EFMI Working Group on Translational Health Informatics – Workshop on How Informatics Can Mitigate Translational Medicine Barriers

Amnon SHABO (SHVO)\textsuperscript{a,1}, Alberto MORENO-CONDE\textsuperscript{b}, Carlos PARRA-CALDERÓN and Mauro GIACOMINI\textsuperscript{d}

\textsuperscript{a}University of Haifa
\textsuperscript{b}Digitalica Health
\textsuperscript{c}University of Applied Sciences Western Switzerland
\textsuperscript{d}University of Genoa, Italy

Abstract Translational medicine is a relatively new discipline targeted at removing barriers in utilizing new scientific discoveries in healthcare, moving from bench to bedside, on to communities and finally to policy. The biomedical discoveries need to be accompanied by studies in areas such as health economy as well as sociological and psychological aspects. In order to manage the information generated by each of these disciplines, it is crucial to use advanced informatics that could fuse the various types of data into computerized systems that support the translational processes. This workshop aims at discussing these challenges.

Keywords. Translation Research, Translational Medicine, Translational Informatics, Biomedical Informatics, Clinical Genomics, Genomic and Precision Medicine

Introduction of the topic

The intended audience for this workshop is biomedical informaticians who nowadays play a major role in the loop of bench to beside and back to the bench. These experts bridge the gaps between bioinformaticians and medical informaticians, as well as clinicians in healthcare and scientists in research.

The knowledge about human health is accumulating rapidly, with enormous amounts of new types of data, in which most of the data items are currently of unknown significance, but with the recognition that they are determinants to our health (e.g., genomics). The classical methodology of controlled clinical trials is not sufficient anymore and there is a need to refine traditionally-established evidences with results of data-driven analytics. Combined together, new insights are gained that could help out at the point of care, in cases where existing clinical guidelines fall short of providing precise advice on the decision needed to be taken.

\footnote{Corresponding Author.}
1. Aim of the discussion

This workshop will be focused on the health informatics needed for accommodating the translational challenges. We will strive for active discussion with the audience on the core challenges of translational health informatics.

In particular, the following issues will be discussed: (1) Explore a proposal for a Translational Health Information Language (see details at http://www.epmajournal.com/content/5/S1/A51), which can underlie translational infostructure; (2) Study feasibility of common translational infostructure for the currently distinct environments of biomedical research and health care; (3) The nature of informatics foundation needed to underlie translational research and medicine; and (4) How can bioinformatics and medical informatics be better interrelated, in particular in light of the new OMICS era yielding enormous amounts of data of yet unknown significance.

2. Contribution from each speaker

Amnon Shabo will discuss the roadmap towards a Translational Health Information Language, described in more detail at his recent paper in the European Personalized Medicine Association Journal. He will show how such a language could underlie a common infrastructure for both healthcare and research, using a warehousing/marts architecture where the warehouse information model is developed based on constraining international standards and interrelating them with each other to fit to the translational goals.

Alberto Moreno-Conde will explain a set of initiatives carried out as part of the IS13606 Cost Action about public-private initiatives for direct to consumer genetic testing. This project will develop a proof of concept for information modeling within the translational informatics field to identify the most common barriers to information flow and possible mechanism to reduce these gaps.

Carlos Parra-Calderon will present the information structure developed within Virgen del Rocío University Hospital to support translational research based on opensource solutions and the reusability of healthcare information.

Mauro Giacomini will discuss the results of the EATRIS (European Infrastructure for Translational Medicine) project and of its national follow-ups. He will consider the possibility to link this working group with the EATRIS network. The main aim of this collaboration could be to disseminate and to currently apply the outcomes of the discussions within the EATRIS group in the platforms proposed the EATRIS network.

In order to foster participation of the attendants each presentation will raise a set of topics that will be discussed with the aim of promoting the adoption of a Roadmap for Translational Health Informatics and a questionnaire will be distributed to determine the priorities that need to be addressed at the current stage.

3. Expected results

Workshop results will further inform the translational informatics community on possible ways to fuse bioinformatics and medical informatics along with other types of information (e.g., economic and socio-psychological aspects) into an integrative info-
structure that can serve both research projects as well as warehousing efforts within the premises of healthcare providers.

In particular, we expect to have results on the roadmap towards a Translational Health Information Language (THIL), through touch point between common and emerging languages, e.g., nano-publications (structured asserts made in scientific literature) can point to the assertion’s provenance through the use of the ISA (Investigation-Study-Assay) format that provide extensive metadata on biological assays.

In addition, emerging ‘bridge’ standards will be discussed, such as the Genetic Testing Report, Diagnostic Imaging Report and Personal Healthcare Monitoring Report. These are standards that can be consumed by healthcare information systems on the one hand, but also encapsulate key chunks of raw / mass data sets obtained through high-throughput omics assays, sensor streaming data and advanced imagining techniques.

The foreseeable collaboration with EATRIS could make possible some applications of the THIL within current researches in this community, in particular, the info-structure component of the broader translational medicine infrastructure.