PRESS RELEASE

Final report of project NF-CZ11-PDP-3-003-2014 National Coordination Center for Rare Diseases at the Motol University Hospital

In July 2014 the Norway Grants project "The National Coordination Center for Rare Diseases at the Motol University Hospital" (Registry Number: NF-CZ11-PDP-3-003-2014; www.nkcvo.cz; NKCVO) was launched and its completion was scheduled for April 30, 2017. The official recipient of funds from the Norway Grants Financial Mechanism was the Motol University Hospital (www.fnmotol.cz; UHM). Overall, UHM was awarded CZK 30 975 417, out of which 80% of the total amount of CZK 24 777 933 was covered by the aforementioned financial mechanism. The co-applicants of the project were Faculty Hospital Brno-Department of Medical Genetics (www.fnbrno.cz/oddeleni-lekarske-genetiky/k1479) and the Faculty of Sciences-Department of Anthropology and Human Genetics (www.natur.cuni.cz/biologie/antropologie). Within this project the cooperation with the Czech Association for Rare Diseases (ČAVO; www.vzacna-onemocneni.cz; CAVO) was also expanded.

The first Norwegian partner to take part in the project was the Norwegian Resource Center for Rare Disorders (www.frambu.no). FRAMBU has been working with NKCVO on raising awareness among rare disease (RD) professionals and the general public in order to improve their early detection and ensured *on site* training of Czech RD patient organizations and/or experts in the area of health and social issues.

The second Norwegian partner was Haukeland University Hospital, Bergen, Center for Medical Genetics and Molecular Medicine (www.helse-bergen.no, HUH Bergen). HUH Bergen is the top Norwegian genetic facility with which NKCVO had collaborated on the improvement of next generation sequencing diagnostics, human phenotype ontology and bioinformatics related to RD.

Within the project, Czech somatometric standards-, 3D models of jaws and upper palate and 3D “facial gestalt” metadata were established in a representative cohort of apparently healthy Czech children between 3-18 years of age. In addition, a proprietary Morpho3metrics program was improved and a pilot exome sequencing in molecular syndromology linked to human phenotype ontology and 3D facial gestalt analysis had been carried. Unique equipment purchased during the project from Norway Grants (comprising e.g. 3dMD facial gestalt 3D scanner, Bravo liquid handling system, special software) will be used within the sustainability period at UHM, and will serve the entire country via NKCVO.

More than ten events for the professional and non-professional public were organized and supported to raise RD awareness. A dedicated monograph “Rare Cancers” was edited and more than 250 RD patients were examined within the project through improved methods of next generation sequencing and molecular cytogenetics, a large DNA biobank from various RD was established, and the system of teaching of pre-graduate students of the 2nd Faculty of Medicine of Charles University was introduced in collaboration with CAVO. In addition, dedicated websites were developed RD (www.nkcvo.cz, www.vzacnenemoci.cz, www.vzacna-onemocneni.cz). A very successful helpmail was introduced and a nationwide population specific genetic variant database was created, all of which provided a basis of excellent sustainability of the project at UHM.

MINISTERSTVO ZDRAVOTNICTVÍ
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Overview of achievements:

1) Lectures for the professional and non-professional public - 55

2) Dedicated articles - 13

3) Expert monographs – 2

4) The chapter of the monograph – 1

5) Number of examined patients with RD using next generation sequencing, arrayCGH, MLPA, Karyomapping - 969

6) Establishment of a DNA biobank – 100 DNA samples

7) Educational events for the professional and lay public - 10

8) Distribution of information leaflets to pediatric and adult general practitioners - 3000

9 Setting up helpemail - 1

10) Establishment of web portals – www.nkcvo.cz, www.vzacnenemoci.cz, www.vzacna-onemocneni.cz

11) Pharmacoeconomic studies - 2

12) Employment of professionals

NKCVO prepared guidelines and endorsed participation of 17 Czech RD experts centers in respective European Reference Networks for rare diseases (ERN; http://ec.europa.eu/health/ern/policy_en). Through the activities of NKCVO the Czech Republic is currently ranked 8th within EU28 countries and first within EU13 (EU New Member States) in terms of the scope and quality of RD ERN participation.

Overall, this project was successfully completed and significantly improved care for RD in the country. We also would like to thank our Norwegian partners for beneficial cooperation and we would like expanded RD-related activities in the future within further Norway Grants funding