Luxembourg Centre for Systems Biomedicine

LCSB 1

LCSB 2
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

IBBL

<table>
<thead>
<tr>
<th>API to IBBL LIMS system</th>
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<tbody>
<tr>
<td>LCSB</td>
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<tr>
<td>RedCap server</td>
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<td>transMART server</td>
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CHL

Data Analysis Tools

LIH/CRP-S

Anonymization and Security Layer

eCRF

Data Standardisation
Collaboration with SAGE Bionetworks to use mPower mobile app

(Mobile Parkinson Observatory for Worldwide Evidenced-based Research)
Key accomplishments related to genomic medicine

De novo mutations in HCN1 cause early infantile epileptic encephalopathy

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

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 Glarb · Christophe Trefois · Isaac Crespo · Thanheer M. Perumal · Wiktor Jurkowski · Paul M. A. Antony · Nico Diederich · Manuel Buttini · Akihiko Kodama · Venkata P. Satagopam · Serge Elies · Antonio del Sol · Reinhard Schneider · Hiroaki Kitano · Rudi Balling

Prediction of intracellular metabolic states from extracellular metabolomic data

Malke K. Aurich · Giuseppe Paglia · Óttur Rolfsón · Sigrún Hrafnsdóttir · Manuela Magnúsdóttir · Magdalena M. Stefaniak · Bernhard O. Palsson · Ronan M. T. Fleming · Ines Thiele

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

Longfei Mao *, Averina Nicolae *, Miguel A.P. Oliveira *, Feng He *a, Siham Hachi *, Ronan M.T. Fleming *a

*a Luxembourg Centre for Systems Biomedicine (LCSB), University of Luxembourg, 7, avenue des Haies-Romains, L-4362 Esch-sur-Alzette, Luxembourg
b Department of Infecion and Immunity, Luxembourg Institute of Health (LIMH), 20, rue Hériti Koch, L-4354 Esch-sur-Alzette, Luxembourg
A community driven Parkinson’s disease map

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http://pdmap.uni.lu/pd_map/
Developing a general concept of disease maps

- Parkinson Disease Map
- Alzheimer Map
- Diabetes Map
- Gliosis Map
- Synapse Map
- Biofilm Map
ELIXIR – A European infrastructure for life sciences
Potential collaborations

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