Nordic Alliance for Clinical Genomics

16th NACG-workshop

Emerging technologies of clinical genomics

Marienlyst Strandhotel, Helsingør, Denmark

25-26th September, 2025

Welcome to the 16th NACG workshop

Programme committee:

Anders Jemt, Ane Yde Schmidt, Beate Skinningsrud, Kaisa Kettunen, Majbritt Busk Madsen, Thomas Damm Als, Vidar Martin Steen

Organizing committee:

Ane Yde Schmidt, Lejla Majdanac, Majbritt Busk Madsen

Nordic Alliance for Clinical Genomics

Agenda

Programme at a glance:

Day 1

Time	Session
10:00 - 10:30	Registration
10:30 - 11:00	**Welcome**
11:00 - 12:00	**Opening keynote lecture 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	Free time for for individual activities
19:30 - 20:00	Pre-dinner drinks 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	**Closing remarks**

25th September 2025

Time	Session	Lead
10:00 - 10:30	**Registration** Coffee and snacks	
10:30 - 11:00	**Welcome**	Maria Rossing + NACG steering committee
11:00 - 12:00	**Opening keynote lecture** **Edwin Cuppen** *Ultima and Roche sequencing technologies for clinical cancer genomics.*	Maria Rossing
12:00 - 12:45	**Rapid-fire talks** **Henrikki Almusa**: *Lab automation and effects on (CNV) analysis, part 2* **Tatjana Pandzic**: *Rapid detection of leukemic abnormalities using nanopore sequencing* **Anna Lyander**: *Benchmarking four new library preparation kits against KAPA HyperPlus/Prep* **Hanne Sørmo Sorte**: *Experience with OGM at AMG, OUS* ***Morten Eike**: Primer Check: Minimize allelic dropouts with gnomAD* **Thomas Damm Als**: *During-Surgery CNS Tumor Classification: A Methylation-Based Approach Using Nanopore Sequencing* **Klaus Tangsgaard (NEB)**: *Methods for sequencing cell-free DNA and cell-free RNA from human plasma* **Christophe Meynier (Seqone)**: *Transforming Clinical Genomics with AI-Powered Workflows*	Anders Jemt Kaisa Kettunen
12:45 - 13:45	**Lunch**	
13:45 - 14:15	**Rapid-fire talks** (continued) **Alina Orozco**: *PSMUDS : a patient specific mutation analysis pipeline* **Jakob Jersild Nielsen**: *Detection of RNA Outliers: A Clinical Implementation* **Marianne Overgaard Hesselager**: *Creating National Consensus on Variant Interpretation* **Morten Dunø**: *Dominant inheritance – when one is not enough* **Arianda Abazi (Centogene)**: *Centogene Biodata Bank / Centogene CentoDX Variant Interpretation Tool* **Ahmet Can Türkoğlu (Genomize)**: *How to Combine Genomics, Deep Phenotyping, and AI to Diagnose Patients with Rare Diseases*	Anders Jemt Kaisa Kettunen
14:15 - 15:15	**Clinical application of emerging technologies for DNA sequencing** Knowledge-sharing of DNA sequencing techniques, hard to solve cases etc. **Aino Palva:** *Comparing low pass WGS and NGS target enrichment as a method to replace genotyping*	Ane Yde Schmidt Edda Elvarsdóttir

	Gregor Gilfillan: *Detecting a cryptic splice variant in the Congenital Adrenal Hyperplasia seg-dup gene CYP11B1 by adaptive sampling* **Håkan Thonberg:** *Clinical long-read sequencing and expansion-detection* **Carolina Diettrich, Moa Hägglund, Anders Jemt and Anna Lyander:** *Evaluation of the Element Biosciences Trinity Fast Hybridization Workflow Using the GMS Myeloid Panel*	
15:15 - 15:45	**Coffee break and sponsor exhibits**	
15:45 - 17:00	**<u>Parallel session - Bioinformatics</u>** **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. **Ksenia Lavrichenko & Eirini Liampa**: *Mapping the shift of diagnostics to GRCh38 genome build* **Alban Obel Slabowska:** *Graph pangenome reference construction and read mapping* **Jesper Eisfedlt:** *De novo assembly analyses in the UDN long read trio cohort*	Anders Jemt Frederik Otzen Bagger
	<u>Parallel session - Clinical</u> **Variant classification** A few variant classifications are still unsolved from the Norwegian data sharing project (VARDE). The WS will both be a benchmarking with the other Nordic departments, and discussions about the use of diverging ACMG-criteria. The draft of new ACMG criteria will also be presented. **Caroline Nangota:** *Overview of DRAFT ACMG/AMP v4 Sequence Variant Guidelines*	Beate Skinningsrud Gry Asker
17:00-19:30	**Free time for personal activities: spa, sauna or other recreation**	
19:30 - 20:00	**Pre-dinner drinks**	
20:00	**Dinner**	

26th September 2025

Time	Session	Lead
Before 9:00	**Checkout, breakfast and access to spa**	
09:00 - 10:30	**Parallel session - Bioinformatics** **Thresholds and quality measures** This session will focus on validation of new methods, finding optimal thresholds, as well as defining robust and standardised quality control (QC) metrics. **Timo Järvinen**: *Validation of DRAGEN for Clinical Use* **Ying Sheng:** *Impact of Sequencing Coverage on the Accuracy of Variant Calling* **Karl Svärd:** *Striking the Balance: Setting Effective QC Thresholds in a Production Environment* **Tony Håndstad:** A proposed standard for WGS QC by GA4GH	Tony Håndstad Thomas Damm Als
	Parallel session - Clinical **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.	Majbritt Busk Madsen Benjamin Schantz-Conlon
10:30 - 11:00	**Coffee break and sponsor exhibits**	
11:00 - 11:30	**Recap of parallel sessions**	
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. **Anna Lindstrand** *Completing the picture: Long-read sequencing, reference genomes and multi-omics strategies in rare disease genomics* **Andreas Ørslev Rasmussen** *Routine RNA-seq as part of germline analysis: Implementation, obstacles and clinical outcome* **Peter Pruisscher** *Use cases for optical genome mapping*	Vidar Martin Steen Anna Lyander
12:30 - 13:30	**Lunch**	
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

	Eirik Bratland *Experience from using Franklin by Genoox for the interpretation of germline variants* **Eirikur Briem** *Implementing SOPHIA DDM: AI-driven enhanced exome interpretation across diverse indications*	
14:15 - 14:45	**Coffee break and sponsor ex**	
14:45 - 15.45	**<u>National / Regional scale initiatives</u>** **Ronja Hotakainen** *Emerging Carrier Screening Approaches in Finland* **Anna Lindstrand** *GMS-LRS1000: A National Long-Read Genome Project.* **Inge Søkilde Pedersen** *Danish initiatives towards national consensus in clinical genomics* **Eiríkur Briem** *Sequencing Panel for the Simultaneous Detection of Common Variants in the Icelandic Population*	Ane Yde Schmidt
15.45 - 16:00	**Closing remarks**	NACG steering commitee

Attendees

Arianda Abazi

Henrikki Almusa

Thomas Damm Als

Anna-Kaisa Anttonen

Gry Asker

Jannie Assenholt

Frederik Otzen Bagger

Mads Bak

Tuva Barøy

Christian Baudet

Eva Berglund

Niclas Björn

Anneli Nordén Björnlert

Eliott Bosshard

Eirik Bratland

Gwenna Breton

Eirikur Briem

Dimitrios Chouliaras

Lise Lotte Christensen

Mette Christiansen

Edwin Cuppen

Nina de Taeje

Morten Dunø

Edda Maria Elvarsdóttir

Morten C. Eike

Jesper Eisfeldt

Jakob Ek

Ida Coordt Elle

Rada Ellegaard

Caroline Nangota Felde

Ida-Renée Forsberg Langemyhr

Ingvild Synnøve Matre Gabrielsen

Gregor Gilfillan

Magnús Halldór Gislason

Victor Enrique Goitea

Andrei Guliaev

Wenche Scheel Hamang

Johan Hallin

Qin Hao

Maria Harbo

Martijn Heeneman

Markus Heidenblad

Viktor Henmyr

Marianne Overgaard Hesselager

Ronja Hotakainen

Moa Hägglund

Tony Håndstad

Tina Catela Ivkovic

Anders Jemt

Trine Maxel Juul

Timo Järvinen

Krishnakumar Kandaswamy

Meri Kaustio

Andreas Kegel

Kaisa Kettunen

Julie Knudsen

Alexander Koc

Sanna Koskela

Klara Alicja Kotkowska

Britta Schlott Kristiansen

Pär Larsson

Ksenia Lavrichenko

Eirini Liampa

Ida Lindegaard

Anna Lindstrand

Anna Lyander

Sara Löfgren

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Lejla Majdanac

Eline Mejlænder-Andersen

Christophe Meynier

Leonardo A. Meza-Zepeda

Lotte Moens

Jakob Jersild Nielsen

Karen Nielsen

Elina Niemelä

Hanne Gro Olsen

Alina Orozco

Aino Palva

Annika Hjerdin Panagopoulos

Tatjana Pandzic

Inge Søkilde Pedersen

Sini Penttilä

Peter Pruisscher

Teija Paakkola

Kari Jerve Ramsøy

Andreas Ørslev Rasmussen

Emilia Regazzoni

Annick Renevey

Maria Revenikioti

Martin Rippin

Maria Rossing

Olaug Rødningen

Janna Saarela

Benjamin Schantz-Conlon

Ane Yde Schmidt

Yngve Sejersted

Ying Sheng

Barbara Siemensen

Ashish Kumar Singh

Beate Skinningsrud

Hanne Sørmo Sorte

Aashish Srivastava

Vidar Martin Steen

Thomas Stautland

Yvan Strahm

Maria Strandh

Karl Svärd

Erik Söderback

Sofia Gruvberger Saal

Klaus Tangsgaard

Esmee ten Berk de Boer

Mari Tervaniemi

Håkan Thonberg

Kasper Thorsen

Mari Tinholt

Dag Undlien

Søren Vang

Ida Maria Westin

Valtteri Wirta

Christina Westmose Yde

Anna Zetterlund

Hanna Klang Årstrand

More information about

Nordic Alliance for Clinical Genomics (NACG) at:
https://nordicclinicalgenomics.org/

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