PhenoMeNal: a gateway to personalised medicine

***PhenoMeNal, an EU-funded project launched today and led by EMBL-EBI, is creating a secure, on-demand e-infrastructure for clinical metabolomics data.***

* PhenoMeNal project to build an e-infrastructure for clinical metabolomics data;
* 13 partners, coordinated by EMBL-EBI and funded with €8 million over 3 years by the EU’s Horizon2020 Programme;
* E-infrastructure will make it possible to analyse large-scale metabolomic and genomic data, improving our ability to detect, treat and manage disease.

1 September 2015 – Today marks the launch of PhenoMeNal, a project to create an e-infrastructure to analyse metabolomic data from clinical and population studies. An international endeavour led by EMBL-EBI, PhenoMeNal is funded with €8 million from the EU’s Horizon2020 Programme to bring state-of-the art methods for understanding the molecular drivers of health, healthy ageing and disease into the clinic.

A large number of European citizens are expected to have their genomes sequenced and added to their medical records over the next decade, but understanding the totality of environmental and genetic factors on a person’s health – in a measurable way – is crucial for providing evidence-based treatment when things go wrong. Linking genotype and metabolome data with other factors influencing a patient’s health captures a surprising amount of this highly dynamic information, at relatively low cost. But the data generated is so incredibly vast that the current infrastructure cannot cope on anything close to European scale.

“A typical national phenotyping centre might manage around 100,000 human samples per year, which generates petabytes of data,” explains Christoph Steinbeck of EMBL-EBI, “The challenge is to standardise the way scientists can access and analyse such large amounts of data. Making personalised medicine feasible for Europe is a major undertaking, and it requires that we create the infrastructure to work on exabyte-scale amounts of biomedical phenotyping data. This can’t be done off-the-shelf – we need to create a viable solution.”

The main goal of PhenoMeNal is to develop and deploy the PhenoMeNal Gateway: an e-infrastructure that makes it feasible for researchers and healthcare providers to process, analyse and mine medical molecular phenotyping and genotyping data. To ensure the service is truly useful, the partners are committed to making it integrated, secure, permanent, privacy-compliant, available on-demand and sustainable.

“We want to be sure that molecular information is captured from the beginning, when a patient first participates in testing,” says Jeremy Nicholson of Imperial College, London. “It’s really important to us to make sure there can be a water-tight audit trail for patients’ metabolic phenotyping data, from collection all the way to biomedical insights and, hopefully, accurate diagnosis.”

The project builds on the previous success of COSMOS (Coordination of Standards in Metabolomics) and its worldwide network for the exchange of metabolomics metadata, MetabolomeXchange.org. Partners include world-leading groups with long experience in developing tools and methods for acquiring, integrating and analysing very large datasets on metabolic phenotypes and genomes, and groups that specialise in High Performance and Distributed Computing.

“The PhenoMeNal Gateway has to be both high-performance and easy to use by people working in many different settings and languages,” says Christoph. “Our approach is to make a federated and secure system, so we’ll be creating open-source solutions and building on the hard-earned progress already made by the community.”

Transmitting datasets of this size between institutions is not yet feasible in any reasonable time frame, so PhenoMeNal partners will be drawing on virtual machines and large-scale technologies such as EMBL-EBI’s Embassy Cloud. The work will be done in consultation with European Grid and Cloud Infrastructures as well as national, European and international grid initiatives, so that ultimately the PhenoMeNal Gateway will be useful on a global scale.

# **About PhenoMeNal**

PhenoMeNal partners include the European Bioinformatics Institute (EMBL-EBI), Imperial College of Science, Technology and Medicine, the University of Birmingham and the University of Oxford in the UK; the Biobanking and BioMolecular Resources Research Infrastructure in Austria; the University of Alberta, Canada; Commissariat à l'énergie atomique et aux énergies alternatives (CEA) and Institut National de la Recherche Agronomique (INRA) in France; Leibniz-Institute of Plant Biochemistry in Germany; Consorzio Interuniversitario Risonanze Magnetiche di Metallo Proteine in Italy; Universiteit Leiden in the Netherlands; Universitat de Barcelona in Spain; Uppsala Universitet in Sweden; the SIB-Swiss Institute of Bioinformatics in Switzerland; and SRI International in the US.