



Norsk forening for  
medisinsk genetik

DEN NORSKE LEGEFORENING



# THE 13th NORWEGIAN NATIONAL GENETICS MEETING

## Post-exome diagnostic strategies

Tromsø 9-10. Nov 2022

### DAY 1

**Wednesday, November 9th 2022**

09.00-10.15 Registration

10.15-10.20 **Welcome** by the local organizing committee and the chair of the Norwegian Society for Medical Genetics (NFMG)

10.20-11.30 **European Reference Networks ERNs, national and local multidisciplinary teams (MTDs) in rare disorders**

Chair of the session: Asbjørg Stray Pedersen

*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, Inger Lise Mero
- MDT for neurofibromatosis/schwannomatosis, Gorlin clinic, Cecilie Rustad
- Skeletal dysplasia clinic, Achondroplasia clinic, ERN-Bond, Cecilie Rustad
- National advisory unit on rare disorders (NKSD), Stein Are Aksnes
- 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 epigenatures 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



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## DAY 1 cont.

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

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**

- ELLA and your ideas for an ideal variant interpretation tool Morten Eike og Tony Håndstad
- Inherited cancer Lovise Mæhle *(in Norwegian)*
- Genetic counselling and videoconferences, Benedicte Tolo *(in Norwegian)*
- Inborn errors of metabolism IEM, Yngve Thomas Blikrud *(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



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## DAY 2

Thursday, November 10<sup>th</sup> 2022

08.15-08.40 Registration

08.45-08.50 **Welcome** by the chair of the Norwegian Society for Human Genetics (NSHG)

08.50-09.20 **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

09.20-09.30 Q&A

09.30-09.40 **«Elevator pitch» – Posters, part 2** (even numbers)

Chair of the session: Marijke Van Ghelue

09.40-10.20 Coffee break with poster viewing

10.20-10.30 **Genetikportalen**, (*in Norwegian*) by Pablo Cortez, Haukeland University Hospital, Bergen

10.30-11.30 Prenatal diagnostics, NIPT, PGD and Biotechnology law changes (*in Norwegian*)

- Status NIPT Norge, fra foster medisinsk side, Ragnhild Glad UNN
- NIPT til kvinner < 35 år, rapport fra Helsedirektoratet, Ingrid Stavenes Andersen HDir
- Ny lab enhet 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.

11.30-12.40 Lunch

12.40-13.25 **Presentation of submitted abstracts** ("frie foredrag") 3x15 min

Chair of the session: Abstract committee representative

13.30-14.45 **Workshops Day 2**

- Analyse og klassifisering av strukturelle varianter Olaug Rødningen (*in Norwegian*)
- Cardio genetics Knut Erik Berge (*in Norwegian*)
- Clinical genetics – Incidental findings and children. When and how to report? Charlotte von der Lippe (*in Norwegian*)

14.45-15.00 **Awards**

15.00-15.10 **Closing remarks**