Nordic Alliance for Clinical Genomics

WORKSHOP AGENDA

Theme: Overcoming short-read insufficiencies using long-read sequencing

NACG 12th Workshop, 28-29 April 2022 in Reykjavik, Iceland

Nordic Alliance for Clinical Genomics

Welcome!

The upcoming NACG 12th workshop will take a different approach from previous workshops, and instead focus on a single theme relevant to clinical genomics, namely **'Overcoming short-read insufficiencies using long-read sequencing'.**

Short-read sequencing (NGS) has led to tremendous progress in medical genomics and is providing a genetic diagnosis to patients who previously went undiagnosed. Despite these improvements, there is still genetic variation that is hard to capture with this method. Hence, some patients are still not diagnosed and genomic laboratories still need to use laborious manual methods such as karyotyping to detect certain causes of disease. In this workshop, we will focus on methods such as long-read sequencing and optical mapping to address to which extent these novel technologies can address current shortcomings in clinical genomics and what it would take to bring the technologies into mainstream healthcare. As always, the workshop will be highly interactive - aiming to foster Nordic collaboration and knowledge sharing.

For site-wise briefing sessions, so far, 8 labs, including SciLifeLab/Karolinska University Hospital, MOMA in Aarhus, Rigshospitalet in Copenhagen and Oslo University Hospital, and UMCG lab in the Netherlands have already signed up to share their experiences in **long-read sequencing** with the NACG community.

Below is the agenda and details of the workshop, as well as practical information for participants.

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Day 1: 28th April 2022 (Room F and G)

8:15 Coffee (Room E)

8:30 Opening and Welcome
Dag E. Undlien, OUS Department of Medical Genetics, NO, NACG chair
Eiríkur Briem, Department of Genetics and Molecular Medicine, Landspitali - The National University Hospital of Iceland

08:40 Keynote lecture by Patrick Sulem, MD, Head of Clinical Sequencing, deCODE: "From Research to Clinics - 25 years of deCODE genetics" (Chair: Eiríkur Briem)

09:30 National updates related to clinical genomics from Nordic representatives (Chair: Dag Undlien) - 5 min presentation + 4 min Q&A

Representatives from: Norway (Dan Undlien), Finland (Janna Saarela), Denmark (Kasper Thorsen & Ole Lund), Sweden (Valtteri Wirta), and Iceland (Eirikur Briem)

10:15 Coffee (Room E)

10:30 Technologies presentation (Chair: Valtteri Wirta) - 15 min presentation + 5 min Q&A

Key technology providers in the long-read sequencing and optical mapping space including **Oxford Nanopore Technologies, BioNano Genomics and Pacific Biosciences** will provide introductions to their clinical applications, as well as recent technology updates.

11:30 Site-wise briefing on long-read/optical mapping implementation Part 1/2 (Chair: Janna Saarela)

- Ebbe Norskov Bak and Ester Ellegaard Sørensen, MOMA (Department of Molecular Medicine) Aarhus University Hospital, Denmark (15 min)
- Martin Larsen, Department of Clinical Genetics, Odense University Hospital, Denmark (15 min)
- Tobias Overlund Stannius and Lusine Nazaryan-Petersen, Center for Genomic Medicine, Rigshospitalet, Denmark (15 min)
- Arvind Sundaram, Department of Medical Genetics, Oslo University Hospital, Norway (15 min)

12:30 Lunch

13:30 Site-wise briefing on long-read/optical mapping implementation Part 2/2 (Chair Janna Saarela)

- Lars Paulin, Institute of Biotechnology, University of Helsinki, Finland (10 min)
- Jesper Eisfeldt & Anna Lindstrand, Karolinska Institute, Sweden (15 min)
- Ulf Birkedal, Department of Clinical Genetics, Rigshospitalet, Denmark (15 min)
- Johanna Lehtonen, Institute for Molecular Medicine Finland (15 min)
- Malin Melin and Joakim Klar, SciLifeLab, Uppsala University/Uppsala University Hospital, Sweden (15 min)
- Dorieke Dijkstra, University Medical Center Groningen, the Netherlands (15 min)

15:00 30-minute booth and networking session (Room E)
15:30 NACG ideation workshop Applications and challenges: Which cases are not solved by short-read sequencing? What are the potential solutions, and how can we get there together in the Nordics? Facilitators: **Valtteri Wirta**, SciLifeLab, Sweden and **Sharmini Alagaratnam**, DNV, Norway
17:30 Conclusion of day 1, **practical information** (Chair: Eiríkur Briem)
19:00 Workshop dinner

Day 2: 29th April 2022 (Room F or G)

8.15 Coffee (Room E)	
08.30 Parallel Session 1 (Room F) **Wet lab - long read challenges and solutions** led by Ebbe Norskov Bak (MOMA)	**08.30 Parallel Session 2 (Room G)** **Bioinformatics – long read challenges and solutions** led by Anders Jemt (SciLife) & Ksenia Lavrichenko (OUS)
10.30 Coffee (Room E)	
10:45 Report back to plenary (Chair: Dag Undlien) **(Room F and G)**	
11:30 Conclusion and depart	

Practical information

Venue and accommodation

The workshop will take place at the Hilton Reykjavík Nordica. Direct booking for hotel rooms at the workshop rate is closed, but please contact miceres@icehotels.is with the reservation number for the group 338393 in case there are still some rooms available at the workshop rate.

Travel

The nearest international airport is Keflavik airport. Please book your flights as soon as possible to ensure you can attend. As the workshop is due to start at 8.30 am on the 28th, it is recommended that you arrive one day prior to the workshop, on the 27th of April.

For airport transfer (hotel to airport), you can book a taxi at:: +354 5885522 (www.hreyfill.is) or +354 5610000 (www.bsr.is).

Requirements for entry to Iceland

Information about entry requirements for Iceland can be found here.

Travel to the NACG meeting should be undertaken in accordance with Covid travel recommendations. NACG and NACG meeting organisers are unable to accept any liability for inability to attend due to Covid restrictions (or other reasons for inability to attend). All activities will be carried out in accordance with local regulations.

Workshop dinner

The workshop dinner will be held on the evening of Thursday 28th April at Vox Hilton Reykavík Nordica.

Resources

Workshop reports from previous events, NACG papers and governing documents are available at https://nordicclinicalgenomics.org/resources

Contact us, sponsorship

If you have questions about the workshop, please contact us, also if you are interested in sponsorship opportunities.

About NACG

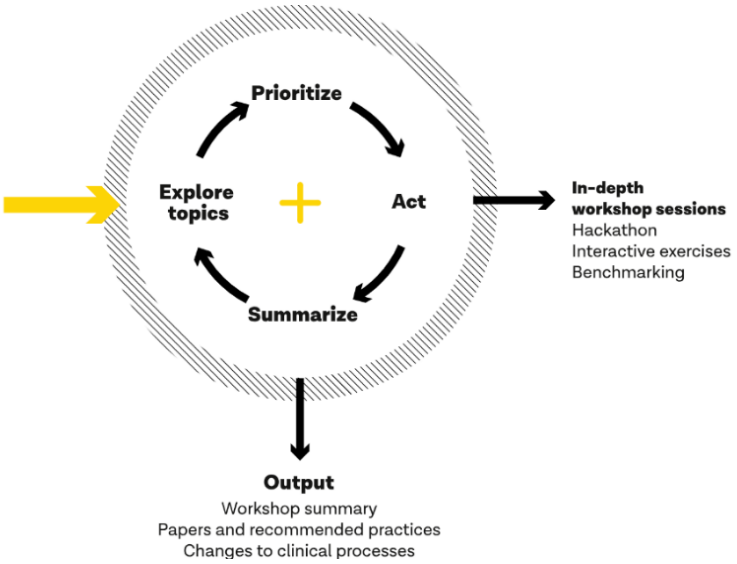
The Nordic Alliance for Clinical Genomics (NACG) is an independent, non-governmental, not-for-profit Nordic association. NACG partners collaborate to identify and address emerging challenges to the implementation of clinical genomics and precision medicine.

Mission

NACG partners work together and learn from each other to lift performance standards. We aim at responsible sharing of trustworthy data for improved diagnosis and treatment, and as a resource for research.

Goals and activities

- Facilitate the responsible sharing of genomic data, bioinformatics tools, sequencing methods and best practices for interpretation of genomic data.
- Enhance quality of genomic data and processes and explore methodologies to provide assurance.
- Understand legal barriers to the implementation of personalized medicine and to engage with key stakeholders that influence these barriers
- Develop demonstration projects that challenge perceived legal barriers that limit responsible and ethical sharing of genomic and health data.
- Build bridges between research and clinical communities, technologies and practices to foster innovation



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