Personalised medicine

European Commission's policy development in the field of personalised medicine

Workshop on personalised medicines: role of patients, consumers and healthcare professionals

EMA, London 14 March 2017

Irene Norstedt, Head of Unit Innovative and Personalised Medicine, DG Research and Innovation, European Commission

Research and Innovation

Personalised medicine at activities at EU level

2010: **Preparatory workshops**

2011: **European Perspectives conference**

2013: **Commission Staff Working Document on "use of '-omics' technologies in the development of personalised medicine"**

2015: **Council conclusions on Personalised Medicine**

2015: **Strategic Research and Innovation Agenda of PerMed**

2016: **Personalised Medicine Conference**

2016: **Launch of International Consortium of Personalised Medicine**

Research areas

- Large scale data gathering and "-on
- Technology development
- Statistics
- Diagnostics
- Biomarkers
- Clinical trial methodologies
- Pre-clinical and clinical research
- Rare diseases: small patient populations
- Omics for health promotion and disease prevention
- Piloting personalised medicine in healthcare

EU funding - over 2 billion EUR to top research

PerMed

Shaping Europe's Vision for Personalised Medicine
Strategic Research and Innovation Agenda (SRIA)

PERSONALISED MEDICINE CONFERENCE 2016
1-2 June, Brussels

Definition of personalised medicine
Council Conclusions on personalised medicine for patients (2015/C 421/03)

"Personalised medicine refers to a medical model using characterisation of individuals' phenotypes and genotypes (e.g. molecular profiling, medical imaging, lifestyle data) for tailoring the right therapeutic strategy for the right person at the right time, and/or to determine the predisposition to disease and/or to deliver timely and targeted prevention"

Definition developed by the Advisory group for the H2020 Health, demographic change and well-being challenge

International Consortium for Personalised Medicine (IC PerMed)

WHAT

Collaboration of research funders and policy makers from EU Member States and beyond

WHY

- Establish Europe as a global leader in PM research
- Support the PM science base through a coordinated approach to research
- Provide evidence to demonstrate the benefit of PM to citizens and healthcare systems
- Pave the way for PM approaches for citizens

HOW

Implementation of a joint action plan based on PerMed Strategic Research Agenda (SRIA)

IC PerMed Challenges and facilitators

Wolfgang Ballensiefen, Ministry of Research (DLR-PT), Germany

Maria Judith Molnar
Health Ministry, Hungary

Policy, Funders, Researchers, Industry, Ethics/Data committees, Patients/Citizens

Policy, Patients/Citizens, Industry, Funders, Researchers

Industry, Policy, Regulators, Funders, Researchers, Patients/Citizens

Daria Julkowska, National Research Agency, France
Hemma Bauer, Ministry of Science, Research and Economy, Austria

Gaetano Guglielmi
Ministry of Health, Italy

Policy, Providers, Health Technology Assessment, Insurances, Patients/Citizens

2 Data and ICT

1 Citizens and Patients

Innovations in Diagnosis, Therapies, Prevention & ICT with **economic value** and **fair access**.

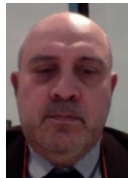**3 Research efforts**

5 Health Systems

4 Market Access

Policy, Industry, Health Technology Assessment, Providers, Researchers, Patients/Citizens

Peter Hongaard Andersen
Innovation Fund, Denmark

Image source © Wolfgang Ballensiefen / PerMed project 2016

Elected IC PerMed Chair and Vice-Chairs

Chair

Vice-Chair

Mairead O'Driscoll
The Health Research
Board - Ireland

Vice-Chair

Wolfgang Ballensiefen
DLR Project Management
Agency (DLR PT) -
Germany

Ain Aaviksoo
Ministry of Social
Affairs, Estonia

Personalised medicine conference

Personalised Medicine Conference 2016

1-2 June, Brussels

The Personalised Medicine Conference 2016 is now over. The organising team thanks all speakers and participants for two interesting and thought-provoking days.

Conference material

You can still browse the **programme** as well as the lists of **speakers** and **participants**.

You can view the videos of the Conference: **1st day** | **2nd day**

Conference presentations

Click on a session title to see presentations

Keynote session

Session1: Developing Awareness and Empowerment

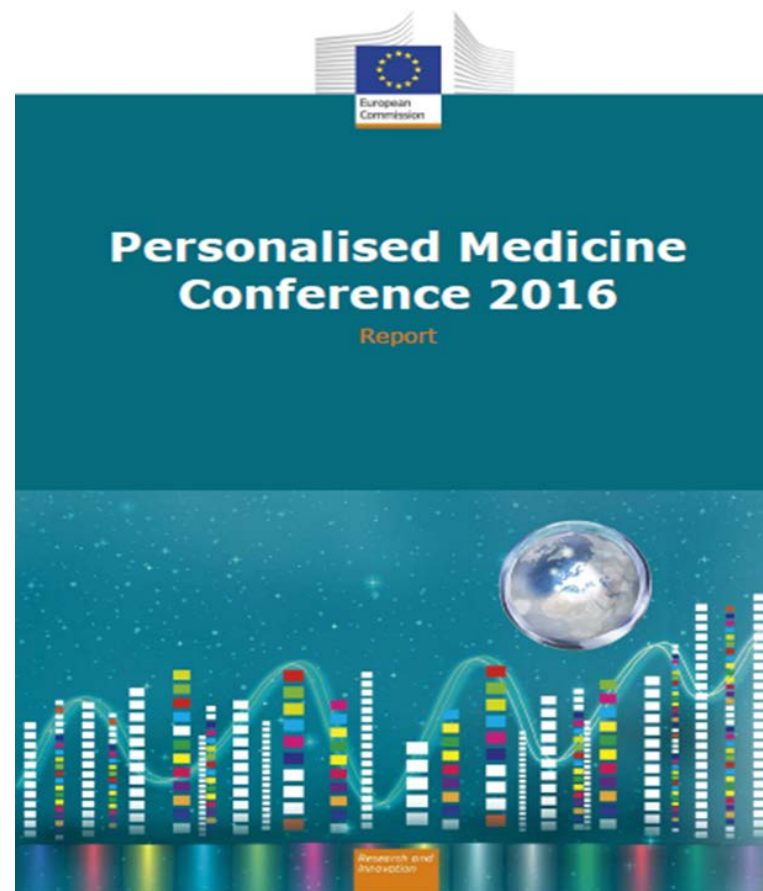**Session 2: Integrating Big Data and ICT Solutions**

Relevant Documents

- Conference Programme 495 KB
- Towards IC PerMed 140 KB
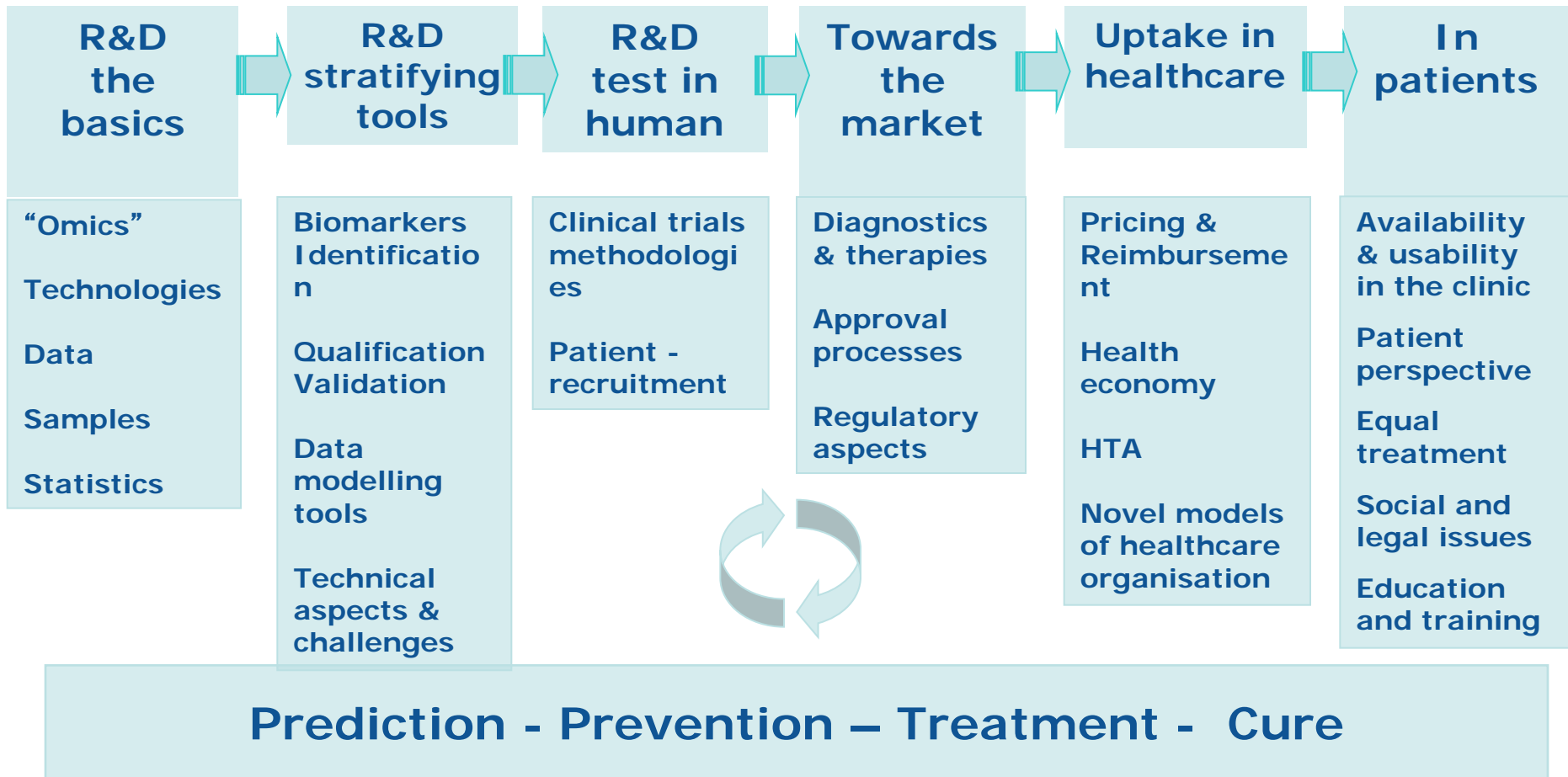- Council conclusions on personalised medicine for patients
- Area map 672 KB

Video

Watch the Conference as it unfolded

- 1st day, 1 June
- 2nd day, 2 June

Personalised Medicine Conference 2016

Report

http://ec.europa.eu/permed2016

Framework for Personalised Medicine

R&D the basics	R&D stratifying tools	R&D test in human	Towards the market	Uptake in healthcare	In patients
"Omics" Technologies Data Samples Statistics	Biomarkers Identification Qualification Validation Data modelling tools Technical aspects & challenges	Clinical trials methodologies Patient - recruitment	Diagnostics & therapies Approval processes Regulatory aspects	Pricing & Reimbursement Health economy HTA Novel models of healthcare organisation	Availability & usability in the clinic Patient perspective Equal treatment Social and legal issues Education and training

Prediction - Prevention – Treatment - Cure

- **Focus on blood epigenomics**
- **Generation of >110 human epigenome maps of blood cells**
- **Almost 300 publications, patents…**
- **All data available freely via sinle-entry data portal**
- **Innovative technologies developed, single cell analysis of DNA methylation**
- **New computational tools for data analysis and sharing, new international standards set**
- **SME Cambridge Epigenetix already commercialises the new technologies**
- **The cornerstone of International Human Epigenome Consortium**

Findings with clinical implications:

- Sepsis as autoimmune disease: epigenetic basis of innate immune memory
- Alzheimer 's disease: implications of novel checkpoint drugs **for microglia development**
- Leukemia: discovery of 3 new subtypes, novel biomarker under development

EuroGentest

- Harmonisation, validation and standardisation in genetic testing
- Support professionals in achieving high quality in all aspects of genetic testing services
- Provide information on genetic testing to professionals and to the public
- Promote the implementation of novel technologies into current practice

www.eurogentest.org
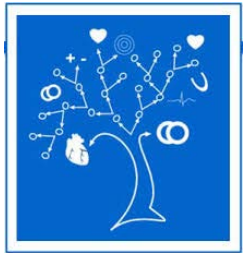
EVINCI STUDY

Evaluation of integrated cardiac imaging for the detection and characterization of ischemic heart disease

- ✓ A pan-European multicentre trial performed on a cohort of 697 patients with suspected ischaemic heart disease (IHD).

- ✓ Creation of the European digital and biological banks for multimodal cardiovascular imaging and blood samples.

- ✓ Definition of biomarkers for the screening of patients with suspected coronary artery disease prior to or together with cardiovascular imaging assessment.

- ✓ Determination of the most accurate non-invasive "anatomo-functional" cardiac imaging strategy for the detection and management of IHD.

- ✓ Development of an advanced clinical and imaging reporting as well as an integrated decision making tool in cardiology.

- ✓ EduCAD - a new web based tool for training young cardiologists in the appropriate and more effective use of imaging tests for diagnosing IHD.
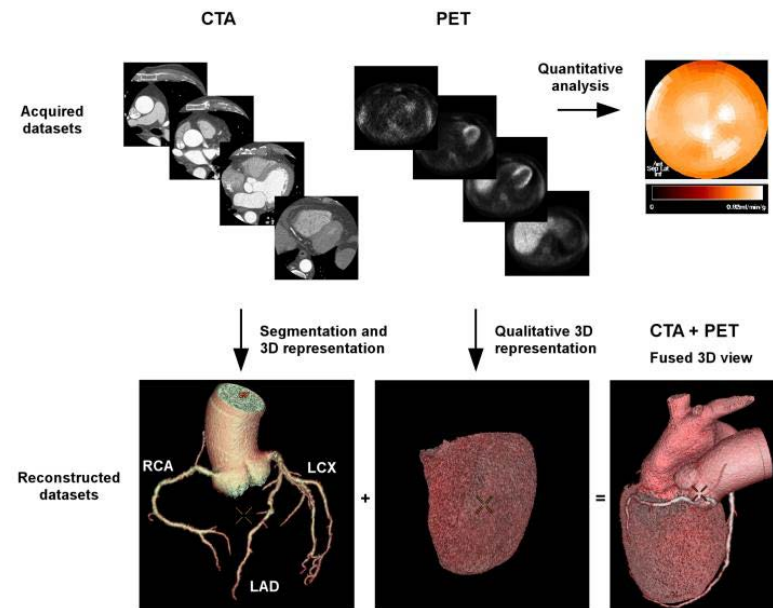
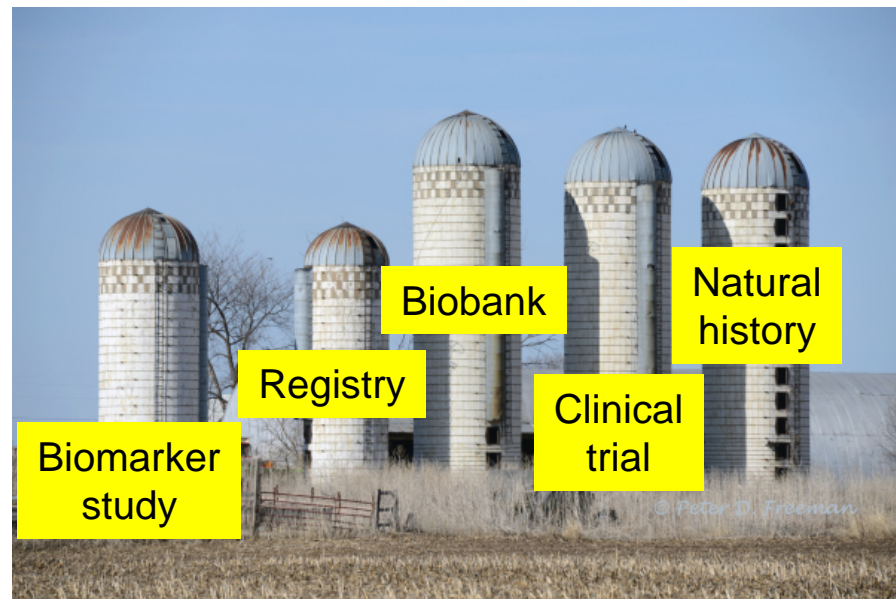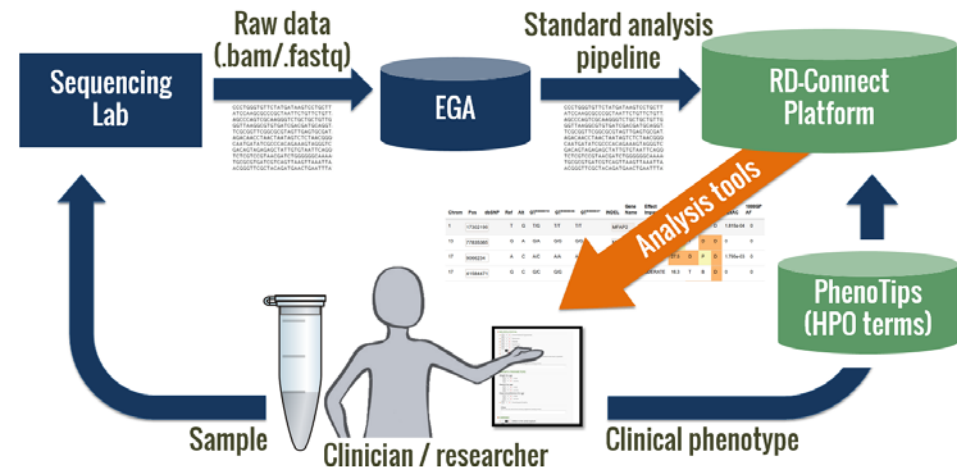| **RD Connect**	Infrastructure for data sharing in rare disease research	Flagship IRDiRC project implementing IRDiRC policies and guidelines on data sharing	EU 7th Framework Programme, 12M EUR, 6 years
Genomic analysis and gene discovery	Standardized phenotypic data collection	Searchable catalogue of biosamples	Data linkage across resources

Overcoming Silos
Data sharing for research and better data analysis

Omics data, clinical data and biosamples from individual with RD

Biomarker study · Registry · Biobank · Clinical trial · Natural history

Raw data (.bam/.fastq) · Standard analysis pipeline · Sequencing Lab · EGA · RD-Connect Platform · Analysis tools · PhenoTips (HPO terms) · Sample · Clinician / researcher · Clinical phenotype
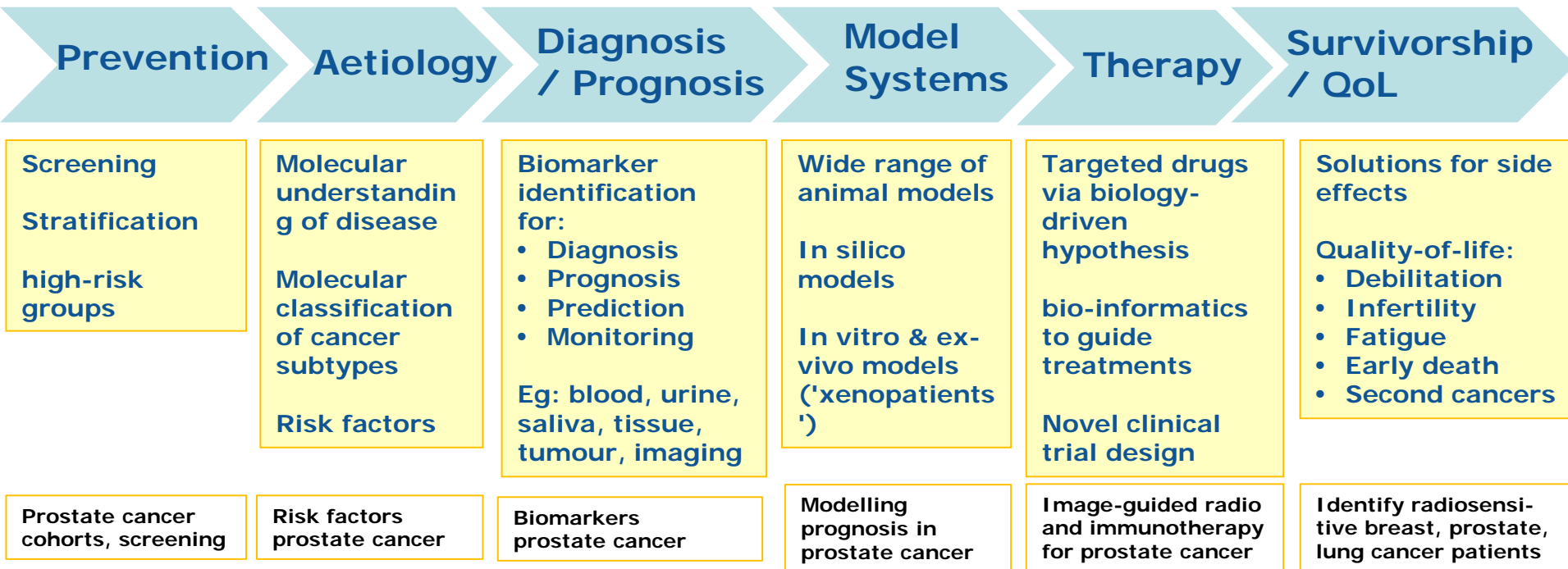
Disease-causing variant can be identified using the **genomics analysis platform**

Sample is findable in the **Sample Catalogue**

Registry data in the **ID-Cards directory** of registries and biobanks

Towards personalised medicine: prostate cancer

Prevention	Aetiology	Diagnosis / Prognosis	Model Systems	Therapy	Survivorship / QoL
Screening Stratification high-risk groups	Molecular understanding of disease Molecular classification of cancer subtypes Risk factors	Biomarker identification for: • Diagnosis • Prognosis • Prediction • Monitoring Eg: blood, urine, saliva, tissue, tumour, imaging	Wide range of animal models In silico models In vitro & ex-vivo models ('xenopatients')	Targeted drugs via biology-driven hypothesis bio-informatics to guide treatments Novel clinical trial design	Solutions for side effects Quality-of-life: • Debilitation • Infertility • Fatigue • Early death • Second cancers
Prostate cancer cohorts, screening	Risk factors prostate cancer	Biomarkers prostate cancer	Modelling prognosis in prostate cancer	Image-guided radio and immunotherapy for prostate cancer	Identify radiosensi-tive breast, prostate, lung cancer patients

Clinical trial methodologies for small populations

March 3, 2016 – Small Population Clinical Trials Task Force Workshop – EMA, London

June 2016 publication of the Small Population Clinical Trials Task Force Workshop Report and Recommendations

April 2017 a meeting is planned at the EMA to discuss project findings with stakeholders for a final joint position statement

Piloting personalised medicine

U-PGx

Ubiquitous Pharmacogenomics: Making actionable pharmacogenomic data and effective treatment optimisation accessible to every European citizen

- Pre-emptive genotyping of multiple important pharmacogenes
- Data collected prospectively and embedded into the electronic records of patients in NL, ES, UK, IT, AT, GR and SL
- Prescribers and pharmacists alerted through electronic clinical decision support systems when a drug is ordered or dispensed for a patient with an at-risk genotype
- Analysis of cost-effectiveness and health outcomes

Innovative Medicines Initiative

> €5 bn

€2.5 bn   Partnership 2008 - 2024   €2.5 bn
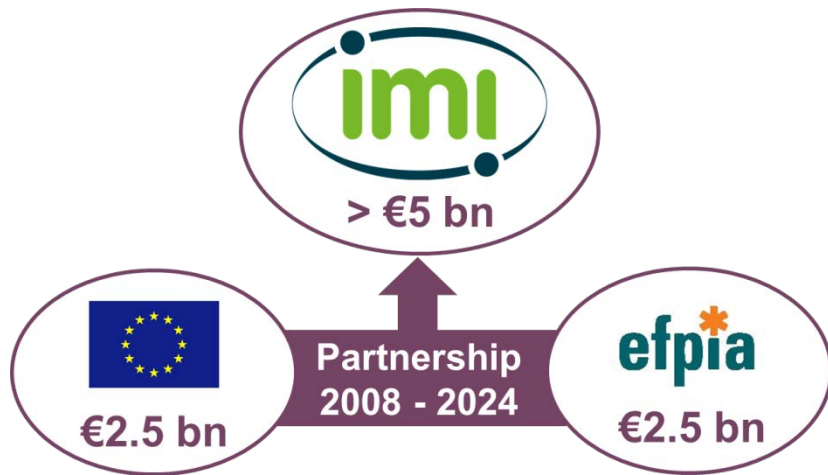
A partnership between EU Commission and the European Federation of Pharmaceutical Industries and Association (EFPIA)

Patient stratification - Efficacy and Safety

Adaptive pathways

Patient engagement/education

Big data for health outcomes

Patient reported outcomes

Etc.

GETREAL - incorporating real-life clinical data in drug development

- *We need data to assess relative effectiveness*
- *Project develops guidance on generating real world data during drug development*
- *Analysis of existing processes and methodologies*

- *Network of regulators, HTA, companies, academics, healthcare professionals, patients etc.*

www.imi-getreal.eu

ADAPT SMART – Coordinating work on MAPPs

- *Coordination & Support Action on Medicines Adaptive Pathways ot Patients (MAPPs)*

- *Project is building a platform with stakeholders to coordinate MAPPs activities in IMI*
  - Gap analysis – lessons learnt from existing IMI projects
  - Informing research activities – facilitate inclusion of tools / methodologies in IMI projects
  - Knowledge management – horizon-scanning of non-IMI activities

- *Recommendations will contribute to aligning understanding of impact of MAPPs vs current paradigm*
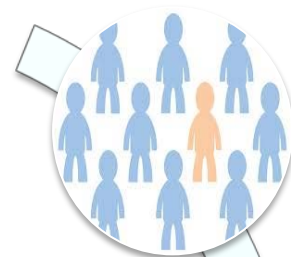
adaptsmart.eu

EU funded rare diseases research

Research priorities

Research Results

Rare diseases

R&I funding

Close to 900 million in more than 160 projects in FP7 and H2020 on: pathophysiology, natural history, delivered new diagnostics and therapies

E-Rare — Linking major EU and national initiatives

E-RARE: research funders collaboration: 25 partners from 17 countries

IRDiRC — International coordination: IRDIRC

IRDIRC: 200 new therapies and means to diagnose most rare diseases achieved in 2017: >40 international partners, policies and guidelines to implement goals

- European reference networks (ERNs) for rare diseases should serve as research and knowledge centres, updating and contributing to the latest scientific findings, treating patients from other Member States and ensuring the availability of subsequent treatment facilities where necessary.

- The definition of ERN should also reflect the need for services and expertise to be distributed across the EU.

  http://ec.europa.eu/health/ern/policy/index_en.htm

Need for a coherent strategy – from bench to bedside

- More efficiently bring the results of research and innovation to the patient

- Programme to implement a research and innovation pipeline, from bench to bedside

- Integrative programme linking major EU and national initiatives – R&D, research infrastructures, registires

- Bridging to ERNs to help implementing research results and taking lessons learned from the clinic back to the bench

Key action 1

Transnational calls for proposals to fund rare diseases research.

Joint funding by EC and national funding agencies.

Key action 2

Virtual platform for coordinated access, data exchange and repository facilities building on existing resources.
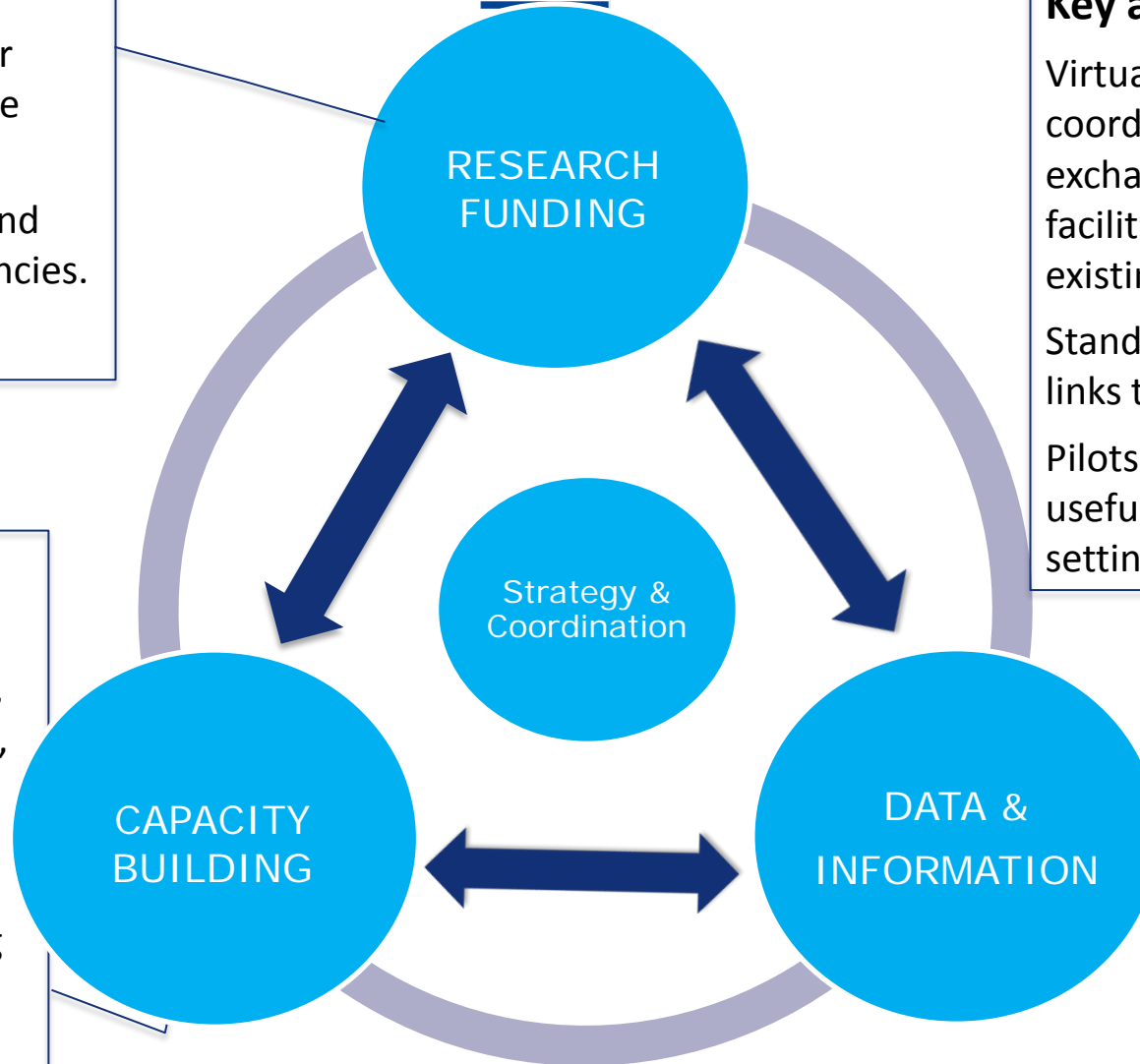
Standards, analysis tools, links to care data.

Pilots to ensure usefulness in clinical setting/ERNs.

Key action 3

Training and support on data management, product development, translational research etc for stakeholders including patient organisations. Sharing best practices.

Tech transfer facility towards industry.

European Commission

RESEARCH FUNDING

Strategy & Coordination

CAPACITY BUILDING

DATA & INFORMATION

What's next ……….

Support IC PerMed Action Plan

Regional activities – smart specialisation

Piloting Personalised Medicine in health care – strengthen evidence base – technical feasibility and financial viability

Data sharing – standards, interoperability, sound legal and ethical frameworks – research – health data

"Bench to bedside" – rare diseases as model

Decision support tools for health care providers

-omics for prediction and prevention (incl. microbiome, epigenome, etc.)

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http://ec.europa.eu/programmes/horizon2020/

http://ec.europa.eu/research/health/index.cfm?pg=home

Funding opportunities:

http://ec.europa.eu/research/participants/portal/desktop/en/home.html