



21 – 25 AUGUST 2017 / UNIVERSITY OF COPENHAGEN

# HUMAN GENOME VARIATION

## — A KEY CORNERSTONE OF PRECISION MEDICINE

**Precision medicine is an emerging approach for disease treatment and prevention that takes into account individual variability in genes, environment, and lifestyle for each person. Genome variation is one of the most frequently measured individual variations and a cornerstone of precision medicine.**

Understanding the nature and consequences of genome variations and translating them into clinical practice is an important aspect of precision medicine and it is of interest not only for all the medical and laboratory specialties and pharmaceutical industry, but to an extent for the whole society.

Understanding genome variations in disease and health is thus of crucial importance as also reflected by establishment of large scale national projects around the world. Very recently, December 2016, the Danish Government and the Regions announced a strategy for establishment of a National Genome Center, which emphasises that genome variation studies will be a focus point in future research and health practice.

### WHAT YOU WILL LEARN

- Basic concepts of precision medicine with focus on genome variation
- Genetic and epigenetic variations and their interplay in disease pathology
- The current methods and technologies to investigate genome variations
- Different public genome variation databases
- Bioinformatic tools and data analysis, including handling of big data
- Data storage and protection – Danish Computerome project
- To evaluate the consequences of genome variations in health and disease

- Current applications of genome variations in precision medicine: Examples from cancer disorders, immunology, psychiatry
- How to translate genetic knowledge to clinical practice, inclusion of the patients/individuals in the process, genetic counselling
- Ethical and juridical aspects of genome variation studies

### COURSE CONTENT

This course will provide insights into key principles of the human genome and a practical understanding of the consequences of genome variations. The course will also cover technical tools, bioinformatics, data storage and projection, ethical and juridical aspects of genome research and inclusion of patients in decision processes. The teaching style is interactive and includes a combination of lectures, discussions, group work and case studies.

*The topics include:*

- Genome variations, pathogenic versus benign
- Genome variation databases
- Architecture of the genome and its complexity
- Genome variation and evolution
- Methods and technologies to investigate genome variations
- National Genome Projects of different countries, including the Danish Genome Project, and importance of these projects in precision medicine
- Bioinformatic tools and data analysis i.e. handling of big data

**“Fantastic program and faculty. Best update I have had in years. Great value for money.”**

*Former participant on a Copenhagen Summer University course*



- Data storage and protection via Computerome
- Ethical and juridical aspects of large scale genome variation studies
- Current applications of genome variations in precision medicine
- Inclusion of patients

#### PARTICIPANTS

The course is for professionals working in the field of medicine, bioinformatics or related health sciences (e.g. public health, human biology and biotechnology), persons with special interest in technical aspects of genome variation and persons from the pharmaceutical industry. The course offer an introduction for those less familiar with human genome variation and will provide a deeper understanding for health professionals with experience within the field. Participants must hold at least a relevant bachelor degree.

#### COURSE DATES

5 days, 21 – 25 August 2017, 9:00 – 16:30 at the University of Copenhagen, Frederiksberg Campus.

#### COURSE DIRECTOR

**Zeynep Tümer**, DMSc, MD, PhD, Kennedy Center, Department of Clinical Genetics, Copenhagen University Hospital, Rigshospitalet

#### Co-directors

**Finn Cilius Nielsen**, Professor, DMSc, Center for Genomic Medicine, Copenhagen University Hospital, Rigshospitalet

**Niels Tommerup**, Professor, DMSc, Wilhelm Johannsen Center for Functional Genome Research, Institute of Cellular and Molecular Medicine, University of Copenhagen

#### OTHER COURSE TEACHERS (in alphabetical order)

**Søren Brunak**, Professor, Research Director, Novo Nordisk Foundation Center for Protein Research, University of Copenhagen; Center for Biological Sequence Analysis, Department of Systems Biology, Technical University of Denmark (DTU)

**Johan den Dunnen**, Professor, Leiden University Medical Center and Human Genome Variation Society, The Netherlands

**Magnus Fontes**, Professor, Director of the Lund University Centre for Data Analysis, Sweden. Founder of the bioinformatics software company Qlucore. Head of International Group for Data Analysis, Institut Pasteur, Paris, France

**Anne-Marie Gerdes**, Professor, Head of Department of Clinical Genetics Copenhagen University Hospital, Rigshospitalet. Member of The Danish Council on Ethics (Det Etske Råd)

**Anders Johannes Hansen**, Associate Professor, Natural History Museum of Denmark

**Torben Hansen**, Professor and Acting Scientific Director, Novo Nordisk Foundation Center for Basic Metabolic Research Metabolic Genetics, University of Copenhagen

**Anna Lindstrand**, Senior Researcher, Department of Molecular Medicine and Surgery, Karolinska Institute, Sweden

**Jens Lundgren**, Professor, Department of Infectious Diseases, Copenhagen University Hospital, Rigshospitalet. Director of CHIP, Centre of Excellence for Health, Immunity and Infections. Head of PERSIMUNE

**Thorkild I.A. Sørensen**, Professor, Center for Basic Metabolic Research, University of Copenhagen

**Thomas Werge**, Professor: Institute of Biological Psychiatry, Mental Health Centre, Sct. Hans, MHS, Capital Region of Denmark. Institute of Clinical Sciences, Faculty of Medicine and Health Sciences, University of Copenhagen

**Torben Franck Ørntoft**, Professor, Head of Department of Molecular Medicine, Aarhus University Hospital

#### COURSE FEE

EUR 2,600/DKK 19,000 (excl. Danish VAT 25%). Fee includes teaching, course materials and all meals during the course.

Approved by the Organisation of General Practitioners in Denmark. If you are a member of the Organisation of General Practitioners in Denmark (PLO) you can apply for reimbursement.

FOR MORE INFORMATION AND REGISTRATION:  
[copenhagensummeruniversity.ku.dk](http://copenhagensummeruniversity.ku.dk)

