Norsk forening for medisinsk genetikk
DEN NORSKE LEGEFORENING

NSHG
NORSK SELSKAP FOR HUMANGENETIKK

THE 13th NORWEGIAN NATIONAL GENETICS MEETING

Post-exome diagnostic strategies

Tromsø, November 9th - 10th 2022

DAY 1

Wednesday, November 9th 2022

09.00-10.30 Registration

10.30-10.35 **Welcome** by the local organizing committee and the chair of the Norwegian Society for Medical Genetics (NFMG)

10.35-11.30 **European Reference Networks (ERNs), national and local multidisciplinary teams (MTDs) in rare disorders**
ERNs – how to apply and how to report? Linking ERNs to national MDTs
Multiple teams presenting *(in Norwegian)*:

- National neuromuscular team and ERN-Euro NMD, Kristin Ørstavik & Andreas Rosenberger
- MetabERN, national network and MDT for metabolic disorders, Trine Tangeraas
- Immunodeficiency team and ERN-RITA, Hans Christian Erichsen
- ERN ITHACA, Sofia D. Houge
- Cranio facial team, Elin Tønne
- National treatment unit for vascular malformations (tba)
- MDT for neurofibromatosis/schwannomatosis, Gorlin clinic, Cecilie Rustad
- Skeletal dysplasia clinic, Achondroplasia clinic, ERN-Bond, Cecilie Rustad
- National advisory unit on rare disorders (NKSD) (tba)
- PM center for rare disorders OUS, Olve Moldestad

11.30-12.30 Lunch break

- LIS lunch: All LIS are encouraged to meet during lunch time

12.30-13.10 **Evolving roles of DNA methylation episignatures in the diagnosis of genetic neurodevelopmental disorders and beyond.** *(Lecture in English)*
Bekim Sadikovic,
Professor and Program Head, Molecular Diagnostics Program, Department of Pathology and Laboratory Medicine, London Health Sciences and St Joseph's Healthcare, Ontario, Canada
Chair of the session: Sofia Douzgou Houge

13.10-13.20 Q&A

13.20-14.00 **Solve-RD: diagnosing Rare Disease patients through large-scale data sharing and collaborative analysis.** *(Lecture in English)*
Sergi Beltran,
Head of Bioinformatics Unit and the Data Analysis team at the National Center for Genomic Regulation, Barcelona, Spain
Chair of the session: Sofia Douzgou Houge

14.00-14.10 Q&A

DAY 1 continue

14.10-14.20 **«Elevator pitch» – Posters,** part 1 (uneven numbers) *(Presentations in English)*

Chair of the session: Marijke Van Ghelue

14.20-15.00 Coffee Break with poster viewing

15.00-15.45 **Presentation of submitted abstracts** ("frie foredrag") 3x15 min *(in English)*
Chair of the session: Abstract committee representative

15.45-17.15 **Workshops Day 1**

- Molecular genetics and bioinformatics, Olaug Rødningen *(in Norwegian)*
  - *a.* Recommendations for CNV classification and nomenclature, an update
  - *b.* CNV calling from NGS data, Ella and other bioinformatics tools. Use of MLPA
- Inherited cancer Lovise Mæhle *(in Norwegian)*
- Genetic counselling and videoconferences, Benedicte Tolo *(in Norwegian)*
- Inborn errors of metabolism IEM, Yngve Thomas Bliksrud *(in Norwegian)*

17.15-17.30 **Closing remarks Day 1**

17.30- General Assembly Norwegian Society of Human Genetics (NSHG) *(in Norwegian)*

18.00- General Assembly Norwegian Society of Medical Genetics (NFMG) *(in Norwegian)*

19.00- Dinner

Norsk forening for medisinsk genetikk
DEN NORSKE LEGEFORENING

NSHG
NORSK SELSKAP FOR HUMANGENETIKK

DAY 2

Thursday, November 10th 2022

08.15-08.50 Registration

08.55-09.00 **Welcome** by the chair of the Norwegian Society for Human Genetics (NSHG)

09.00-09.30 **Germline and somatic variants in bone marrow failure disorders.** *(Lecture in English)*
Why and when and what to test, and how to interpret the variants?
Bianca Tesi,
MD PhD Department of Molecular Medicine and Surgery, Center for Molecular Medicine, Karolinska Institutet, and Department of Clinical Genetics, Karolinska University Laboratory, Karolinska University Hospital, Stockholm, Sweden.
Chair of the session: A. Stray-Pedersen

09.30-09.40 Q&A

09.40-09.50 **«Elevator pitch» – Posters,** part 2 (even numbers)
Chair of the session: Marijke Van Ghelue

09.50-10.30 Coffee break with poster viewing

10.30-10.40 **Genetikkportalen**, *(in Norwegian)* by Pablo Cortez, Haukeland University Hospital, Bergen

10.40-11.45 **Prenatal diagnostics, NIPT, PGD and Biotechnology law changes** *(in Norwegian)*

- Status NIPT Norge, fra fostermedisinsk side, Ragnhild Glad UNN
- NIPT til kvinner < 35 år, rapport fra Helsedirektoratet, Ingrid Stavenes Andersen HDir
- Ny labenhet prenatal diagnostikk og infertilitetsutredning, Anne Blomhoff AMG OUS
- Status PGD ved OUS, transport-PGT, behandlingstider, Barbro Stadheim AMG og Prof. Peter Fedorcsak, reproduksjonsmedisinsk avdeling OUS
- Status PGD ved St. Olavs og transport-PGT, behandlingstider, Hanna Schilling, Fertilitetsseksjonen og Julie Paulsen Med gen St Olav.

11.45-12.45 Lunch

12.45-13.30 **Presentation of submitted abstracts** ("frie foredrag") 3x15 min
Chair of the session: Abstract committee representative

13.30-14.45 **Workshops Day 2**

- New technologies for detection of structural variants, Optical mapping and long read sequencing, Olaug Rødningen (*in Norwegian*)
- Cardio genetics – Evaluering av nasjonal anbefaling for genetisk kardiologi (*in Norwegian*)
- Clinical genetics – Incidental findings and children. When and how to report? (*in Norwegian*)

14.45-15.00 **Awards**

15.00-15.10 **Closing remarks**