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IMGGE biobank of thrombosis from the past to the future

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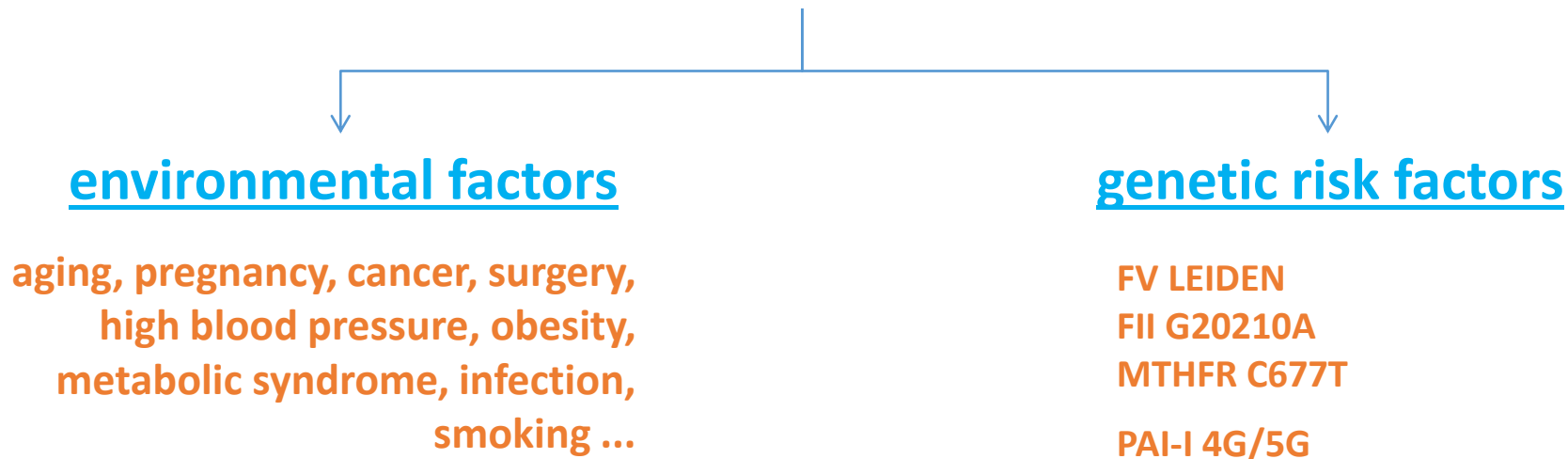
Outline

- ❑ Thrombosis and Biobanking, our research
- ❑ The Structure and the Interface of the Database
- ❑ The Future and Applications
- ❑ Challenges

Thrombosis

THROMBOPHILIA is an increased tendency to form abnormal blood clots in blood vessels.,, A service of the U.S. National Library of Medicine, 2015

multifactorial disease



Clinical manifestation:

stroke, myocardial infarction, deep vein thrombosis, pulmonary embolism, rare cases of thrombotic events... and fertility and pregnancy complications



- ❑ 20 years research on the genetic basis of thrombophilia
- ❑ First routine genetic analysis in Serbia 2002.
- ❑ Collaboration with medical institutions
- ❑ Thrombophilic patients database and DNA repository of 6,000 patients with different thrombotic events, fertility and pregnancy complications, as well as healthy subjects (control groups)
- ❑ Better understanding genetic basis of thrombophilia

<http://www.imgge.bg.ac.rs>

Biobanks

- ❑ Collecting and storing of biological material with appropriate clinical and epidemiological data
- ❑ The data and the samples are collected continuously
- ❑ Used for current and future research projects

Human genetic research databases (HGRDs)

Population genetic databases

Repository

Tissue bank



<http://www.ctmm-trait.nl/work-packages/work-package-3-biobanking>

The data in database

- ❑ general (gender, BMI, age...)
- ❑ contact (personal, clinical...)
- ❑ specimen (type, date, concentration...)
- ❑ clinical data (type, location and date of thrombotic event...)
- ❑ biochemical data (hemostasis, biochemical tests...)
- ❑ epidemiological data (lifestyle data, risk factors, family history...)
- ❑ genetics (genes, gene variants...)
- ❑ ID numbers (patients, specimens...)

Timeframe of database building



2000

2010

2017

Brief overview of the REDCap

- ❑ REDCap is a tool for building online surveys and databases
- ❑ Projects are webpages (data collection instruments), used for data entry
- ❑ Designed to be neutral, can capture almost any type of data, for any purpose
- ❑ Entering data while logged, using REDCap as a database
- ❑ Entering data without logging in, using REDCap as a survey
- ❑ Data entry process is intuitive, instructional text and prompts provide guidance at every step
- ❑ Most frequently used for clinical and research purposes, but it is designed to support any type of work
- ❑ REDCap mobile phone version

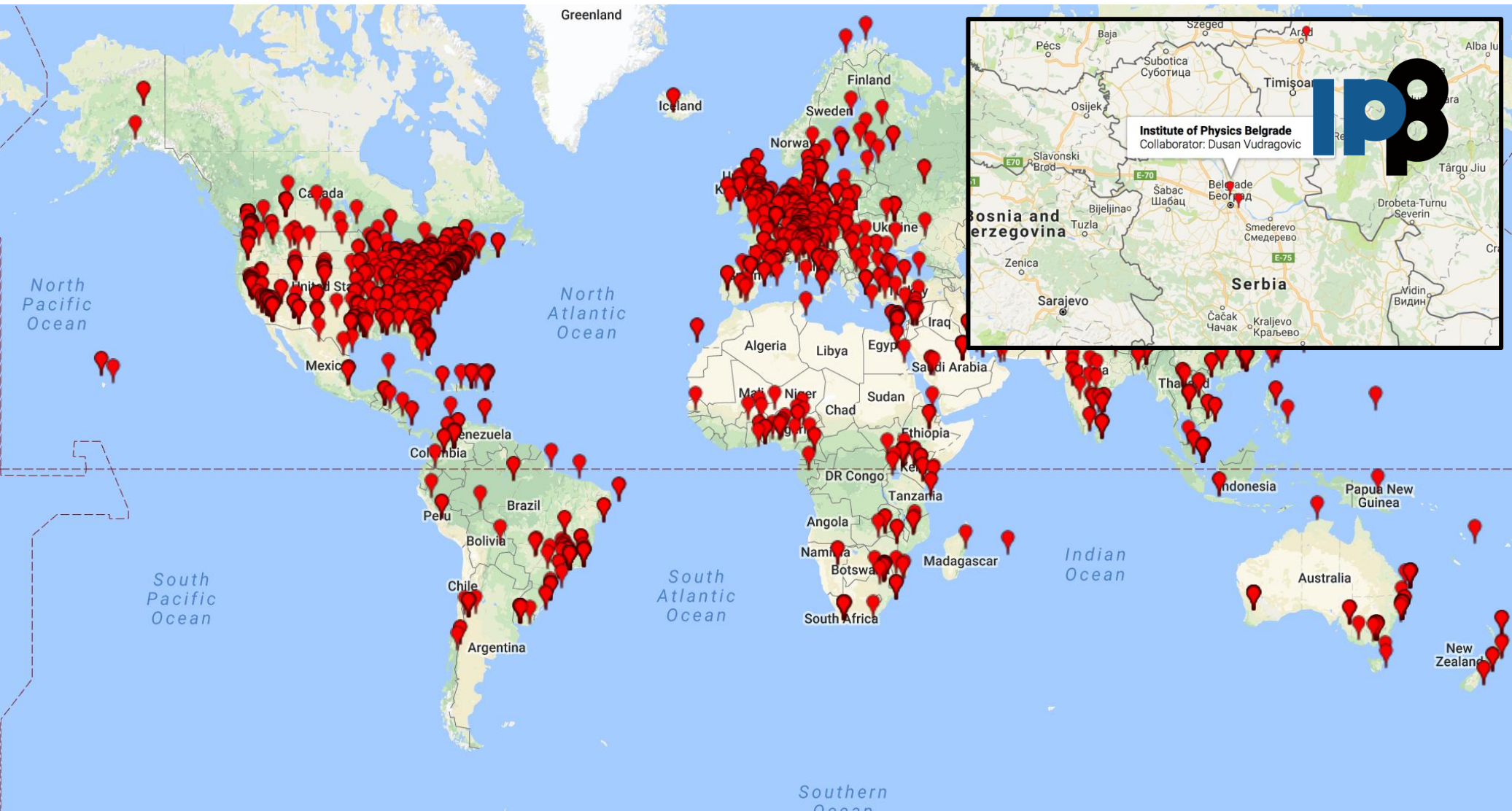
REDCap project development

- ❑ Customizing the questions is part of project development
- ❑ Development process can involve advanced features to improve your data collection strategy: data validation, branching logic, calculated fields, matrix ranking, slider scales
- ❑ Instruments can be built online in real-time through the Online Designer or built offline through a spreadsheet
- ❑ Project can use only one data collection instrument or multiple instruments (control access)
- ❑ These instruments are where project data can be added, modified or deleted

REDCap built-in tools

- ❑ Many data management tools, here few the most important
- ❑ User rights – users of the project
 - ❑ Access level for data collection instruments and applications can be set
- ❑ Report builder – the search engine of a REDCap project
 - ❑ View multiple records worth of data without exporting it
- ❑ Data export tool – snapshot of current dataset
 - ❑ Saved as a file that can be download and used externally
 - ❑ Several common statistical packages formats are supported
- ❑ Data import tool – import data for multiple fields over multiple records
 - ❑ Existing data in a spreadsheet or other database can be transferred into REDCap
 - ❑ Can be used to modify existing data or add entirely new data

REDCap instances



Biobank structure and data

- ❑ Biobank
<https://redcap.ipb.ac.rs/>
- ❑ The following instruments are created
 - ❑ Contact information
 - ❑ General information
 - ❑ Biochemistry
 - ❑ Epidemiology
 - ❑ Clinical information
- ❑ Imported records from 5,053 patients
 - ❑ Gender
 - ❑ Date of birth
 - ❑ FV Leiden
 - ❑ FII G20210A
 - ❑ MTHFR C677T
 - ❑ PAI I 4G/5G
 - ❑ Type of sample

Output papers

More than **60 papers**:

- ❑ frequencies of mutations in Serbian population
- ❑ correlation with clinical manifestations
- ❑ correlations with age of the first thrombotic event
- ❑ ...

New thrombotic biomarkers

Prothrombin (FII) gene variants- 7 new detected *prothrombin Belgrade* Antithrombin resistance

NCBI Resources How To

PubMed.gov
US National Library of Medicine
National Institutes of Health

PubMed Advanced

Abstract Send to:

J Thromb Haemost. 2013 Oct;11(10):1936-9. doi: 10.1111/jth.12367.

A novel prothrombin mutation in two families with prominent thrombophilia--the first cases of antithrombin resistance in a Caucasian population.

Diordjevic V¹, Kovac M, Milic P, Murata M, Takaqi A, Pruner I, Francuski D, Kojima T, Radojkovic D.

Author information

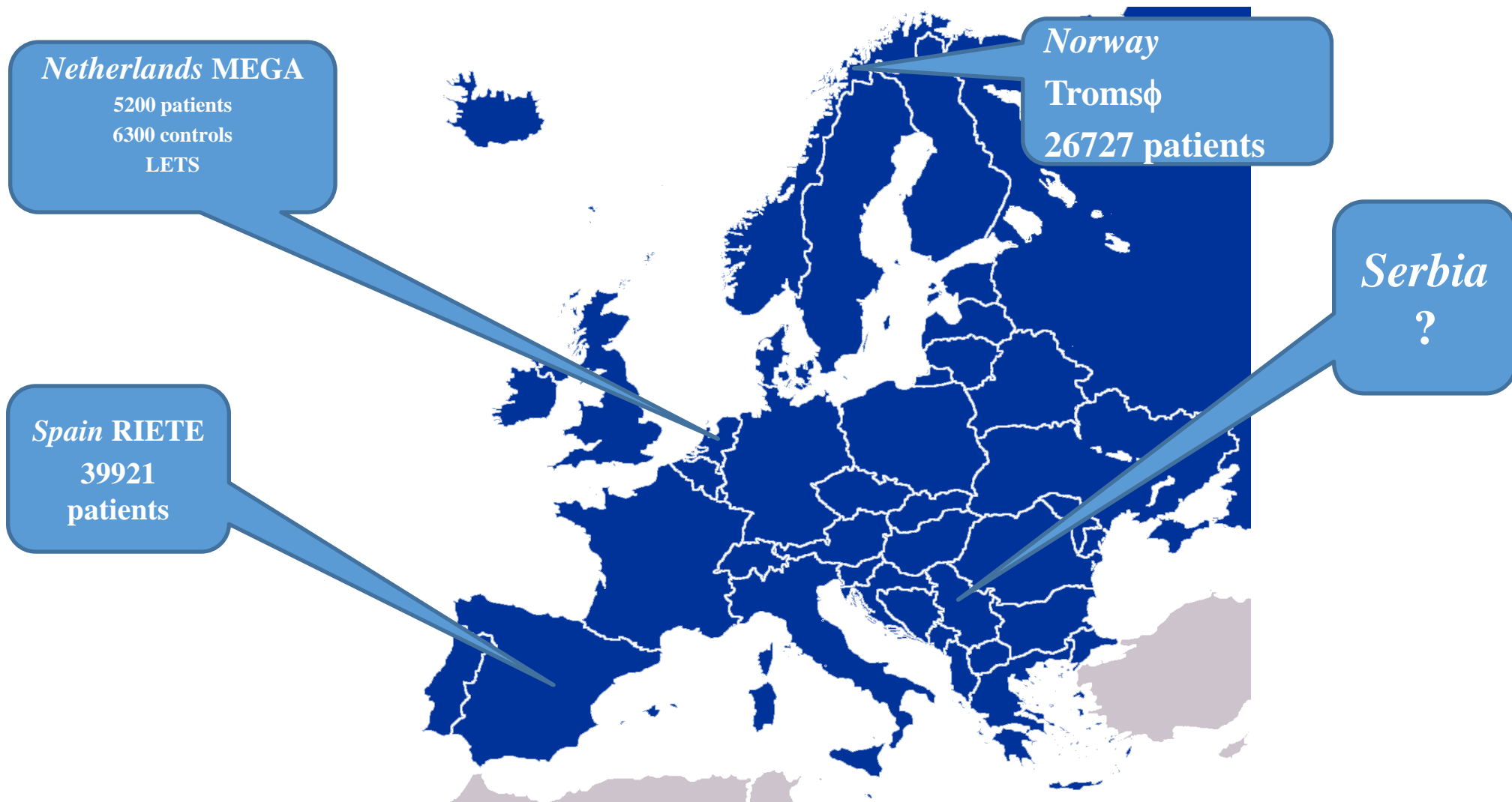
¹Institute of Molecular Genetics and Genetic Engineering, University of Belgrade, Belgrade, Serbia.

Ann Hematol. 2015 Oct 19. [Epub ahead of print]

The first case of antithrombin-resistant prothrombin Belgrade mutation in Japanese.

Kishimoto M¹, Suzuki N², Murata M³, Ogawa M⁴, Kanematsu T⁴, Takaqi A³, Kiyoi H⁴, Kojima T³, Matsushita T².

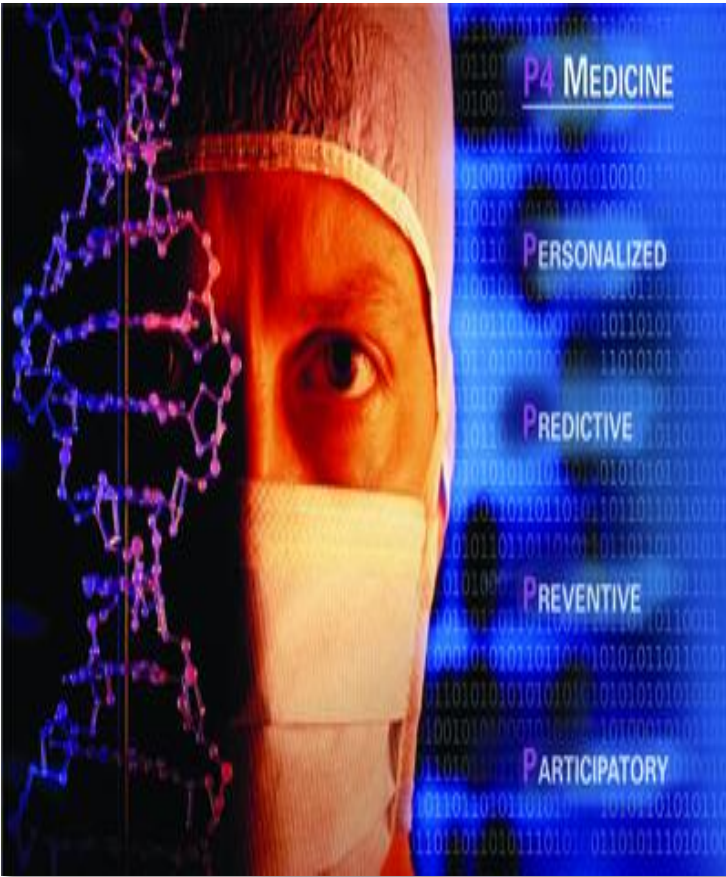
The thrombo biobanks/databases in Europe



The FUTURE and APPLICATIONS

- ❑ Build algorithm for different types of multifactorial diseases, with unknown cause, ethiology and progression
- ❑ Facilitated diagnosis
- ❑ Improving prevention and the ability to track patient
- ❑ Quick and cheaper diagnosis and more detailed analysis
- ❑ Higher quality of treatment and prevention
- ❑ Personalized approaches
- ❑ Application in diagnostics, prevention and treatment

P4 Medicine



1. ***“personalized”** which reflects the individual “personal digital genome”;*
2. ***“predictive”** which is due to the ability to predict the risk of certain diseases based on “personal digital genome” information in combination with lifestyle data, age, sex, occupation etc.;*
3. ***“preventive”** that is based on individualized risk prediction,*
4. ***“participatory”** of the individual concerned in proactively maintaining their health.*

<http://genomicsforeveryone.org/genomics-and-p4-medicine-ethics-and-policy/>

Challenges of the biobanks



- ❑ Conceptual and technical organisation
- ❑ Fundings
- ❑ Legislative regarding organisation and ethical issues
- ❑ Involves the formation of administrative bodies (eg. ethics committee) and procedures that protect the rights and interests of the sample donors (GDPR)



Image Credit: Pe3k/Shutterstock

Welcome to Belgrade

Belgrade Bioinformatics Conference *BELBI2018*

Belgrade, Serbia
June, 18-22, 2018



Topics:

- ☐ *Theoretical Approaches to BioInformation Systems*
- ☐ *Bioinformatics and Data Mining of Biological Data*
- ☐ *Biomedical Informatics*

<http://belbi.bg.ac.rs/>