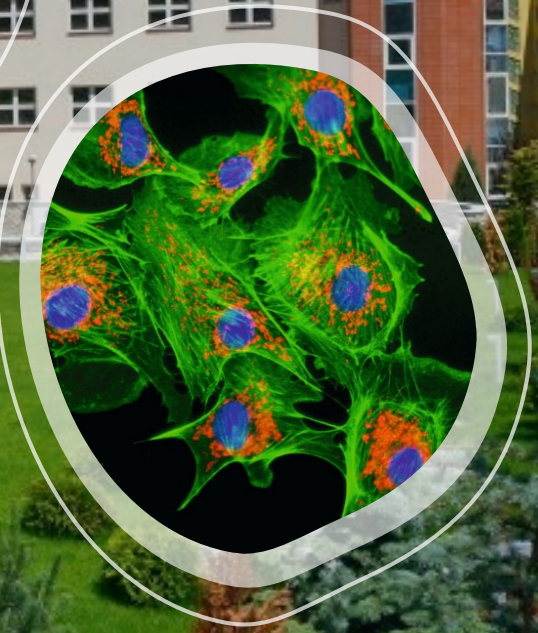
Iceland Liechtenstein Norway grants

RESEARCH CENTER FOR FUNCTIONAL GENOMICS, BIOMEDICINE AND TRANSLATIONAL MEDICINE



GENOMICS

THE VALUES OF THE UMPh IULIU 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

GENOMICS

PUBLICATION 2014

TOTAL ISI PUBLICATIONS **30**

TOTAL IF 237.458
AVERAGE IF 7.91

PUBLICATION 2015

TOTAL ISI PUBLICATIONS **37**

TOTAL IF 109
AVERAGE IF 2.94

PUBLICATION 2016

TOTAL ISI PUBLICATIONS **24**

TOTAL IF 98.957
AVERAGE IF 4.12

PUBLICATION 2017

TOTAL ISI PUBLICATIONS **30**

TOTAL IF 153.477
AVERAGE IF 5.11

PUBLICATION 2018

TOTAL ISI PUBLICATIONS **48**

TOTAL IF 203.809
AVERAGE IF 4.24

PUBLICATION 2019

TOTAL ISI PUBLICATIONS **54**

TOTAL IF 234.19
AVERAGE IF 4.33

PUBLICATION 2020

TOTAL ISI PUBLICATIONS **62**

TOTAL IF 267.439
AVERAGE IF 4.31

PUBLICATION 2021

TOTAL ISI PUBLICATIONS **52**

TOTAL IF 326.015
AVERAGE IF 6.27

PUBLICATION 2022

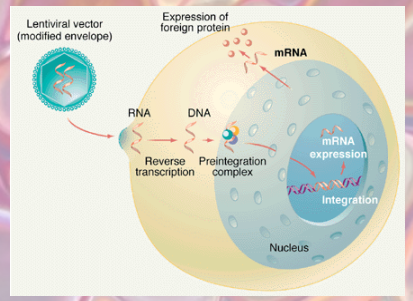
TOTAL ISI PUBLICATIONS **48**

TOTAL IF 318.771
AVERAGE IF 6.641

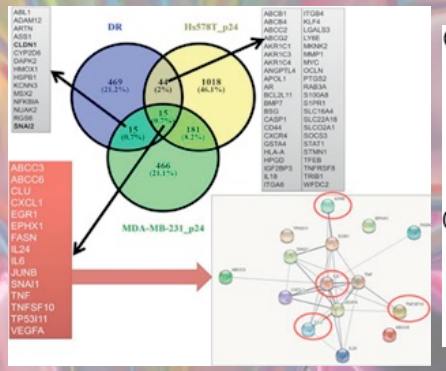
GENOMICS TEAM TOP 2% MOST CITED AUTHORS WORLDWIDE IN 2020, 2021

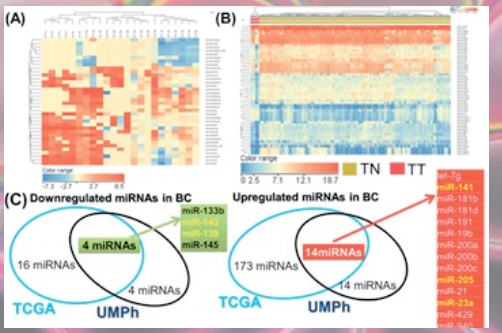
GENOMICS

1. Genetic engineering and biotherapies

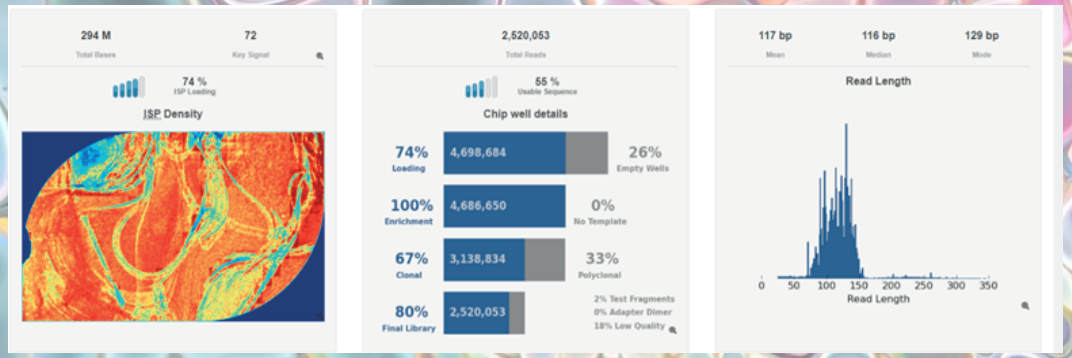


3. Microarray

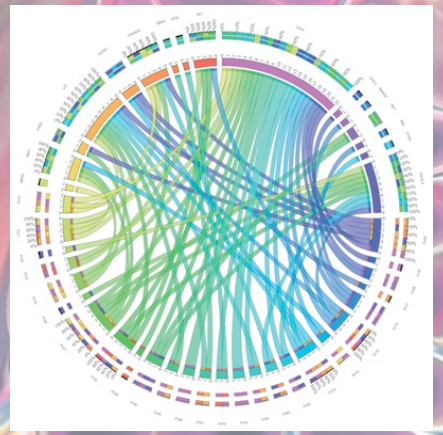




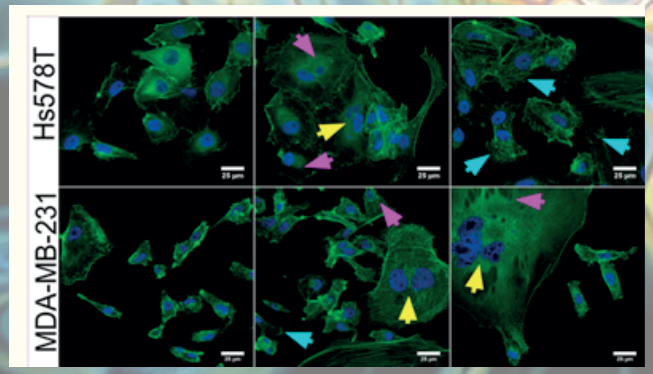
2. NGS



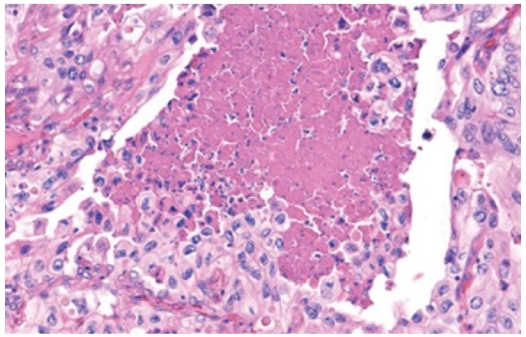
4. Bioinformatics

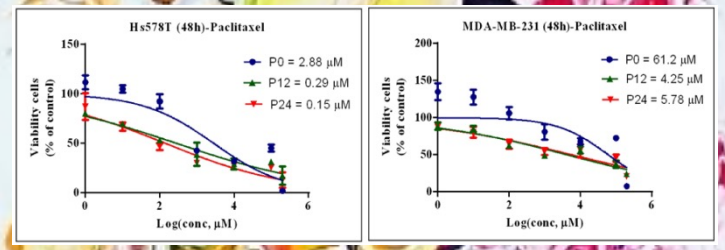


5. Cell cultures

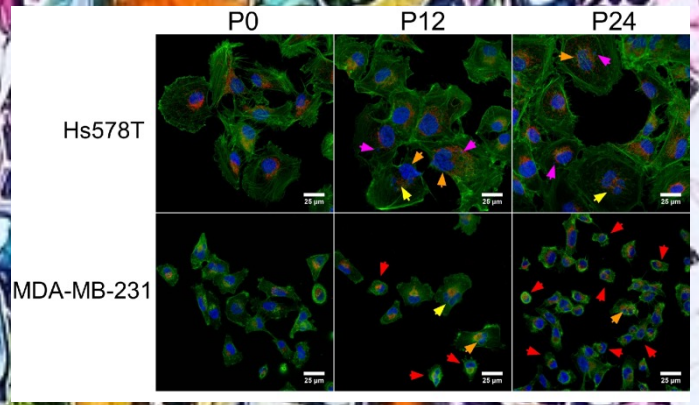


6. Translational pathology



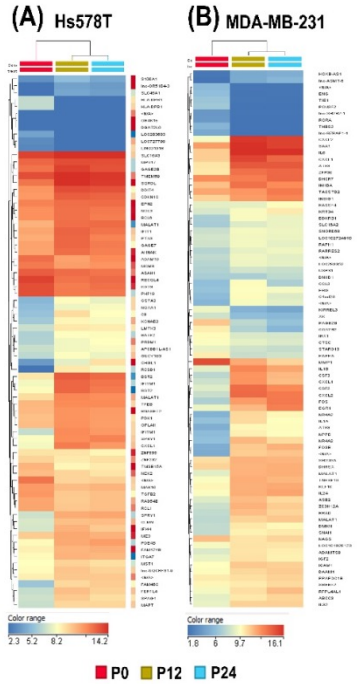


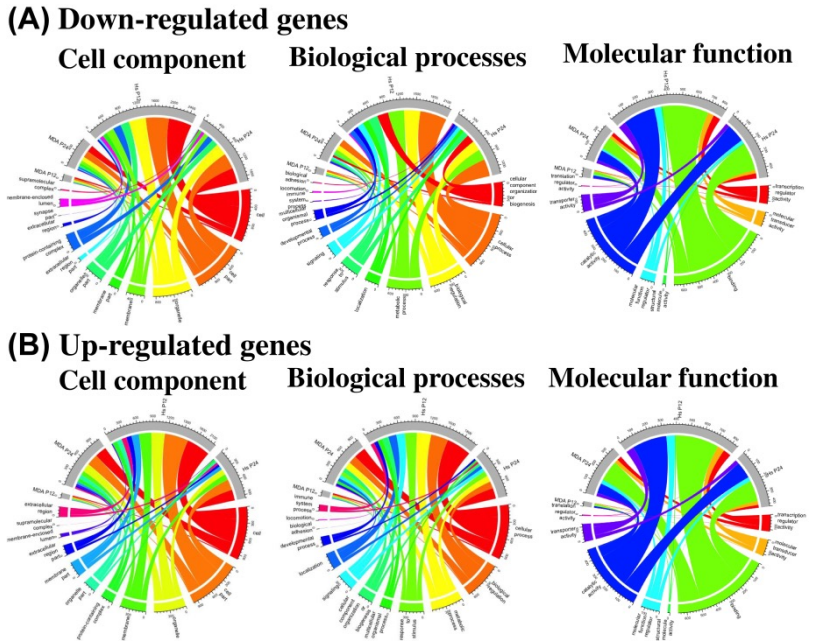
DO YOU HAVE AN IDEA? BUT DON'T KNOW HOW TO DEVELOP IT?



WHY US?

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





GENOMICS

Biological samples
Cell lines
Animal models
Exosomes

Animal models
Cell lines

1600 gene expression analysis transcriptome / miRNome

1048 gene expression samples (coding genes and lncRNA)

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

GENOMICS

(A)

Mutation	c.421↑58 A>G ERBB4	c.352↑40 A>G PIK3CA	c.3849-24 C>A KDR	c.798↑54 G>A KDR	c.*37delT CSF1A	c.*35instA CSF1A	c.35 G>A HRAS	c.817 A>G HNF1A	c.1310-3 T>C FLT3	c.469 G>T TP53	c.215 C>G TP53	c.4732_4734 delGTG NOTCH1
HS578T p0												
HS578T p12												
HS578T p24												

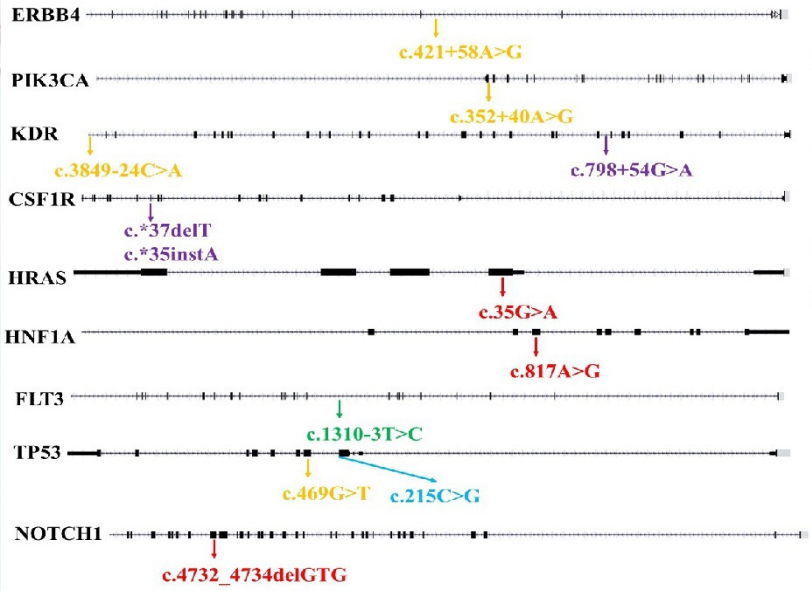
(B)

Sample	G>T BRAF	G>A KRAS	G>A TP53	A>G PIK3CA	G>A KDR	c.*37delT CSF1A	c.*35inst A CSF1A	C>G TP53	C>T SMAD4
MDA-MB-231 p0									
MDA-MB-231 p12									
MDA-MB-231 p24									

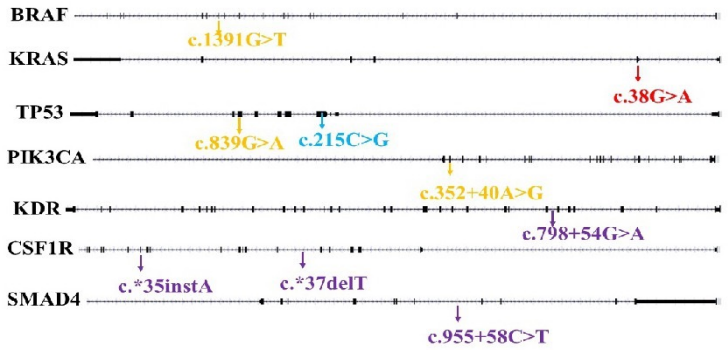
(C)

- drug response
- benign
- likely pathogenic
- pathogenic
- variant of unknown significance

(D)

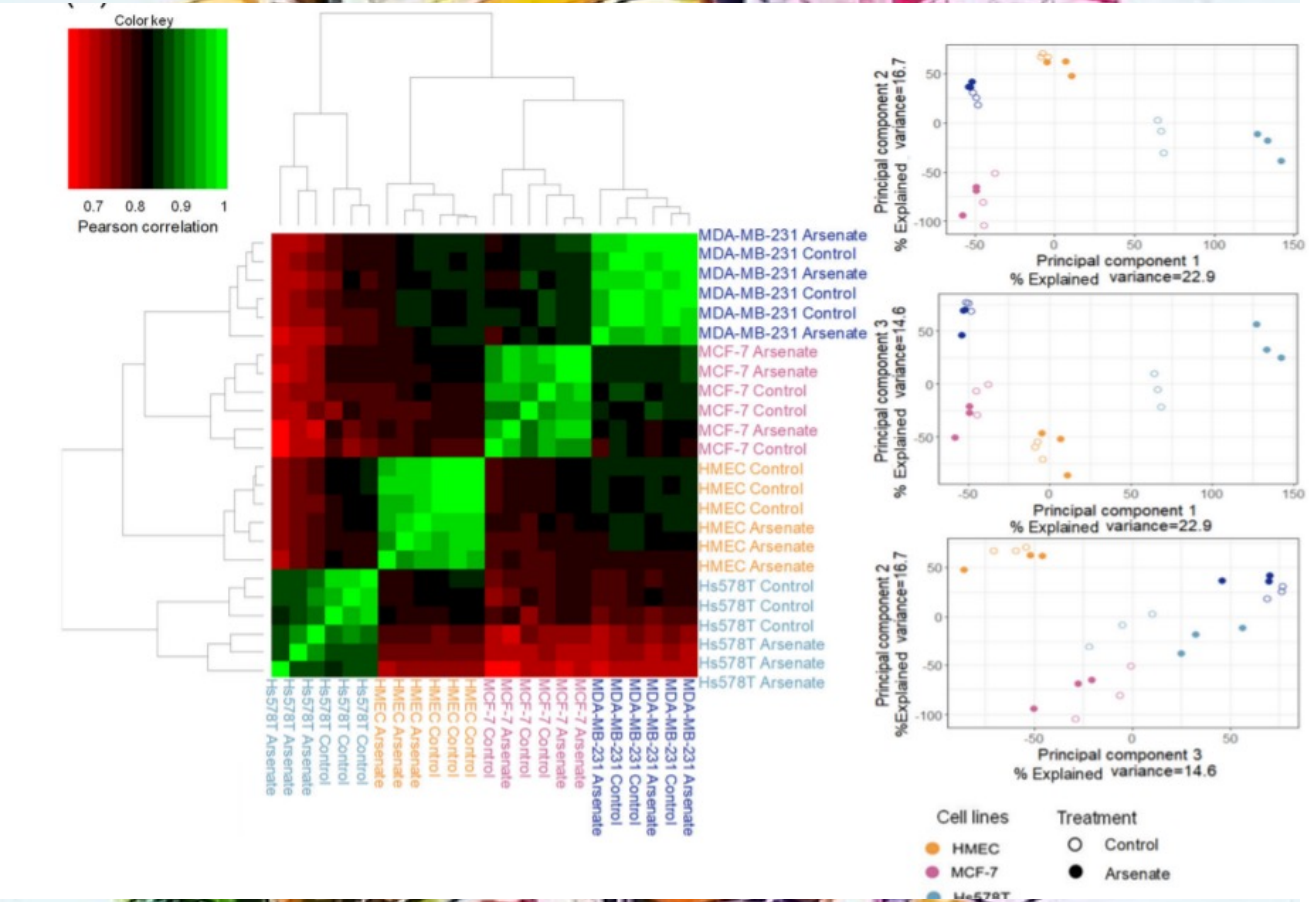


(E)

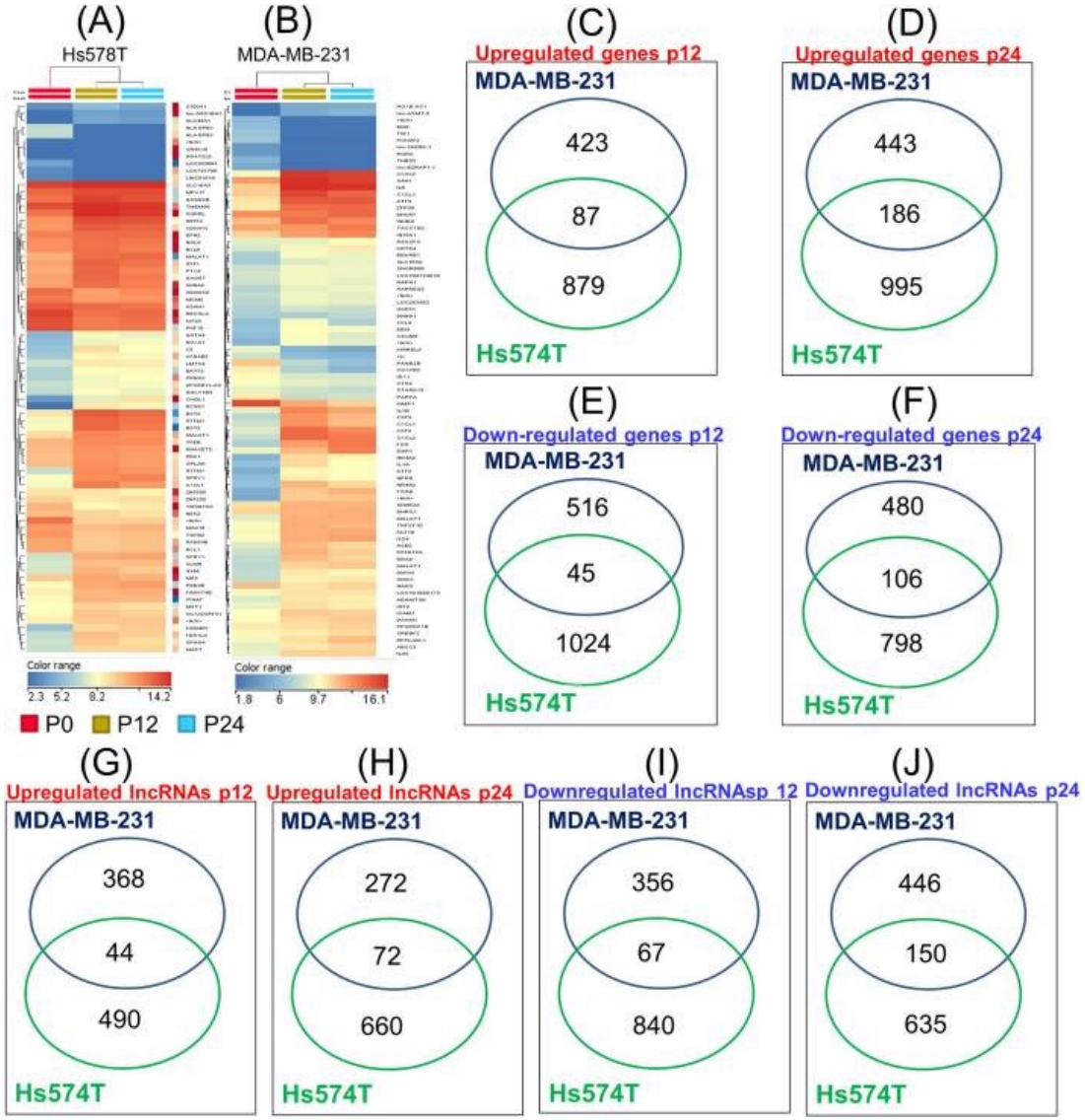


Evaluation of mutation patterns in TNBC cell lines, Hs578T/Pax and MDA-MB-231/Pax, post-therapy with paclitaxel using Ion Torrent PGM Machine and Ion AmpliSeq Cancer Hotspot Panel Pool

Arsenic activates specific mechanisms in breast cancer cell lines

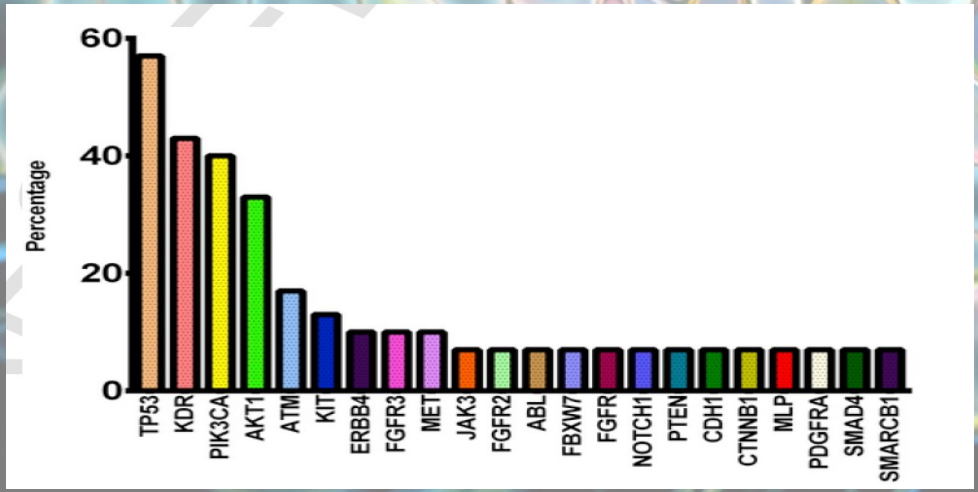


Activation of doxorubicin resistance mechanisms in triple-negative breast lines - microarray study



Next generation sequencing

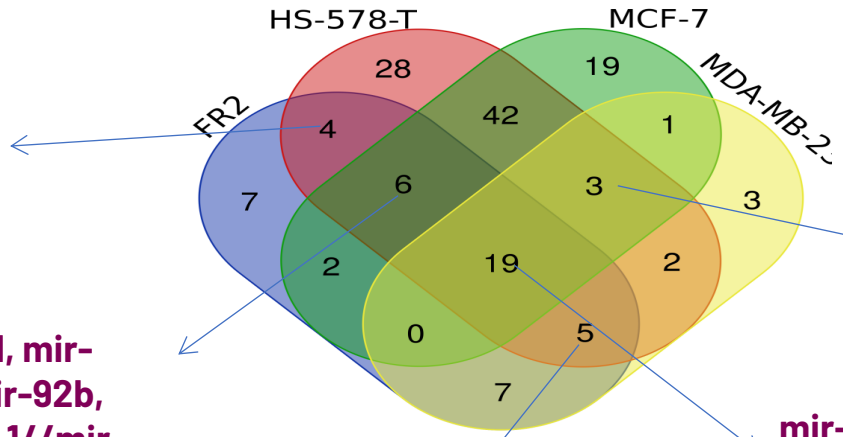
600 Cancer panel analysis
Patients
Cell lines
Animal models



411
BRCA1/2

	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%

29 miRNA seq



mir-145
mir-503
mir-143
mir-155

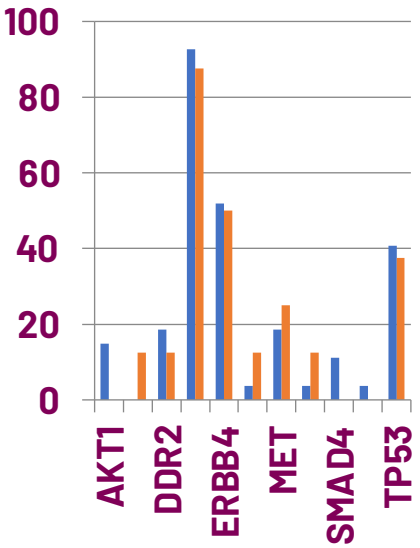
mir-7-1, mir-877, mir-92b, mir-16-1//mir-16-2, mir-30e, mir-361

mir-222 mir-887 mir-15b mir-221 mir-29a

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

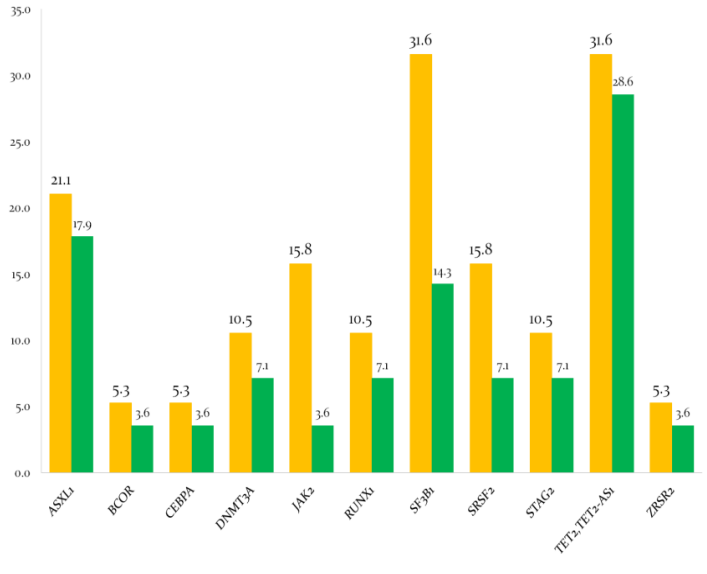
35
Lung and Colon Panel

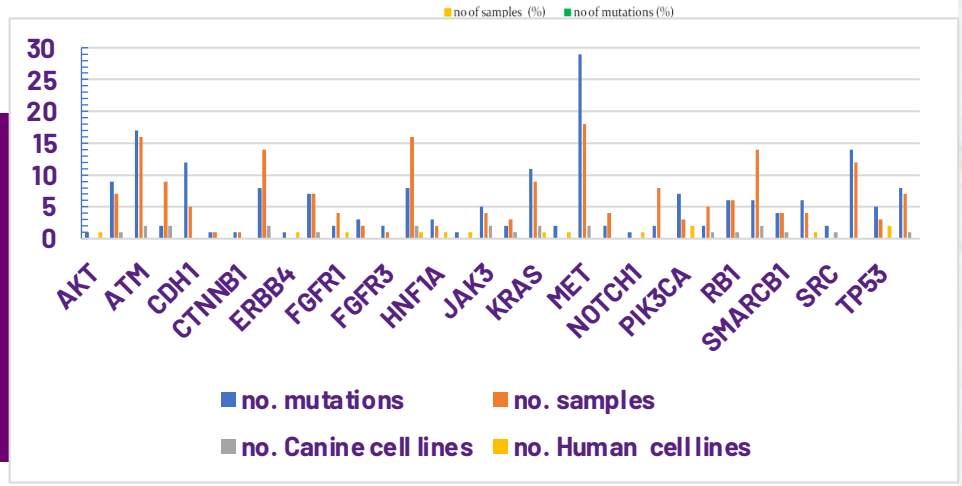


% cancer samples (n=27)

% pneumonia samples (n=8)

19
Oncomine Myeloid panel





19
Custom cancer panel for dog samples

HID NGS Panel

snp	GENE	chr
rs1490413		1
rs7520386		1
rs4847034		1
rs560681	LY9	1
rs10495407		1
rs891700	CHRM3-AS2	1
rs1413212		1
rs876724		2
rs1109037	LOC105373422	2
rs993934		2
rs12997453	CERKL	2
rs907100		2
rs1357617		3
rs4364205		3
rs1872575	QTRT2	3
rs1355366		3
rs6444724	ATP13A4	3
rs2046361		4
rs6811238	PALLD	4
rs1979255		4
rs717302		5
rs159606		5
rs7704770	PWWP2A	5
rs251934		5
rs338882	ADAMTS2	5
rs13218440	HIVEP1	6
rs214955	SYNE1	6

Comprehensive cancer panel 400 genes

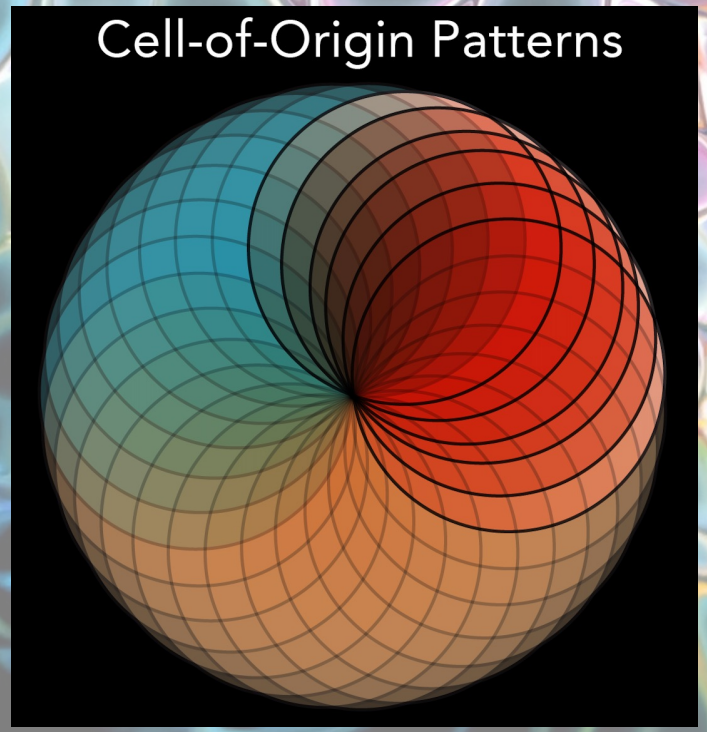
Locus	Genotype	Ref	Genes	% Frequency	Strand	Exon	Transcript	Coding
chr1:144866643	G/A	G	PDE4DIP	22.96	-		NM_0011988 3434.3	c.5599C>T
chr1:144879090	T/C	T	PDE4DIP	38	-		NM_0011988 2734.3	c.4360A>G
chr1:144879264	G/G	A	PDE4DIP	100	-		NM_0011988 2734.3	c.4186T>C
chr1:144909956	G/A	G	PDE4DIP	63.64	-		NM_0011988 1734.3	c.2232C>T
chr1:144918957	T/A	T	PDE4DIP	42.18	-		NM_0011988 1034.3	c.1229A>T
chr1:144931087	T/C	T	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	C	PDE4DIP	22.39	-		NM_0011988 134.3	c.74G>T

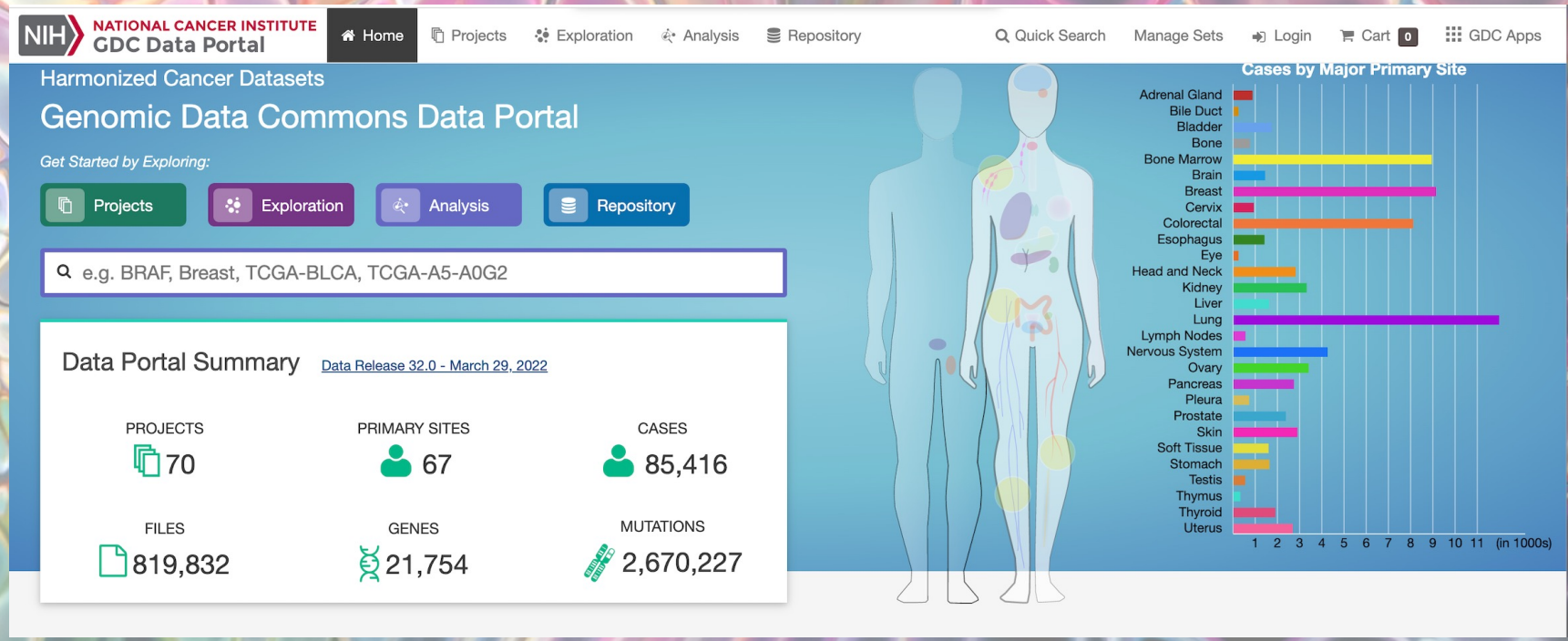
Custom NGS Panels for different diseases (COVID, Lymphoma, Hereditary Telangiectasia Type 2, etc)

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

GENOMICS

Bioinformatic analysis
20.000
TCGA, MIRNA, GENESPRING,GENE EXPRESSION, FEATURE EXTRACTION, STRING, Ingenuity Pathway Analysis, MIRBASE, MRNA-MIRNA NETWORK MIRTARGETLINK, VARIANT CALLER

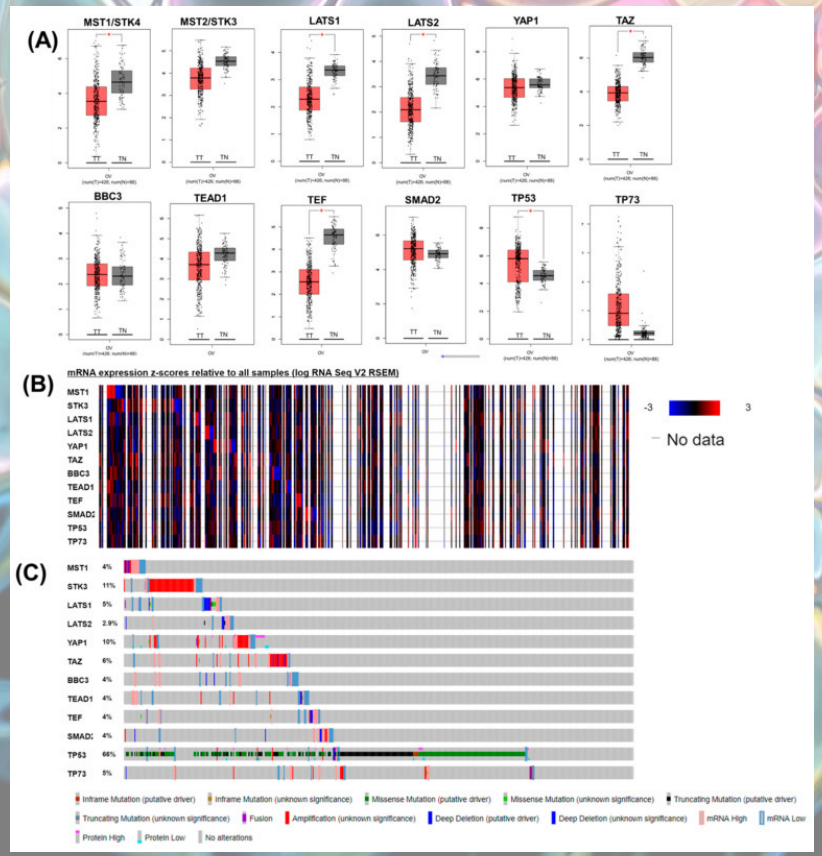


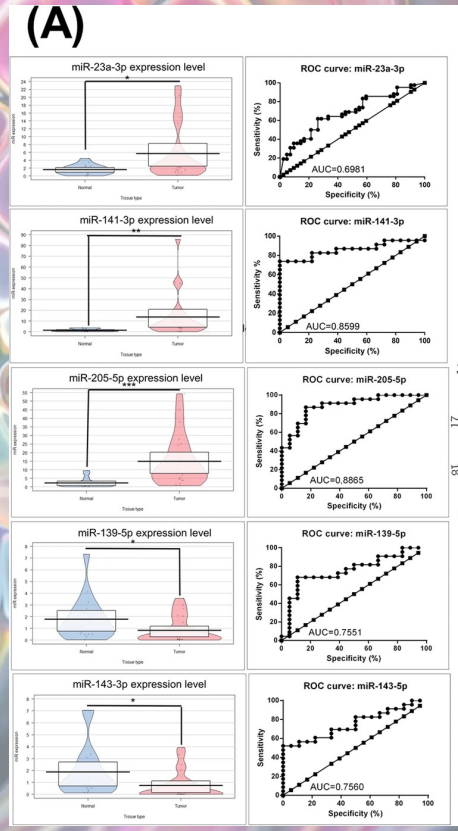


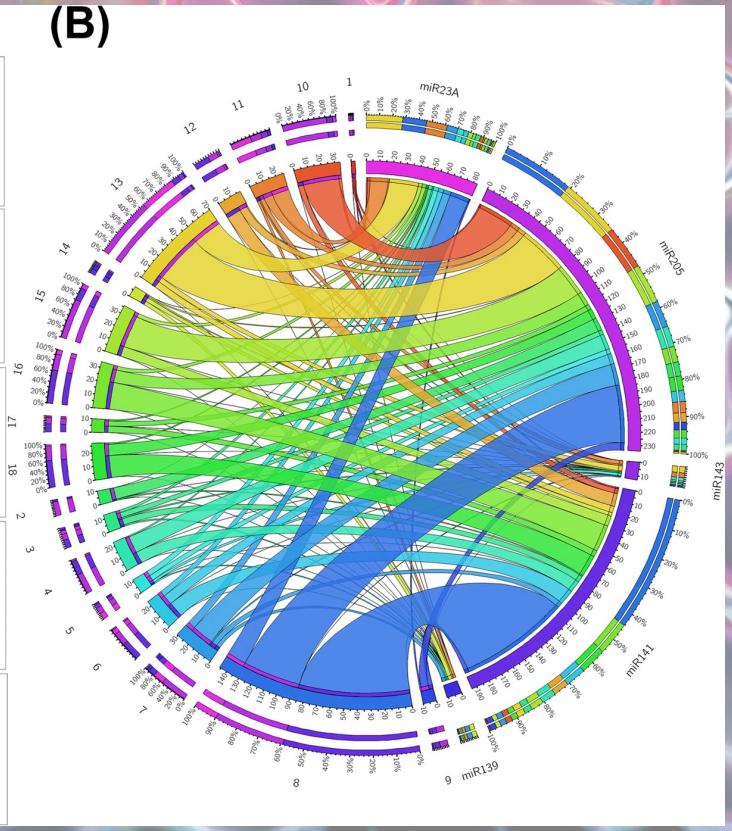
qRT-PCR

24.000 gene expression tests, miRNA, lncRNA

Cells, exosomes, tissues, serum, plasma, blood, urine samples

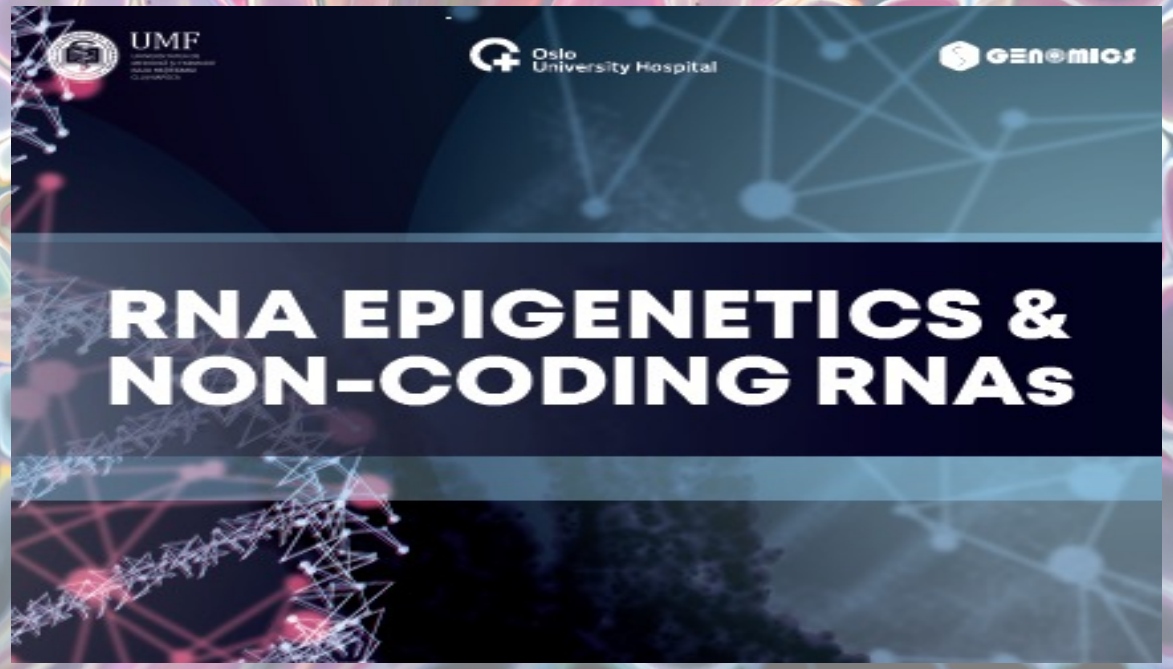


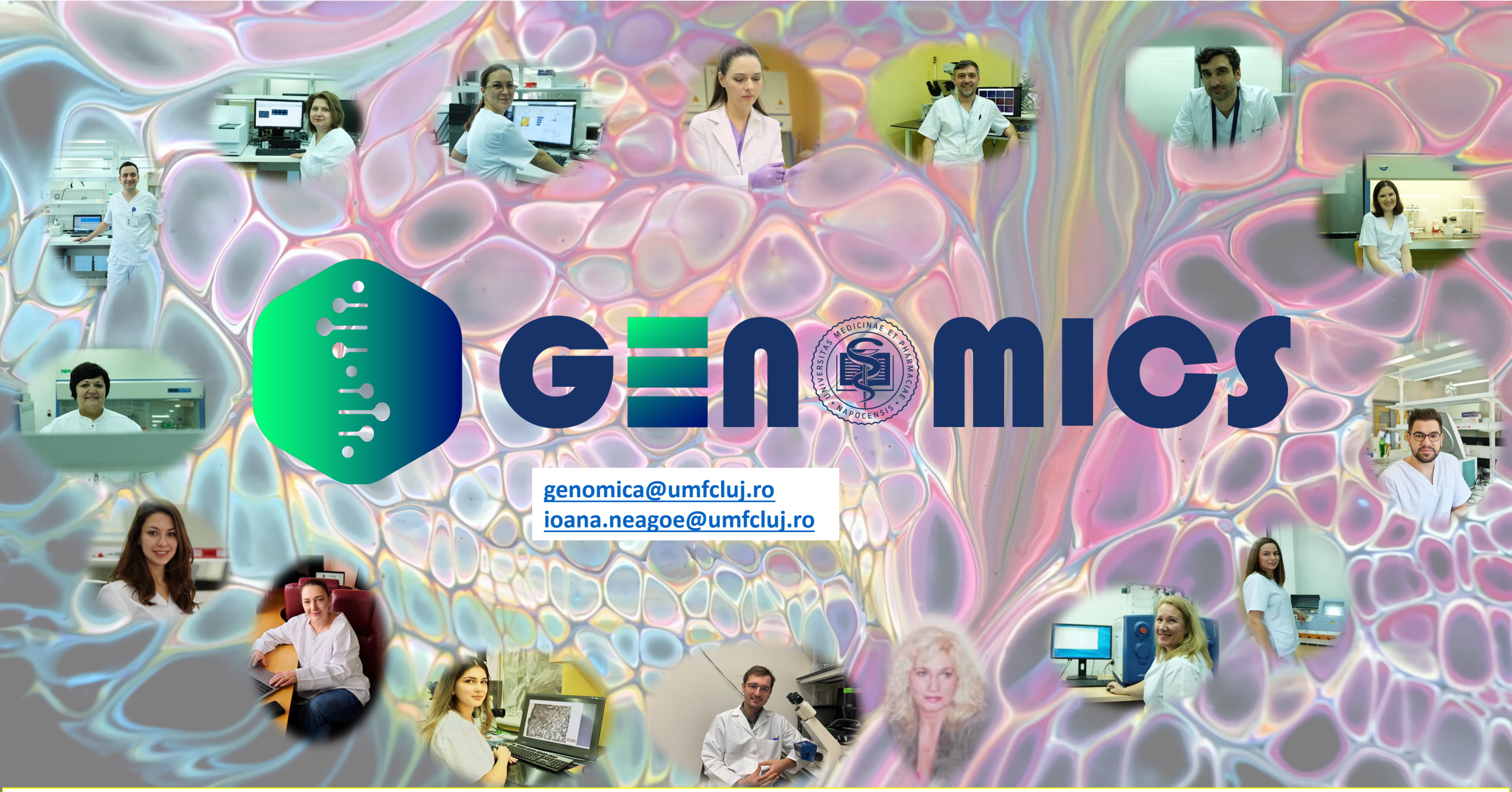




qRT-PCR
60.000 tests done for SARS-CoV-2 diagnosis

Volunteering programs, InternshipsTrainings, Workshops, Mentoring the next generation of scientists.


UMF
Oslo University Hospital
GENOMICS
RNA EPIGENETICS &
NON-CODING RNAs



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