

2nd Joint Workshop

Norway Grants project

NF-CZ11-PDP-3-003-2014

Národní koordinační centrum pro vzácná onemocnění ve Fakultní nemocnici v Motole
(National Coordinating Centre for rare Diseases at the Motol University Hospitals)

Title: NEXT GENERATION SEQUENCING & BIOINFORMATICS IN RARE DISEASE RESEARCH
University Hospital Prague Motol (CZ) and Haukeland University Hospital Bergen (NO)

Date: January 22, 2016

Time: 9 00 – 16 30

Venue: Department of Biology and Medical Genetics University Hospital Motol
4th Floor Pediatric Polyclinic tract G (www.fnmotol.cz/ublg)

09 00 - 09 30 *Arrival of participants*

09 30 - 09 45 *Welcome and presentation of workshop participants*

Milan Macek, Gunnar Houge

09 45 -10 00 *Introduction and current outcomes of the PDP3 project*

Milan Macek

SHARING EXPERTISE

Bergen

10 00-10 30 *Genomics of rare diseases: Bergen experience*

Gunnar Houge

Prague

10 30-11 00 *Molecular syndromology and phenotyping*

Jana Paděrová

11 00-11 30 *Cardiomyopathies*

Patricia Norambuena

11 30-12 00 *Facial gestalt analysis by 3D phenotyping*

Veronika Cagáňová

12 00-12 30 *Interesting cases from molecular cytogenetics*

Jana Drábová

12 30-13 00 *Autistic spectrum disorders*

Markéta Havlovicová / Zdeněk Sedláček

Bergen / Prague

13 00-13 30 *Outlooks for collaboration, visit of the UBLG genomic facility and 3D phenotyping*

13 30-14 30 *Lunch University Hospital Cafeteria*

NEXT GENERATION SEQUENCING BIOINFORMATICS

14 30-14 45 *UBLG bioinformatics pipeline: an overview*

Jan Geryk

14 45-15 30 *Whole genome sequencing - detection of causative variants in rare monogenic disorders*

Tomas Stokowy

15 30-16 30 *Practical demonstration activities, discussion on bioinformatics approaches*

16 30 *Workshop adjourns and transfer to the dinner*

Transfer to Hotel Pyramida (www.hotelpyramida.cz).



MINISTERSTVO ZDRAVOTNICTVÍ
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