# Winter School "Clinical and Genetic Epidemiology – Strategies to Drive Personalized Medicine"

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Personalized medicine is a medical procedure that **separates** patients into different groups - with medical decisions, practices, interventions and/or products being tailored to the individual patient based on their predicted response or risk of disease. The terms personalized medicine, precision medicine, stratified medicine and P4 medicine are used interchangeably to describe this concept though some authors and organisations use these expressions separately to indicate particular nuances. While the tailoring of treatment to patients dates back at least to the time of Hippocrates, the term has risen in usage in recent years given the growth of new diagnostic and informatics approaches that provide understanding of the molecular basis of disease, particularly genomics. This provides a clear evidence base on which to stratify (group) related patients.

(Wikipedia, accessed Feb 5, 2017)

#### **Development of concept**

In personalised medicine, diagnostic testing is often employed for selecting appropriate and optimal therapies based on the context of a patient's genetic content or other molecular or cellular analysis. The use of genetic information has played a major role in certain aspects of personalized medicine (e.g. pharmacogenomics), and the term was first coined in the context of genetics, though it has since broadened to encompass all sorts of personalization measures.

(Wikipedia, accessed Feb 5, 2017)

Personalized medicine is an emerging practice of medicine that uses an individual's genetic profile to guide decisions made in regard to the prevention, diagnosis, and treatment of disease. Knowledge of a patient's genetic profile can help doctors select the proper medication or therapy and administer it using the proper dose or regimen. Personalized medicine is being advanced through data from the Human Genome Project.

(Genetics Home Reference, U.S. National Institutes of Health, previous definition.

Now preferred term: Precision medicine)

Personalized medicine is the use of detailed information about a patient's genotype or level of gene expression and a patient's clinical data in order to select a medication, therapy or preventative measure that is particularly suited to that patient at the time of administration. The benefits of this approach are in its accuracy, efficacy, safety and speed. The term emerged in the late 1990s with progress in the Human Genome Project. Research findings over the past decade, or so, in biomedical research have unfolded a series of new, predictive sciences that share the appendage omics (genomics, proteomics, metabolomics, cytomics). These are opening the possibility of a new approach to drug development as well as unleashing the potential of significantly more effective diagnosis, therapeutics, and patient care.

(www.biomedicine.org)

A form of medicine that uses information about a person's genes, proteins, and environment to prevent, diagnose, and treat disease. In cancer, personalized medicine uses specific information about a person's tumor to help diagnose, plan treatment, find out how well treatment is working, or make a prognosis. Examples of personalized medicine include using targeted therapies to treat specific types of cancer cells, such as HER2-positive breast cancer cells, or using tumor marker testing to help diagnose cancer. Also called precision medicine.

(NCI Dictionary of Cancer Terms)

Briefly, P4 medicine describes a systems approach that includes predictive, personalized, preventive, and participatory aspects. This approach extends beyond genomic medicine because "genes and their products almost never act alone, but in networks with other genes and proteins and in context of the environment." P4 medicine proposes to integrate numerous biologic data points "including longitudinal molecular, cellular, and phenotypic measurements as well as individual genome sequences" to better define health or wellness for each person, to predict disease transitions, and to target medical interventions. The premise is that P4 medicine will lead to powerful new diagnostics and therapeutics for treatment and prevention, based on each person's unique biologic characteristics (e.g., inherited variation to drug response) and disease processes (e.g., tumor genomic characteristics). (Khoury et al., A population approach to precision medicine, Am J Prev Med 2012;42:639-645)

# **Topics covered in the Winter School**

- Cancer Genomics
- Predictive Medicine
- Prediction Models
- Clinical Studies and Translational Medicine
- Ethical Aspects of Clinical and Genetic Epidemiology
- Health Economics
- Metabolomics
- Pharmacogenomics
- Statistics for High-Dimensional Data
- Basic NGS Data Processing Exercise
- Personalized Medicine in Cardiology
- Data Integration and Knowledge Exploration
- Direct-to-consumer genetic testing

### Short summaries for each day

- A team of 2-3 participants prepares a short summary of the key messages & most relevant statements of a particular day of the Winter School
  - a short summary of 5 min will be given by this team at the beginning of the first morning session on the next day (short & simple, no slides), with additional remarks by the other participants.
- The same team will provide a longer summary with slides (15 min) in the wrap-up session, morning of last Friday (February 17th), followed by additoinal remarks and discussion by the other participants.



- Participants who want to earn 3 ECTS credit points have the opportunity to take a written exam on the last day of the Winter School
  - Friday, February 17th, 2016, afternoon (14:15-15:45)
- Prerequisite is attendance of the Winter School (with at most two days of absence).