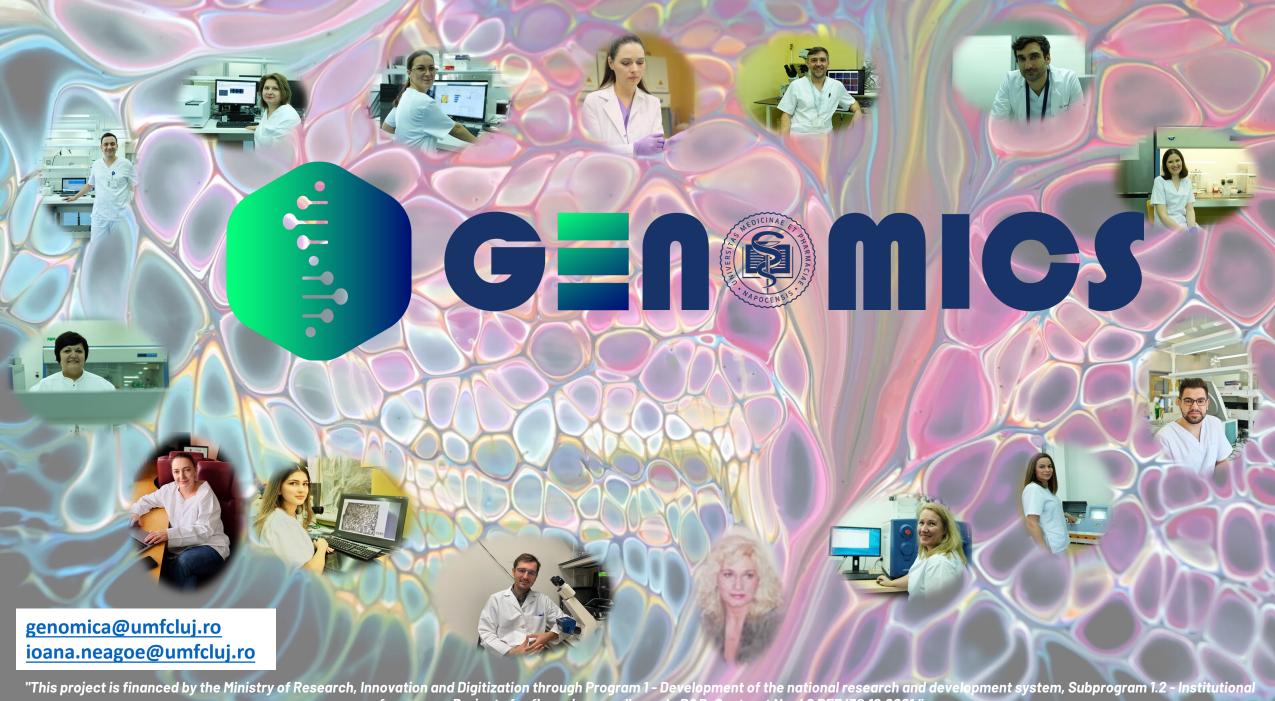






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