

# Luxembourg Centre for Systems Biomedicine



# Primary activities related to genome medicine in Luxembourg

## Establishment of a :

- National Centre of Excellence in Research in PD (NCER-PD)
- National Cohort of PD patients (HELP-PD)

## European projects - Focus on Curation and Standardisation:

- Integration of biological and clinical data (ETRIKS/TranSMART)
- Mechanism-based Aetiology (AETIONOMY)

## Preparation for a :

- “ELIXIR”-node for Translational Medicine IT infrastructure

# National Centre of Excellence in Research



Duration: 8 + 4 years  
Funding: 16 + 8 Mio €



## National Centre of Excellence in Research

*on the topic of*

### Early Diagnosis and Stratification of Parkinson's Disease

*coordinated by*

Professor Rudi Balling,  
Luxembourg Centre for Systems Biomedicine.

*Submitted on behalf of the institutions*

University of Luxembourg,  
Centre Hospitalier de Luxembourg,  
Centre de Recherche Public de la Santé,  
Integrated BioBank of Luxembourg,

*with the partner institutions*

Paracelsus-Elena-Klinik, Kassel,  
Philipps-Universität Marburg,  
Ruhr-Universität Bochum,  
Universitätsklinikum Tübingen.



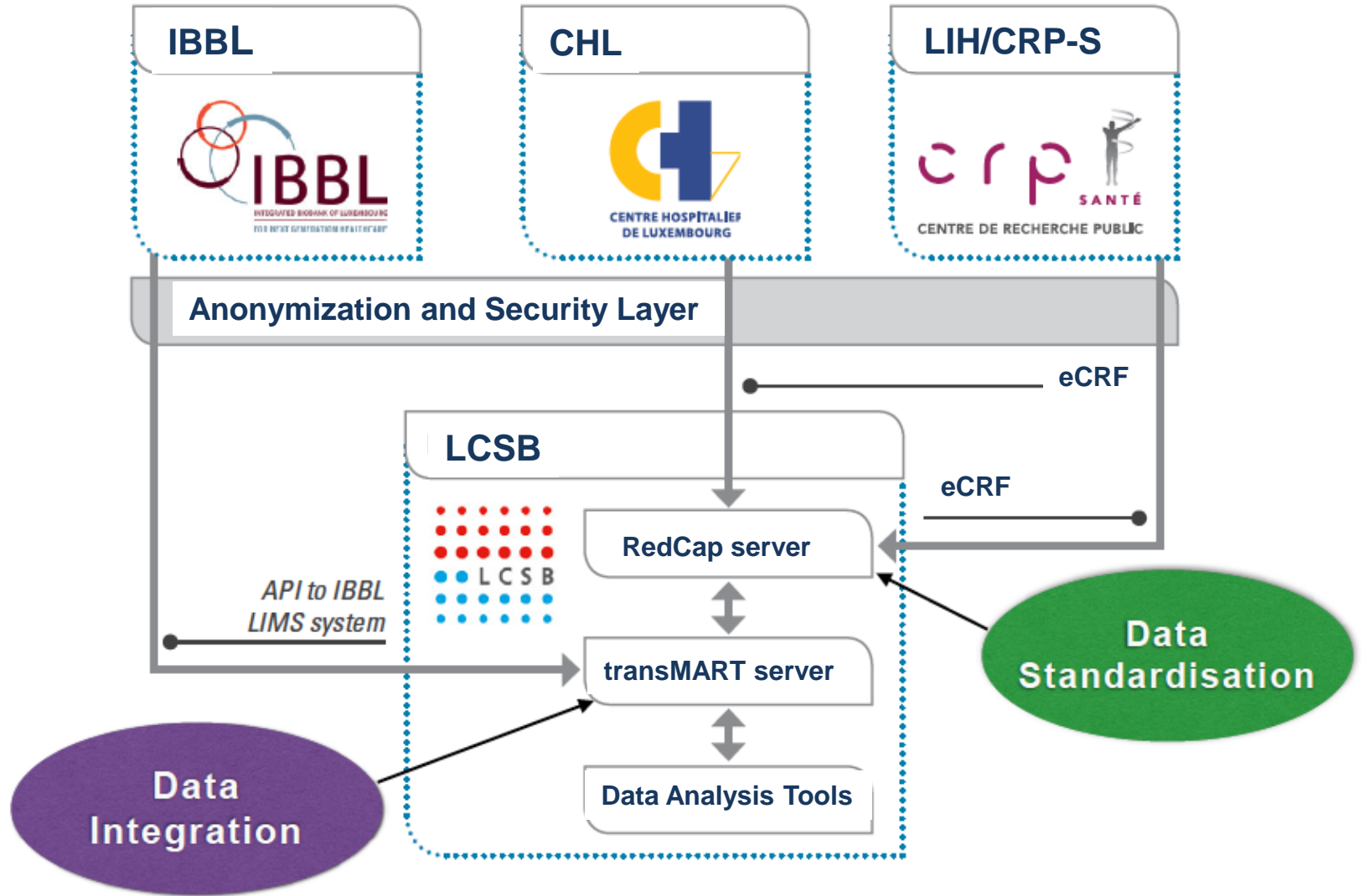
# National Longitudinal Parkinson's Disease Cohort

## Health of the **Elderly Luxembourgish Population** (**HELP-PD**)



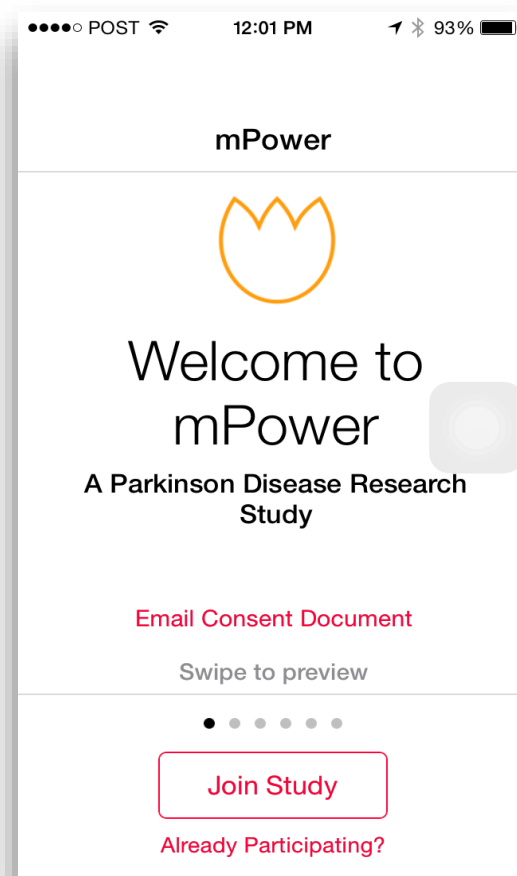
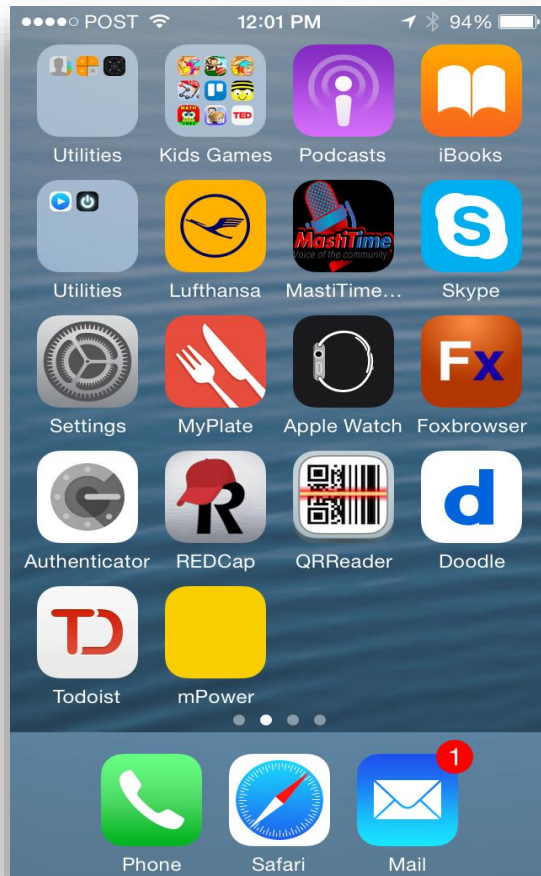
- Genetic Parkinson Disease patients
- Sporadic Parkinson Disease patients
- Enriched risk subjects
- Healthy ageing controls

# Data Integration & Data Standardization



# Collaboration with SAGE Bionetworks to use mPower mobile app

(Mobile Parkinson Observatory for Worldwide Evidenced-based Research)



# Key accomplishments related to genomic medicine

nature  
genetics

Nava, C. . et al. 2014

*De novo* mutations in *HCN1* cause early infantile epileptic encephalopathy

nature  
genetics

Schubert, J. . et al. 2014

Mutations in *STX1B*, encoding a presynaptic protein, cause fever-associated epilepsy syndromes

nature  
genetics

Syrbe S. et al. 2015

*De novo* loss- or gain-of-function mutations in *KCNA2* cause epileptic encephalopathy

## Integrating Pathways of Parkinson's Disease in a Molecular Interaction Map

Kazuhiro A. Fujita • Marek Ostaszewski • Yukiko Matsuoka • Samik Ghosh • Enrico Glaab • Christophe Trefois • Isaac Crespo • Thanneer M. Perumal • Wiktor Jurkowski • Paul M. A. Antony • Nico Diederich • Manuel Buttini • Akihiko Kodama • Venkata P. Satagopam • Serge Eifes • Antonio del Sol • Reinhard Schneider • Hiroaki Kitano • Rudi Balling

Metabolomics (2015) 11:603-619  
DOI 10.1007/s11306-014-0721-3



ORIGINAL ARTICLE

## Prediction of intracellular metabolic states from extracellular metabolomic data

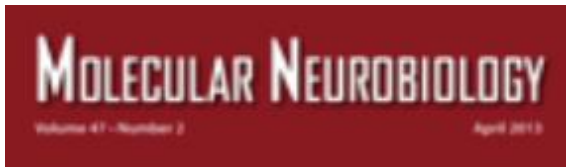
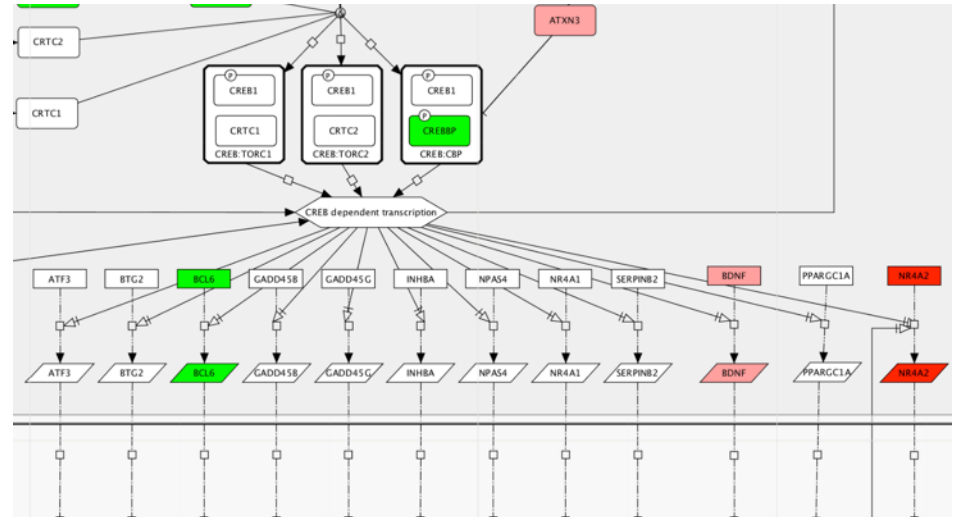
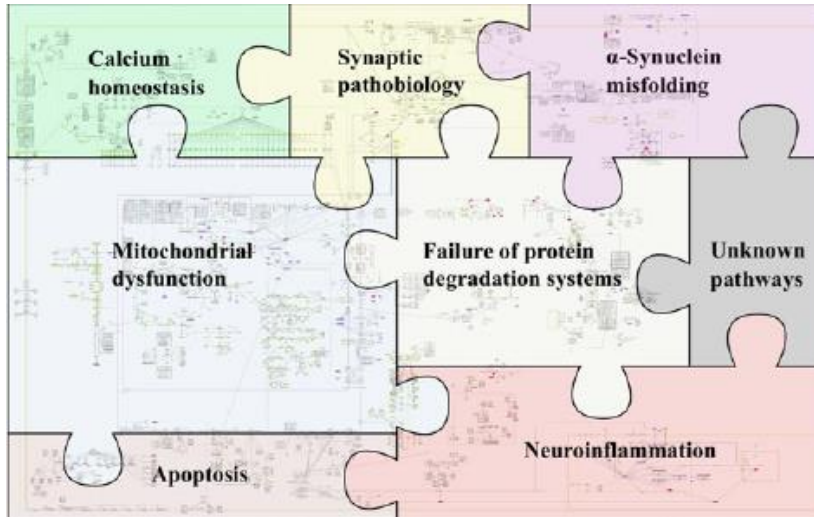
Maike K. Aurich • Giuseppe Paglia • Óttar Rólfsson • Sigrún Hrafnadóttir • Manuela Magnúsdóttir • Magdalena M. Stefaniak • Bernhard Ø. Palsson • Ronan M. T. Fleming • Ines Thiele

A constraint-based modelling approach to metabolic dysfunction in Parkinson's disease

Longfei Mao <sup>a</sup>, Averina Nicolae <sup>a</sup>, Miguel A.P. Oliveira <sup>a</sup>, Feng He <sup>a,b</sup>, Siham Hachi <sup>a</sup>, Ronan M.T. Fleming <sup>a,\*</sup>

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# A community driven Parkinson's disease map



Vol. 49, 88-102, 2014

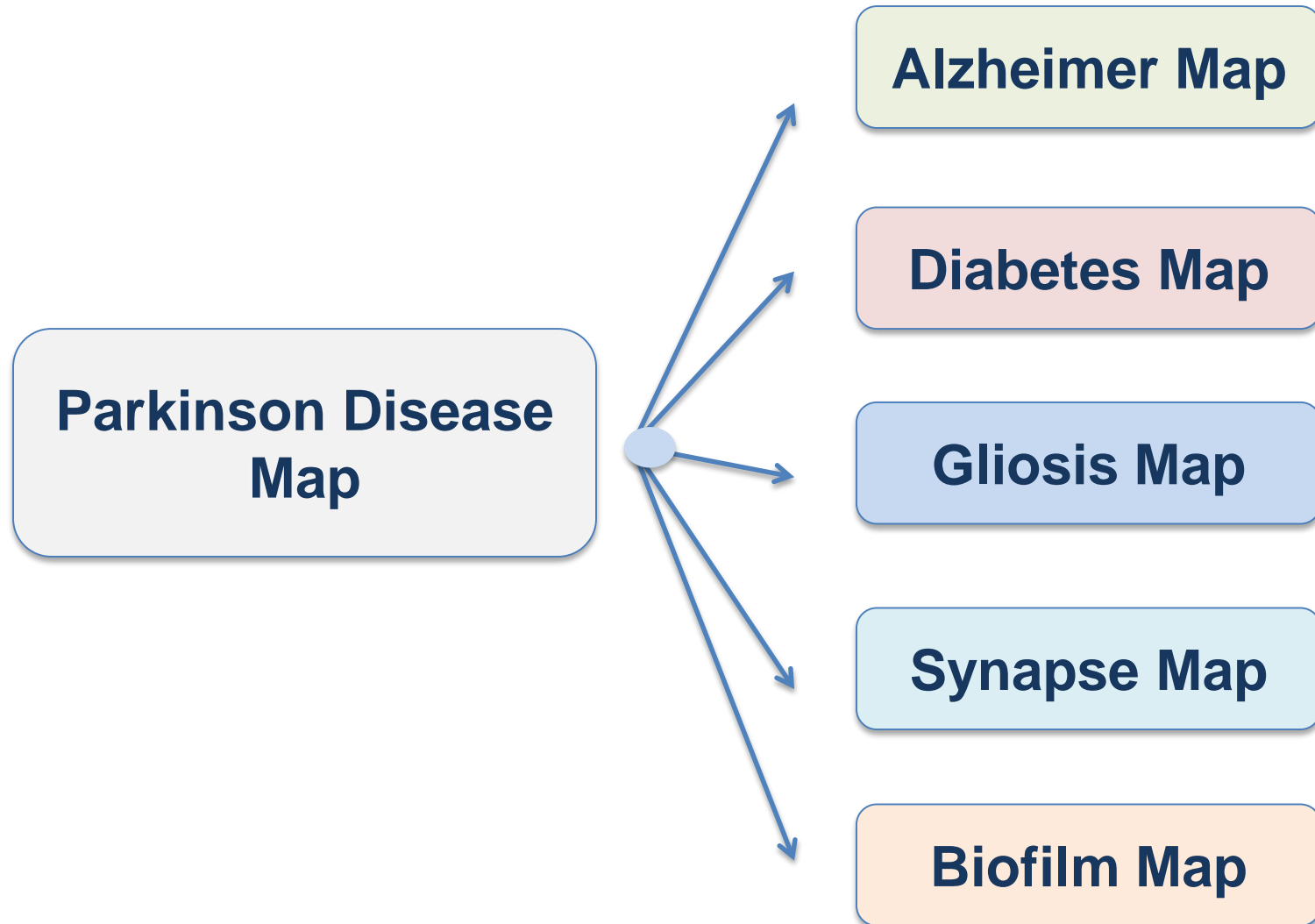
[http://pdmap.uni.lu/pd\\_map/](http://pdmap.uni.lu/pd_map/)

## Integrating Pathways of Parkinson's Disease in a Molecular Interaction Map

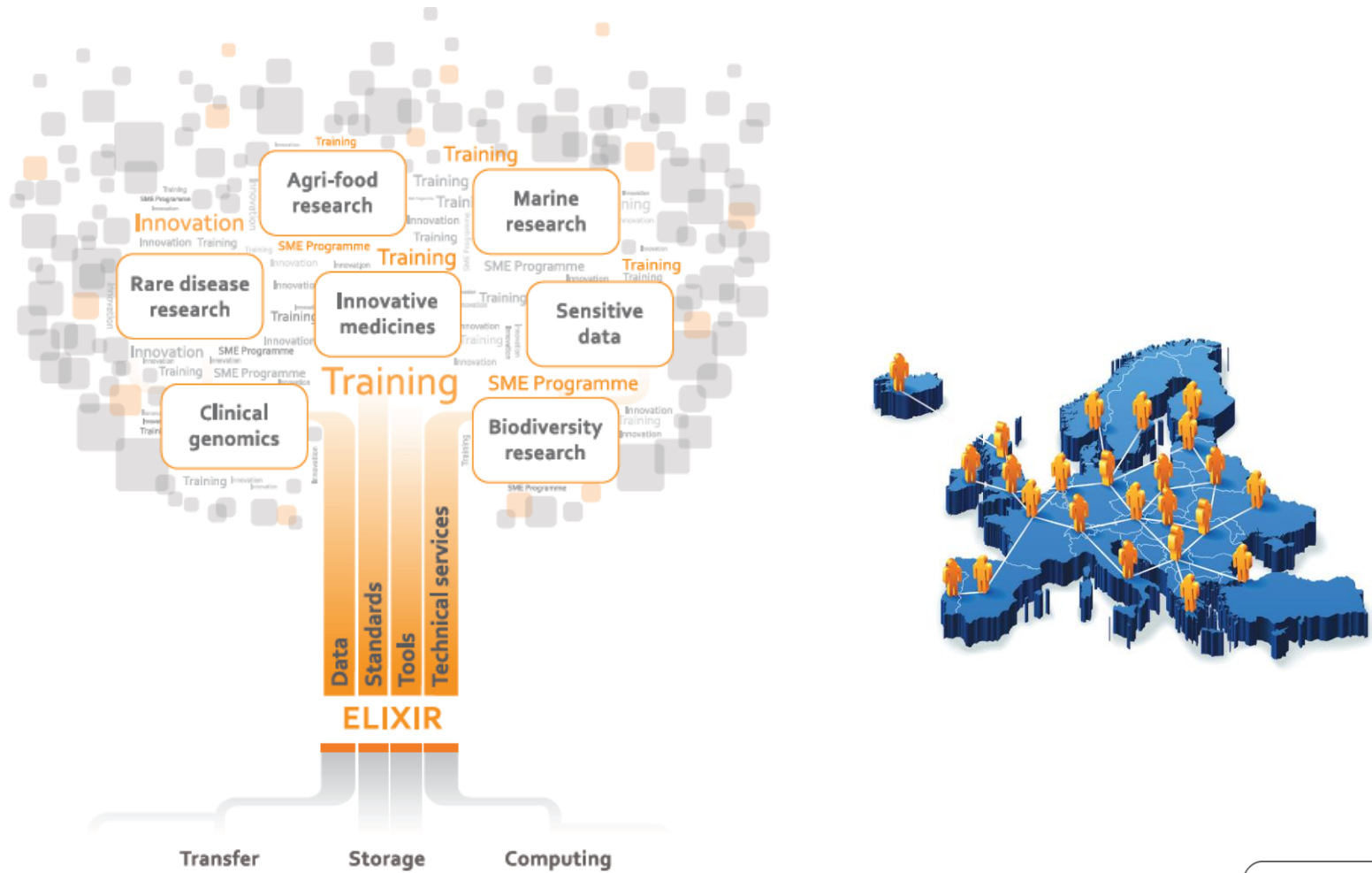
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# Developing a general concept of disease maps



# ELIXIR – A European infrastructure for life sciences



# Potential collaborations

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- **Whole genome sequencing & genome analysis projects**
- **Bio- and medical informatics**
- **Computational disease modeling**
- **Cellular & animal models of neurodegenerative disease**