pMedGR

The Greek Research Infrastructure for Personalized Medicine

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Fast Forward in Biomedical Innovation!

- New Biology – New momentum

- New phase in Biomedical Innovation

- New opportunities for academic research

Can we succeed?

- Legal / Regulatory frameworks suitable for Research

- Foster Immediate Launching and Use of New Technologies

- Motivate top level personnel for recruitment

pMedGR: The Greek Research Infrastructure for Personalized Medicine

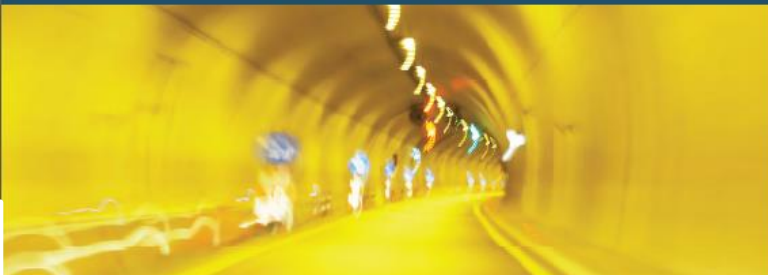Proteomics   Genomics   CyTOF   Single Cell Analysis   Bioinformatics

National Roadmap for Research Infrastructures

2014

The Greek Research Infrastructure for Personalised Medicine (pMedGR)

Following recent technological breakthroughs, such as rapid sequencing of the human genome, the concept of personalised medicine/health has become key in understanding, classifying, preventing and treating human disease. Capitalising on top clinical and basic research teams, the pMedGR infrastructure aims to support research towards patient stratification, biomarker development, tailored healthcare interventions and personalised treatment strategies to help bridge the gap between genomic information and clinical practice. pMedGR is particularly significant for the Greek population and neighbouring countries, which represent a genetic pool that differs from Central and Northern European populations, thus offering unique potential for the development of targeted therapies and diagnostic modalities specifically for this population.

The objectives of the new pMedGR infrastructure are to: (a) support research aiming at the transition from traditional symptom-based healthcare models to omics-based approaches for health and disease; (b) allow the in-depth description of individual phenotypes at a systems level by providing access to cutting-edge technological platforms, clinical data and biological specimens; (c) generate technological ICT solutions that facilitate the processing, integration and modeling of the output of several technological platforms; (d) train the next generation of physicians and bioscientists that will develop and implement personalised medicine; and (e) lead industrial innovation towards novel diagnostic and therapeutic modalities and advanced knowledge for personalised healthcare.

pMedGR will have close contacts with BBMRI-GR, the biobanking RI, in order to align activities in providing access to biological specimens and data. The ICT modules of pMedGR, which will be responsible for data analysis, integration and model building, will cooperate closely with ELIXIR-GR, the data storage infrastructure. Key interactions will be pursued also with INFRAFRONTIER-GR/Phenotypos, the mouse archiving and phenotyping infrastructure, which is expected to provide preclinical platforms and proof-of-principle projects for further clinical development. Lastly, pMedGR will cooperate with BioImaging-GR for the development of advanced imaging platforms with clinical applications for personalised medicine. pMedGR will also liaise with the European counterparts of these and other ESFRIs in order to establish an international network of partners that can provide relevant know-how and expertise.

pMedGR will provide a hub for the implementation, coordination and integration of personalised medicine approaches in the region and as part of a pan-European and global network, thus offering centralised information on patient stratification efforts, susceptibility factors and response to treatments for the regional population. This hub will effectively serve as a single entry point for researchers and industry interested in this area. Furthermore, Greece's strategic geopolitical position together with the region's genetic characteristics render pMedGR an ideal paradigm for personalised approaches that target an extended regional area, including southern Italy, the Balkans & Turkey. Through pMedGR, Greece has the potential to become a South East European Node for Personalised Medicine, linking Europe to emerging markets such as Asia, Africa and the Middle East.

National Biomedical Research road map
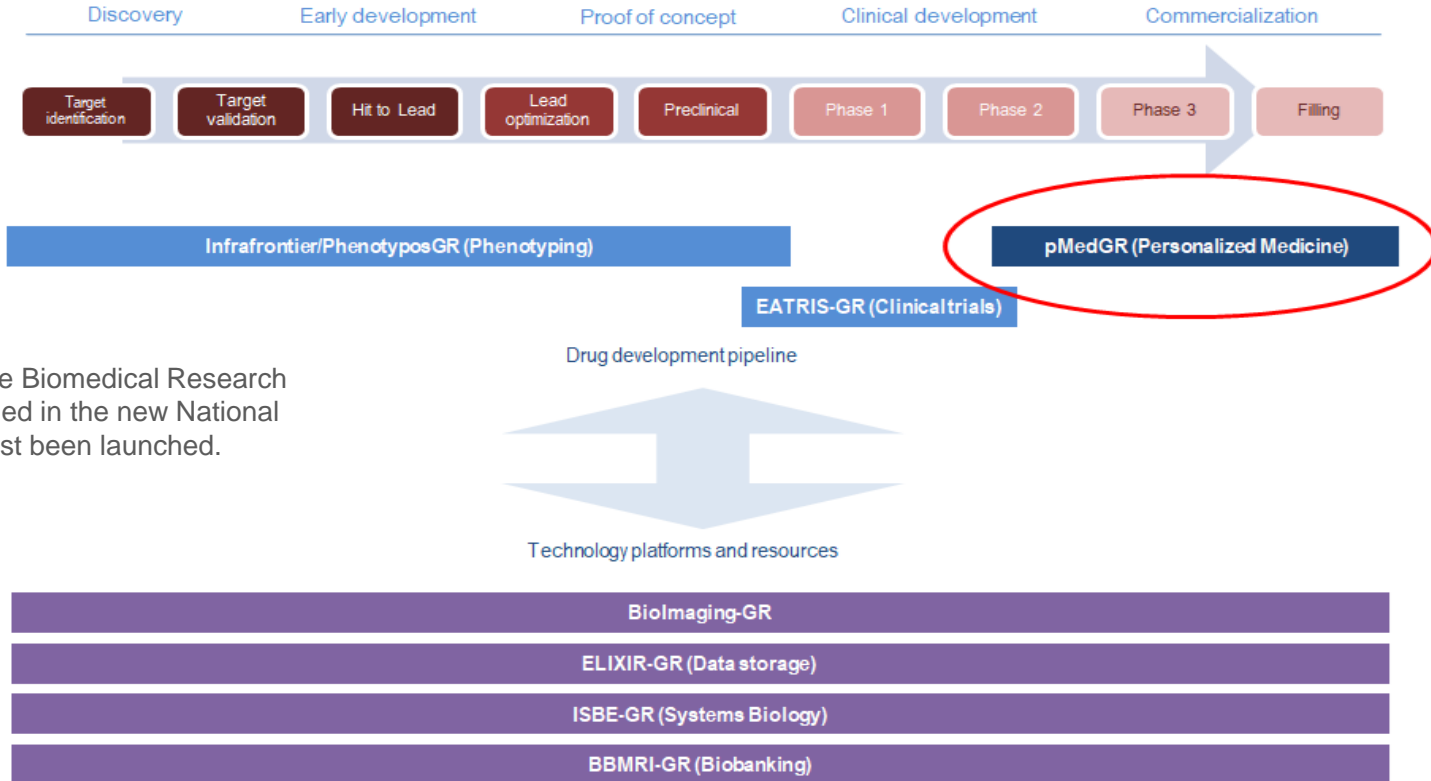
pMedGR is one of the Biomedical Research Infrastructures included in the new National Roadmap that has just been launched.

pMedGR - Aims

- Strengthen basic research and public health

- Join the "Big-Data" communities

- Revise clinical trial designs

- Move regulatory science forward

- Educate new generation of Doctors and PhDs

- Become a bridge between industry and academia

- Ensure development of precision medicine in a safe technical and ethical framework

Coordinators

Prof. Petros Sfikakis

- President and Professor of Internal Medicine & Rheumatology at the Medical School of National and Kapodistrian University of Athens

Prof. George Kollias

- Professor of Experimental Physiology at the National and Kapodistrian Medical School of the University of Athens
- President and Scientific Director at the Biomedical Sciences Research Center BSRC "Alexander Fleming"
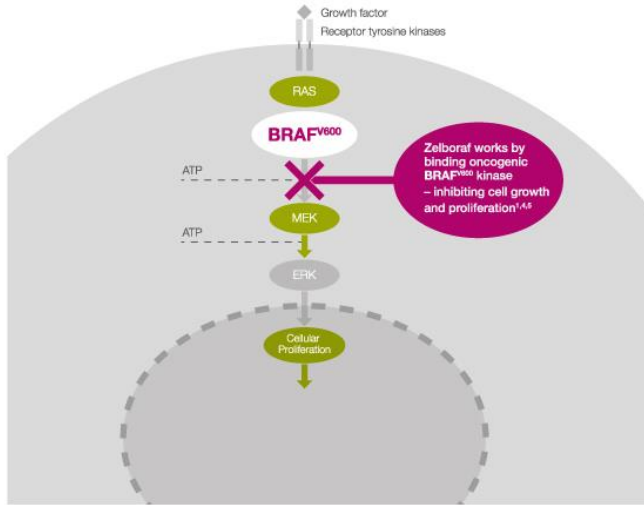
About pMedGR

4.000.000 Euros

4 years (started 19/12/2017)

Independent 400m^2 space at the Medical School

Zelboraf MOA

Zelboraf™
vemurafenib
The power of personalization

Identification of $BRAF^{V600}$ mutations is key to optimizing treatment in metastatic melanoma, as only patients with the $BRAF^{V600}$ mutations may benefit from Zelboraf therapy.
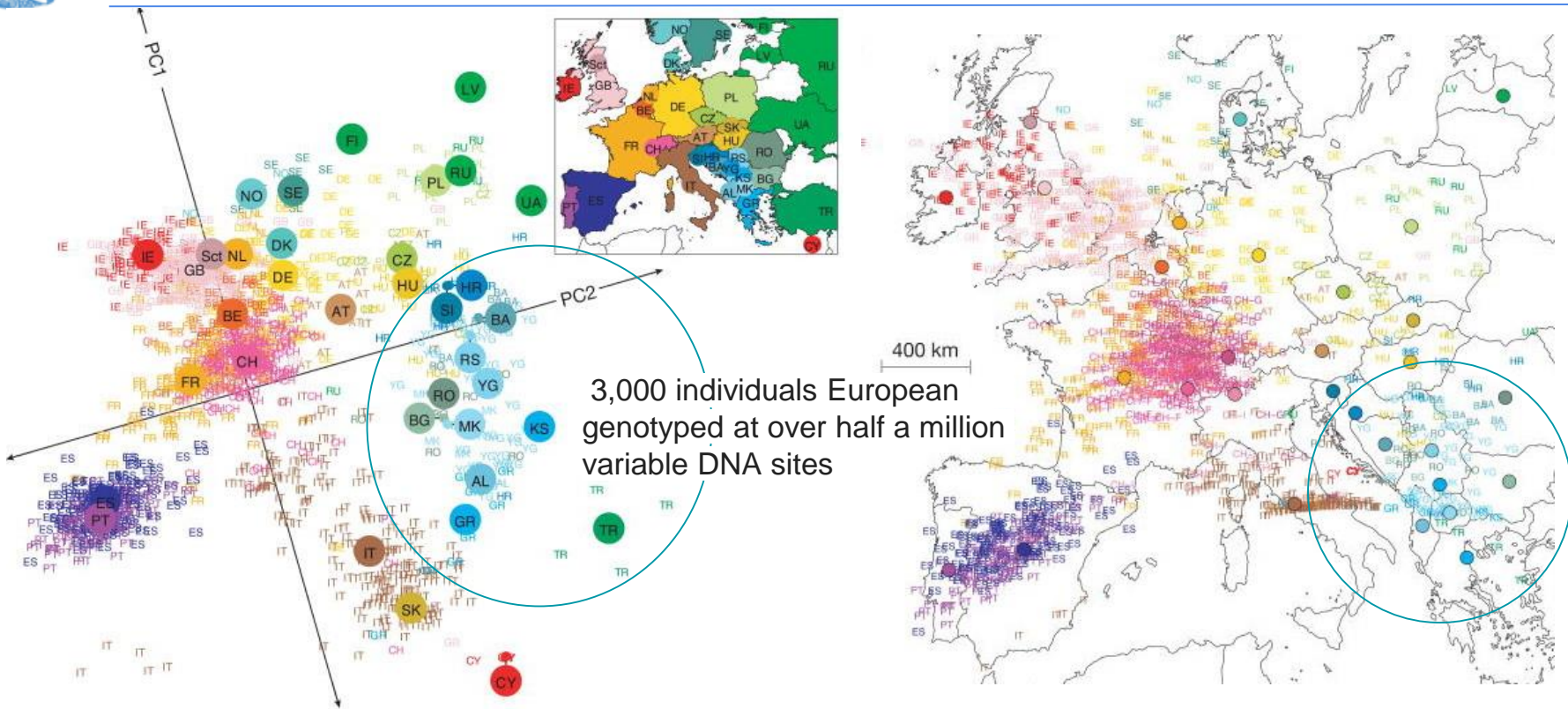
Approximately 50% of metastatic melanoma patients are positive for $BRAF^{V600}$ mutations.

To be eligible for Zelboraf, patients must have their $BRAF^{V600}$ mutation-positive tumor status confirmed by a clinically validated test.

81% of metastatic melanoma patients with a B-RAF^{V600} activating mutation responded to treatment with Zelboraf.
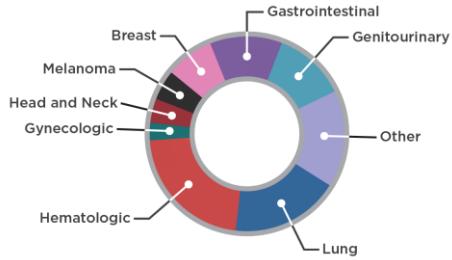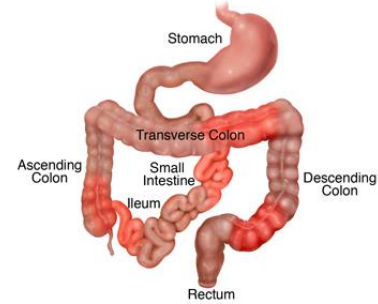
Genetic diversity even within Europe

3,000 individuals European genotyped at over half a million variable DNA sites

Genes mirror geography within Europe, NATURE, Vol 456|6 November 2008

CANCER

CHRONIC INFLAMMATORY DISEASES

NEURODEGENERATIVE

pMedGR and Bioinformatics data production

Illumina NGS 550

10X Genomics single cell transcriptomics and
CyTOF Helios – single cell Mass Cytometer

Thermo Scientific Q Exactive HF-X Hybrid Quadrupole-Orbitrap
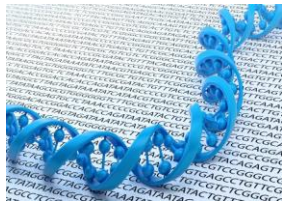Mass Spectrometry System

Servers

pMedGR - Structure

Clinical Tissue Sampling Facility

The Unit will determine strategies and implement best practices for collecting, cataloguing, and storing samples and specimens (fresh, frozen or FFPE samples) for use.

Personalised Genomics Facility

The Unit will provide services and support in high-throughput, genome wide research, including genomic applications (whole genome sequencing, exome sequencing, whole genome mapping, genotyping etc), transcriptomic (RNA-Seq, smallRNA-Seq), epigenomic (MeDIP-Seq, ChIP-Seq, bisulfide sequencing etc), metagenomic and genotyping services.

Proteomics and Metabolomics

The Unit will provide the following services:
- improved sample separation and sensitivity
- accurate quantization in parallel with identification
- high-throughput analysis of proteins and metabolites
- metabolic profiling and fingerprinting
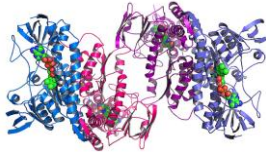
Data Analysis, Integration and Modeling Unit

The Unit will provide bioinformatic and data analysis resources for individual medical genomic applications through the following pipelines:
Analysis of genetic variability
- Transcriptome profiling
- Pharmacogenomic analyses
- Individual epigenetic profiling
- Modeling
- Efficient reference genome indexing
- ExomeSeq data analysis
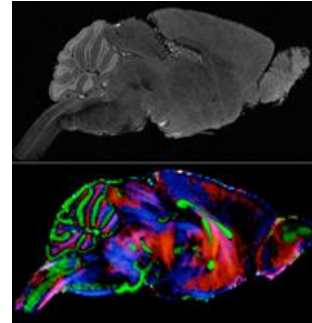
Advanced Imaging Facility

The Unit will employ new approaches for the discovery and validation of novel biomarkers. These include:
- light sheet and multi-photon microscopy system
- echographic apparatus for assessing novel treatment strategies for heart and vascular diseases
- probe-based in vivo imaging for assessing novel biomarkers for disease progression

pMedGR and elixir

- Services and pipelines

- Data production

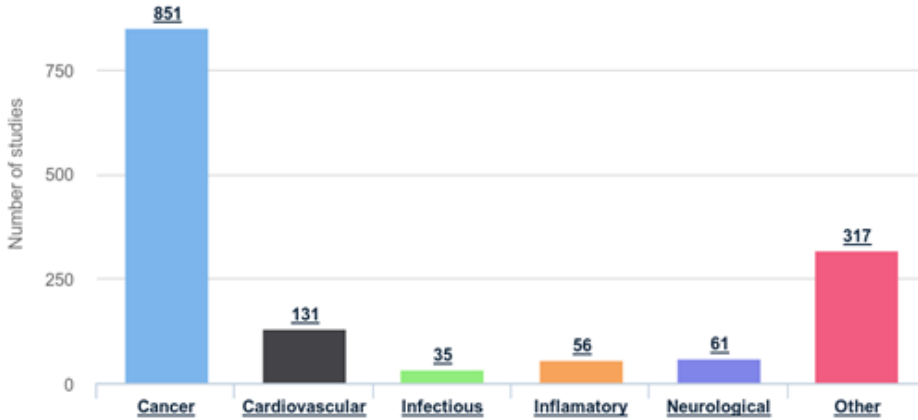- Data storage

- Data analysis

- Data sharing
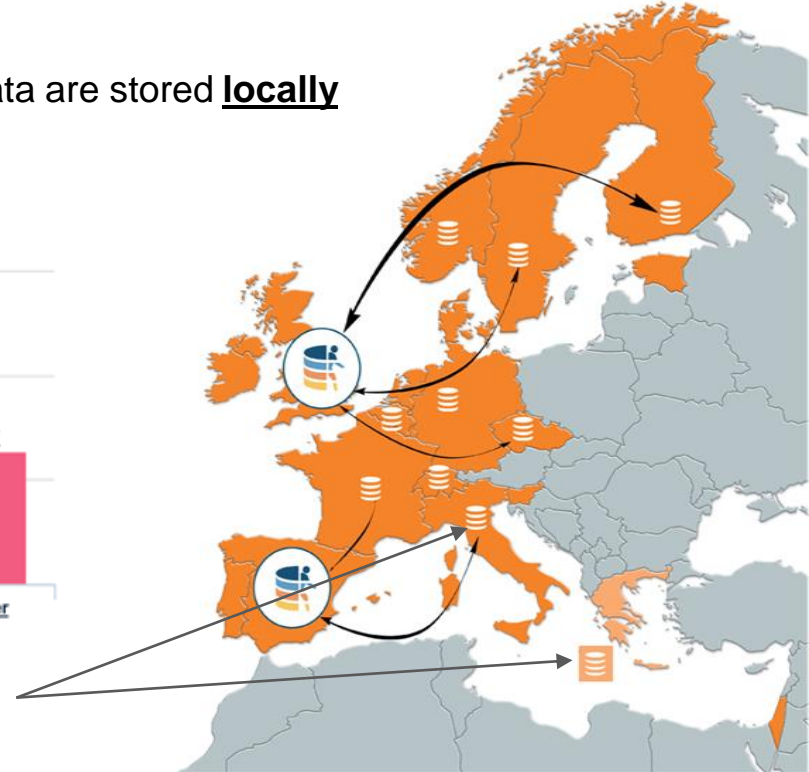
pMedGR and elixir

Sensitive controlled-access data are stored **locally**

Local EGA

pMedGR and elixir
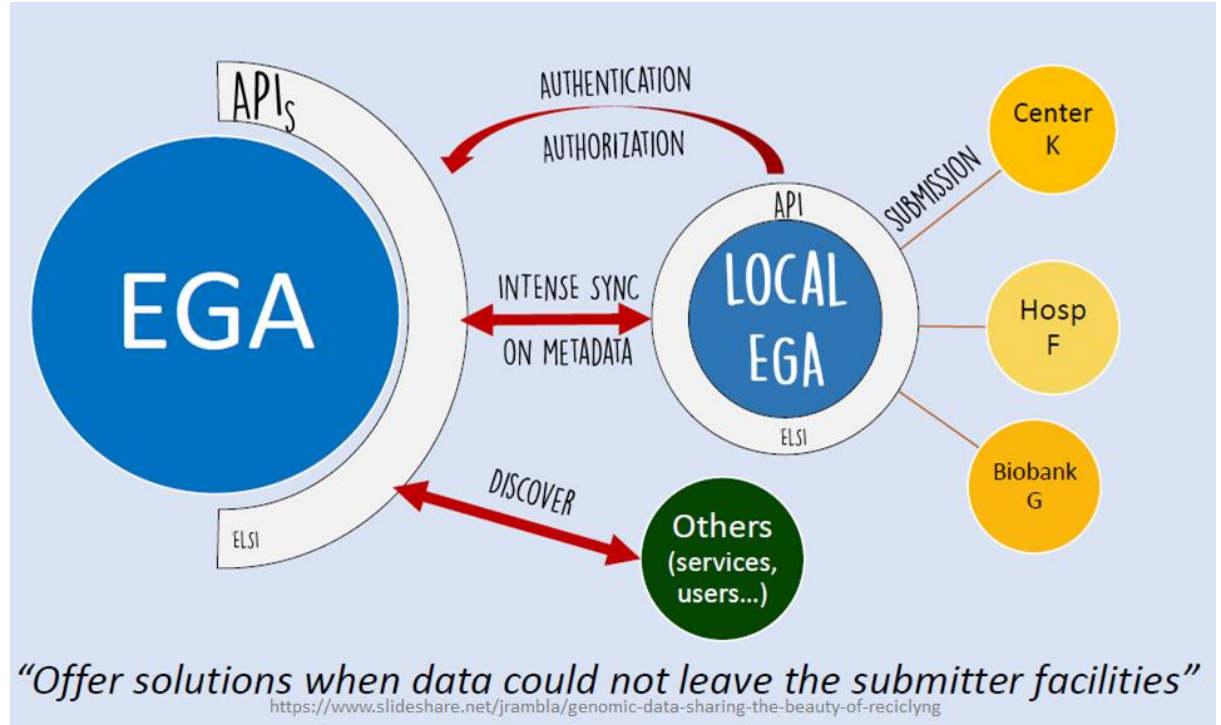
- Data are stored locally

- Metadata are shared

"Offer solutions when data could not leave the submitter facilities"
https://www.slideshare.net/jrambla/genomic-data-sharing-the-beauty-of-reciclyng

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Thank you

PARTNERS

University of Athens   BSRC Fleming   BRFAA