Nordic Alliance for Clinical Genomics

16th NACG workshop

Emerging technologies of clinical genomics

Marienlyst Strandhotel, Helsingør, Denmark

25-26th September 2025

Preliminary Agenda

Preliminary programme at a glance:

Day 1

Time	Session
10:00 - 10:30	Registration
10:30 - 11:00	**Welcome address**
11:00 - 12:00	**Keynote by Edwin Cuppen**
12:00 - 12:45	**Rapid fire talks**
12:45 - 13:45	Lunch
13:45 - 14:15	**Rapid fire talks** (continued)
14:15 - 15:15	**Clinical application of emerging technologies for DNA sequencing**
15:15 - 15:45	Coffee break
15:45 - 17:00	**Parallel sessions:** **Bioinformatics: Reference genomes** **Clinical: Variant classification**
17:00 - 19:30	Time on your own
19:30 - 20:00	Pre-dinner drink and social activity
20:00	Dinner

Day 2

Time	Session
09:00 - 10:30	**Parallel sessions:** **Bioinformatics: Thresholds and quality measures** **Clinical: Variant interpretation and visualisation**
10:30 - 11:00	Coffee break
11:00 - 11:30	**Recap of parallel sessions**
11:30 - 12:30	**Clinical application of emerging technologies**
12:30 - 13:30	Lunch
13:30 - 14:15	**AI based interpretation tools**
14:15 - 14:45	Coffee break
14:45 - 15.45	**National / Regional scale initiatives**
15.45 - 16:00	**Summary & farewell, announcement of next NACG**

25th September 2025

Time	Session	Lead
10:00 - 10:30	**Registration** Coffee and snacks	GM organisers
10:30 - 11:00	**Welcome address**	GM organisers + NACG steering commitee
11:00 - 12:00	**Keynote speaker** Edwin Cuppen *Ultima and Roche sequencing technologies for clinical cancer genomics.*	Scientific committee
12:00 - 12:45	**Rapid fire talks** Brief yet impactful presentations (maximum 5 minutes) on topics relevant to NACG activities. Whether it's about cutting-edge tools, innovative workflows, fascinating clinical cases, everyday lab dilemmas encountered or anything else relevant, this is your chance to share and shine!	Anders Jemt Kaisa Kettunen
12:45 - 13:45	**Lunch**	GM organisers
13:45 - 14:15	**Rapid fire talks** (continued)	Anders Jemt Kaisa Kettunen
14:15 - 15:15	**Clinical application of emerging technologies for DNA sequencing** Knowledge sharing of DNA sequencing when it comes to the emerging technologies such as longread, Ultima, Optical genome mapping. We also welcome hard-to-solve cases that have been solved with some of these technologies.	Ane Yde Schmidt Edda Elvarsdóttir
15:15 - 15:45	**Coffee break and visit to sponsor booths**	GM organisers
15:45 - 17:00	**Parallel session - Bioinformatics** **Reference genomes** With most clinical labs now operating on GRCh38 and growing interest in the T2T reference, the choice of reference genome continues to have a significant impact on variant discovery and interpretation. This session will explore the clinical implications of reference selection, including compatibility with existing databases and pipelines. We will also touch on the emerging human pangenome and graph-based genome representations, which aim to more accurately reflect the complexity and diversity of human genetic variation.	Anders Jemt Frederik Otzen Bagger
	Parallel session - Clinical (lab + interpretation) **Variant classification** A few variant classifications are still unsolved from the Norwegian data sharing project (VARDE). The WS will both be	Gry Asker Beate Skinningsrud Majbritt Busk

	a benchmarking with the other Nordic departments, and discussions about the use of diverging ACMG-criteria. We will also highlight the new ACMG guidelines.	Madsen
17:00-19:30	**Time on your own** - spa, sauna or free time	
19:30 - 20:00	**Pre-dinner drink and social activity**	GM organisers
20:00	**Dinner**	GM organisers

26th September 2025

Time	Session	Lead
Before 9:00	**Check out, breakfast and access to spa**	
09:00 - 10:30	**Parallel session - Bioinformatics** **Thresholds and quality measures** This session will focus on sharing insights and experiences related to the application of sensitivity and specificity thresholds in the validation and implementation of new methods. Presenters will explore strategies for selecting the optimal balance between sensitivity and specificity, as well as approaches for defining robust quality control (QC) parameters.	Tony Håndstad Thomas Als
	Parallel session - Clinical (lab + interpretation) **Variant interpretation and visualisation** In this session we will dive into the various ways of interpretation and visualisation of genetic variants, and explore the functionalities and limitations of different software and tools, and their usage for the emerging sequencing technologies. We encourage participants to share their experiences.	Majbritt Busk Madsen Beate Skinningsrud Gry Asker
10:30 - 11:00	**Coffee break and visit to sponsor booths**	GM organisers
11:00 - 11:30	**Joint session** Recap of parallel sessions	Scientific committee
11:30 - 12:30	**Clinical application of emerging technologies** In many cases, routine diagnostics with short read WES or WGS come out with normal findings. This session is dedicated to presentation of clinical cases and subtypes of variants that have been solved with emerging technologies such as long read seq (PacBio, ONT), RNA seq, DNA methylation (EpiSign), optical genome mapping (Bionano) etc.	Vidar Martin Steen Anna Lyander
12:30 - 13:30	**Lunch**	GM organisers
13:30 - 14:15	**AI based interpretation tools** Several AI-based interpretation tools are now available, mainly from commercial suppliers. We encourage labs to present their user experiences with such tools.	Vidar Martin Steen Edda Elvarsdóttir
14:15 - 14:45	**Coffee break and visit to sponsor booths**	GM organisers
14:45 - 15.45	**National / Regional scale initiatives** Presentation of large-scale (national/regional/multinational) projects within Clinical Genomics (Approx. 10 min talks)	Ane Yde Schmidt
15.45 - 16:00	**Summary & farewell, announcement of next NACG**	NACG steering commitee

More information about Nordic Alliance for Clinical Genomics (NACG) can be found at https://nordicclinicalgenomics.org/

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Organizing committee:

Ane Yde Schmidt, Lejla Majdanac, Majbritt Busk Madsen

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