PRESS RELEASE

The University Hospital in Motol became the recipient of the Norway Grants. These funds will be used for the project of the National Coordination Centre for Rare Diseases at the Institute of Biology and Medical Genetics of the University Hospital in Motol (Reg. No. NF-CZ11-PDP-3-003-2014). The total project budget is CZK 24,601,526, while the amount of grant is CZK 19,681,220.

The partner on the Norwegian side is the **Norwegian Resource Centre for Rare Disorders** (www.frambu.no). FRAMBU is a government's expert agency for rare disorders, which is working on a project in the area of raising awareness of professionals and the general public on the issue of rare diseases in order to improve the detection of genetic risk families. Another Norwegian partner is the **Centre for Medical Genetics and Molecular Medicine at the Haukeland University Hospital in Bergen** (www.helse-bergen.no). This is a leading Norwegian academic medical facility with which the University Hospital in Motol will cooperate in improving clinical genetic methods for next-generation sequencing (NGS), syndromological examination, detection of inborn errors of metabolism, and development of molecular cytogenetic and genetic methods for early prenatal diagnosis, bioinformatics, and digital phenotyping of rare diseases.

The project aims to:

1) Increase awareness and knowledge about rare diseases in both lay and professional public
2) Improve detection and allow for secondary prevention of genetic risk families
3) Prepare the concept of diagnosis and medical and preventive care for rare diseases based on the experience of the Norwegian project partner
4) Carry out model pharmacoeconomic studies in selected rare diseases for objectification of reimbursement for diagnostic or therapeutic procedures

norway grants

The project supports in particular:

1) Development of website (www.vzacnenemoci.cz), training of professionals and the lay public, including use of the experiences of the Norwegian project partner in this area
2) Preparation of expert advice and publications on rare diseases for the professional and general public
3) Implementation of pharmacoeconomic studies in selected rare diseases
4) Improvement of arrayCGH, MLPA, QFPCR, and NGS methods for preimplantation, prenatal and postnatal diagnosis of rare diseases
5) Using of the latest NGS method for secondary and tertiary prevention of rare diseases

The project's target group are people or families with rare diseases

On 1 December 2014, the inaugural conference was held at the University Hospital in Motol, which brought together work teams from Norway - FRAMBU and University of Bergen and the Working Group of the Institute of Biology and Medical Genetics and Czech Association for Rare Diseases (ČAVO; www.vzacna-onemocneni.cz). This joint meeting contributed to experience sharing on the concept of diagnosis and therapeutic-preventive care for rare diseases between the Norwegian and Czech project partners. Also the principles of mutual cooperation during the project solution phase and at the time of project sustainability were established. The project parties also discussed the reciprocal training of individual specialists and patients associations provided at FRAMBU and raising of awareness of rare diseases in collaboration with practical paediatricians in 2015 (www.detskylekar.cz).

In Prague, on February 2, 2015