

MEDGENET NEWSLETTER Issue III

June 2017

Dear MEDGENET friends,

We are already halfway through the project and we would like to share our progress with you. Mentoring secondments, project workshops and courses helped us to improve the research capacity of CEITEC MU as well as advance research in the field of Medical Genomics and its applicability for routine clinical practice.

We would also like to inform you about the project activities that occurred during the last half year: we've organised our first summer school as well as several work-

shops, courses, and mentoring secondments. You can also find information about our plans for upcoming events during the Autumn semester of 2017.

MEDGENET team



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This project has received funding from the *European Union’s Horizon 2020 research and innovation programme* under grant agreement No 692298. This Newsletter reflects only the author’s view and the Research Executive Agency is not responsible for any use that may be made of the information it contains.

HALFWAY THROUGH THE PROJECT

During the first half of the project, the MEDGENET consortium focused on the main project aim, which is creating a well-educated taskforce of bio-medical researchers while also strengthening CEITEC's capacity and scientific performance. To connect scientists, researchers and students from all partner institutes and facilitate discussions and future collaborations, workshops focusing on areas such as high performance, high-throughput bio-informatics and immunogenetics were organized and held at CERTH, Thessaloniki, Greece and Uppsala University (UU), Sweden, respectively. In addition, four workshops were co-organised by the Host University, CEITEC Masaryk University together with other local partners in Brno, Czech Republic. To advance their skills and knowledge in the area of genomics and next-generation sequencing (NGS), seven young researchers participated in EMBL courses with an additional course organised at CEITEC MU together with CERTH. To facilitate knowledge transfer and the adoption of new techniques within the host institute, CEITEC MU, fourteen secondments were arranged within the mentoring programme. These activities significantly contributed to the improvement of research quality at CEITEC MU and helped to expand and strengthen existing collaborations. Collectively, the MEDGENET mentoring secondments and workshops offered CEITEC MU researchers various opportunities to familiarize themselves with a plethora of advanced wet-lab protocols, computational and bioinformatics tools (e.g. emerging NGS platforms, RNA-Seq data analysis, cloud computing for high-throughput bioinformatics studies, etc.), which have now been implemented in ongoing research projects. As an illustrative example, CEITEC MU researchers learned how to use state of the art RNA-Seq tools and gained experience in working with 'delicate' RNA samples. Through attendance at courses and secondments at EMBL Heidelberg and EMBL-EBI, young researchers have obtained or upgraded their skills in metagenomics, microbiome analysis, and protein-protein interaction analysis. The activities of the MEDGENET project have also focused on the development of new bioinformatics tools that not only will assist in the



analysis of data for translational research projects but also could eventually be applicable and beneficial for routine clinical practice. Collaborations with UU, CERTH, and EMBL also brought advancement in the area of immunogenetics and laboratory-based procedures for library preparation.

Another important aspect of this project is to not only strengthen and expand existing collaborations but also form new collaborative links between partner institutions. The intensive exchange of scientists within the MEDGENET consortium, as a result of the activities summarized above, strengthened existing partnerships that have now progressed into more practical collaborations. This will result in the further advancement of genomic research not only at CEITEC MU, but also throughout Europe. To date, several collaborative research projects are being prepared that will lead to joint publications. A joint H2020 has already been submitted and several joint publications have already been published in well-known journals in the field of genomics and hematology, including *Haematologica*, *Leukemia*, and *Epigenetics*.

Additionally, being a young research institute, CEITEC MU faces the challenge of increasing its visibility and proving its competitiveness. Besides scientific dissemination and project development, the MEDGENET project also seeks to increase awareness and the reputation of CEITEC via participation in international conferences, contributing to events for general public, publication of a regular newsletter, etc.

NEW COLLABORATIONS

In addition to strengthening and expanding existing collaborations, the MEDGENET project also facilitated the initiation of new collaborative links. The main source of connecting researchers were mentoring secondments and MEDGENET workshops.

During the implementation of the MEDGENET project, new collaborative links were established within the area of the genomic analysis of haematological malignancies. One such example is the analysis of DNA obtained from Czech and Swedish family sample sets to investigate the hereditary component of these malignancies (Sarka Pospisilova (CEITEC MU) and Panagiotis Baliakas (UU). Sarka Pospisilova's team (CEITEC MU) has strengthened the collaboration with Richard Rosenquist's (UU) group in their efforts to decipher the genomic landscape in selected haematological disorders and identify recurrent gene mutations that could be utilized for prognostic stratification in the future.

New projects were also initiated in the field of immunogenetic research, particularly with regards to database construction and are currently being outlined in detail by Sarka Pospisilova's group (CEITEC MU), Kostas Stamatopoulos' group (CERTH) and Richard Rosenquist's group (UU). The MEDGENET Grid and Cloud Technologies for high-performance high-throughput bioinformatics studies workshop' was held at CERTH in December 2016 and resulted in a new collaboration with the EGI Foundation aimed at designing novel solutions for ongoing CEITEC MU research and also discussing opportunities for future cooperation.

Additionally, MEDGENET mentoring programme strengthened collaboration between the ERA Chair Mary O'Connell's group at CEITEC MU and Anders Virtanen's group (UU) in the area of mRNA

stability during the innate immune response. These groups will now explore possible changes in Poly(A)-Specific Ribonuclease (PARN) activity during IFN induction.

The CEITEC MU group of Jan Palecek (Jiri Fajkus research group) and the Haering lab at EMBL Heidelberg have both worked on Structure Maintenance of Chromosome (SMC) complexes for years, however there was no practical collaboration between the groups prior to the MEDGENET project. The MEDGENET mentoring programme enabled these two groups to join their expertise and set new topics for collaboration with future potential for joint paper(s).

The mentoring program also served as an important component for facilitating future collaboration between the group of Dalibor Blazek at CEITEC MU and the Genomics Core facility led by Vladimir Benes at EMBL, Heidelberg. This collaboration will be focused on the preparation of samples for NGS projects and subsequent bioinformatics and statistics analyses.

A new collaboration was also initiated with the Enright group at EMBL-EBI where a young researcher from Marek Mraz's group learned new bioinformatics approaches for the analysis of expression data and their relevance to the B-cell signaling pathways and possible novel targeted therapies. Since many of the tools used during the mentoring visits were developed in the group (e.g. Chimira, Sylamer, or Kraken tools), best practices for running the tools and interpreting the data generated were discussed with the authors. The authors of the tools could see their tools being applied to real-life data, which initiated a close link between the groups. New collaborations are planned for testing these tools and expanding their applications.



MEDGENET NEWS

During the first six months of 2017 the MEDGENET consortium continued its activities devoted to educating and developing expertise amongst young researchers. Two workshops, one summer school, and one course took place. A

number of secondments in the mentoring programme were undertaken, and further EMBL courses were available for PhD students and postdocs. More details on these activities are in the following pages.

Summer school in new approaches in experimental biology and omics

The summer school took place at CEITEC MU from 12th to 16th June 2017. The summer school was dedicated to PhD and Master degree students of biological sciences who are interested in modern approaches of experimental research and practical applications of recent findings in the field of genome and proteome research. Lectures from leading experts from EMBL (Neil Dear, Jan Provaznik) and CEITEC MU (Zbynek Zrala, Vitezslav

Bryja, and Ctirad Hofr) covered theoretical background and principles that are applied in genome manipulation and maintenance. More than 60 students attended the lecture on the first day. Next days were devoted to practical lessons with Neil Dear, Jan Provaznik, Ctirad Hofr and Ondrej Šedo. 11 students were selected based on their motivation letters for participation in the practical sessions.



Protein Data Bank – services, API and related tools workshop

The workshop took place between March 9th - 10th, 2017 at CEITEC MU. The workshop introduced participants to the Protein Data Bank in Europe (PDBe) as a service together with the LiteMol tool (developed as a result of cooperation between PDBe and CEITEC MU). The two-day workshop consisted of theoretical and practical sessions on biomacromolecular and PDBe data

processing. The trainers were CEITEC MU researchers, Dr Radka Svobodova and Lukáš Pravda, and invited expert Dr John Berrisford from EMBL-EBI PDBe. Importantly this workshop facilitated knowledge transfer between method developers and end-users at CEITEC MU and other MU faculties. More than 20 CEITEC MU students and young researchers benefited from this workshop.



The workshop on next-gen sequencing technologies and applications

The workshop was organised for January 18th – 20th, 2017 at CEITEC MU. PhD students and young researchers were first instructed on the basic terminology of NGS sequencing, different applications and technology, by the head of the Genomics Core Facility, Dr Boris Tichy. Participants were also given the opportunity to generate a high quality genomic DNA library using a library preparation kit from KAPA. Due to laboratory capacity, the number of participants for the practical part of the workshop was limited to 16. During the second day, lectures from invited speakers on emerging NGS technologies, basic bioinformatics approaches and practical insights into the cancer biology of leukemia took place. Experts from all three MEDGENET partner institu-

tions were invited to speak: Dr Vladimir Benes, Dr Jan Provaznik, and Dr Jonathan Landry from EMBL, Dr Andreas Agathangelidis and Dr Nikos Papakonstantinou from CERTH, and Dr Lesley-Ann Sutton from UU. The lectures were open to everyone interested in the topics. More than 70 researchers and students attended the lectures in this open part of the workshop.



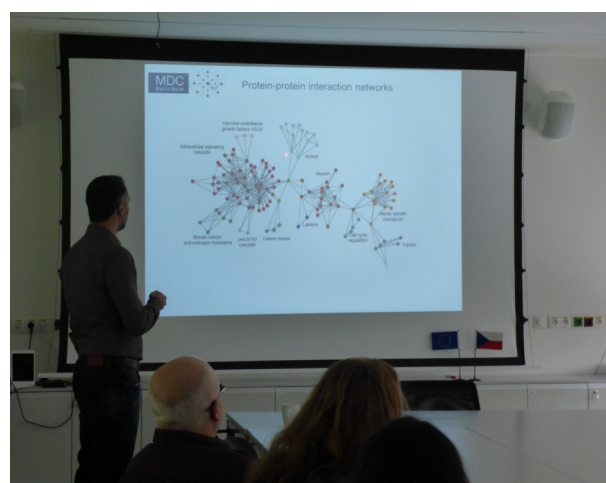
Transcriptome analysis course

The course focused on culturing and handling mesenchymal stem cells (MSCs) which inducibly overexpress the polyglutamine protein ATXN1. In order to investigate the effect of ATXN1 overexpression on the transcriptional profile of these cells, the RNA sequencing analysis had to be performed at different time points. The course was organised by the CEITEC Genomics Core Facility led by Dr Boris Tichy in collaboration with CERTH scientists Stamatia Laidou and Dr Spyros Petrakis.



Guest lecture “Identification of molecular pathogenic mechanisms through protein-protein interaction networks”

In addition to the Transcriptome analysis course, Dr. Spyros Petrakis was willing to present his talk “Identification of molecular pathogenic mechanisms through protein-protein interaction networks” for CEITEC colleagues at the Molecular Medicine research programme.



MEDGENET Mentoring programme

The MEDGENET Mentoring programme started its second year. More CEITEC MU PhD students and young researchers have traveled on secondments to their mentors at MEDGENET partner institutions.

The experience of two researchers is presented below. If you are interested in participating in the MEDGENET mentoring programme, contact [Ms. Iva Vrabelova](#).

Kateřina Stařo Kozubík and Ivona Blaháková, Uppsala University, Sweden

The aim of the educational visit was to learn about new diagnostic panel for inherited breast and ovarian cancer and for leukemias based on the HaloPlex target enrichment system using massive parallel sequencing (MPS). The first week was under the instruction of Prof Marie-Louise Bondeson who introduced the diagnostic approach and research projects on inherited diseases. Dr Panagiotis Baliakas was the trainer for the second week, he showed us the entire procedure of biological samples processing, diagnostic algorithm, molecular methodologies, data analysis and results interpretation in inherited oncologic diseases and leukemias. We also visited the Science for Life Laboratory at the Uppsala Genome Center where sequencing based on SMRT principle was introduced to us for BCR/ABL fusion gene detection and inherited disorders.



Vojtěch Bystrý and Karla Plevová EMBL, Heidelberg, Germany

The aim of the secondment was to get help with the analysis of WGS data from patients with Chromotripsis, improve bioinformatics skills in the field of structural analysis, and learn the concepts of maintaining and managing a high quality Genomic Core facility. In the GeneCore Lab at EMBL, we were introduced to new tools and techniques, and discussed various important topics such as: Variant calling IGV batchmode, Annotation with VEP, BAM cross-contamination sequencing checks, VerifyBamID algorithm, Relatedness sequencing testing, Cross-sample snp correlation check, Allele specific phasing, Snp based ancestry determination, RNA fusion gene detection tools, Microarrays chip analysis tools and SV detection in large sample cohorts.



EMBL courses

3 PhD students and young researchers have participated in EMBL courses since the beginning of 2017. The experience of one course participant—

Filip Pardy is featured below. If you are interested in participating in EMBL courses, contact [Ms. Iva Vrubelova](#).

Filip Pardy: RNA Sequencing Library Preparation - How low can you go?

The aim of the study was extend and improve my knowledge about RNA-Seq methods based on instruction from staff at one of Europe's top laboratories. I have attended a practical course that comprised mostly of wet lab work, we also had instructive QA session with NEB application specialists and introduction to bioinformatics. I have been successful in library preparation and I am thus able to spread this knowledge further in environment of CEITEC MU.



UPCOMING EVENTS

Workshop on advanced microscopy and spectroscopy methods in quantitative analysis of biomolecular interactions

WHEN: 23rd-26th October 2017

WHERE: CEITEC MU, the Czech Republic

TOPIC: This workshop will be dedicated to modern microscopic techniques used for the study of biomolecular interactions and functions. The program will consist of one day of lectures given by outstanding and internationally recognized speakers. The lectures are open to broad audience. The lecture day is followed by a four day practical workshop that will be focused on training of students that will use the sophisticated microscopic approaches in their research.

SPEAKERS: will be announced soon

Bioinformatics course – NGS analytical pipeline design and management

WHEN: November 2017

WHERE: CEITEC MU, the Czech Republic

TOPIC: Next generation sequencing (NGS) is becoming a method of choice for an ever growing number of applications in both research and clinics. However subsequent NGS data analysis still creates a backlog in the process, since designing appropriate analytical pipeline tailored to the data is a daunting task. In this course we will present basic logical steps and reasoning behind a creation of analytical pipelines for the most common NGS experiments. The course will also address the problem of organising and managing analytical workflows in order to maintain a sustainable bioinformatics service.

SPEAKERS: trainers from EMBL will be invited

Events updates will be available in [Upcoming Events](#).

Ethics in research projects

WHEN: 13th October 2017

WHERE: CEITEC MU, the Czech Republic

TOPIC: Understanding ethics in biomedicine and the relationship between science and society has become an essential element of all research projects, especially international collaborations. This workshop will focus on ethical issues in biomedicine and research projects, as well as the process of resolution of ethical issues during project preparation and realisation. The workshop will take place within MU Grants Week.

INTERESTING SPEAKERS: To be announced.

Intellectual Property Rights in research projects

WHEN: 13th October 2017

WHERE: CEITEC MU, the Czech Republic

TOPIC: The seminar will be devoted to discussing challenges and sharing best practices in IPR management in Biomedical research, enforcement strategies and advice on avoiding common pitfalls. The seminar will take place within MU Grants Week.

SPEAKERS: To be announced.

The workshop and seminar updates will be available in [Upcoming Events](#).

PROJECT DISSEMINATION

MEDGENET dissemination activities

In 2017 our researchers continued representing MEDGENET project at international events all over Europe, such as the European Society of Human Genetics ESHG conference in Copenhagen, Denmark, the XVII International Workshop on Chronic Lymphocytic Leukemia in New York, USA or The 22nd Annual Meeting of the RNA Society in Prague, the Czech Republic.

The project was also presented at the Business Research Forum at CEITEC MU in April 2017. Additionally, a one-day educational program for elementary schools “A little scientist for a while” was organised aimed at promoting science among school children. This event accommodated 64 participants.

