# 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)***

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***Date:* January 22, 2016**
***Time:* 9 00 – 16 30**
**Venue: Department of Biology and Medical Genetics University Hospital Motol**
***4th Floor Pediatric Policlinic tract G (www.fnmotol.cz/ublg)***

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| **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**

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

**Prague**

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| **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***

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| **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

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| **14 30-14 45** | *UBLG bioinformatics pipeline: an overview* | Jan Geryk |

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

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

**16 30** *Workshop adjourns and transfer to the dinner*

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

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