Nordic Alliance
for Clinical Genomics

Welcome to

the 15th NACG workshop

Holmen fjordhotell, 26-27 September 2024

Agenda

Main theme:

Long read sequencing in clinical genomics

26th September, Day 1

Time	Item	Responsible
10:00 - 10:30	Registration and coffee	Oslo University Hospital
10:30 - 11:00	Welcome address	Dag Undlien
11:00 - 12:00	**Invited keynote speaker**: Prof. dr. Christian Gilissen, Radboud university medical center, The Netherlands. Experiences from long read sequencing	
12:00 - 13:00	**National updates** relating to development in national programs in genomic medicine, precision diagnostics and precision medicine	Anna-Kaisa Anttonen Søren Vang Dag Undlien Eirikur Briem Valtteri Wirta
13:00 - 14:00	Lunch	
14:00 - 15:30	**Parallel session 1** – long read sequencing. Bioinformatics Introductions: - "Long read phasing and de novo assembly", Jesper Eisfeldt - "Structural variant analyses in long read sequencing", Anuradha Ravi - "Repeat expansion detection with targeted long read sequencing", Pádraic Corcora *Methods, tools and pipelines*	Ksenia Lavrichenko, Jesper Eisfeldt
	Parallel session 2 – long read sequencing. Wet lab; Experiences and challenges Introductions: - "Optimization of Nanopore Adaptive Sampling for rapid, cost-effective targeted native sequencing", Ebbe Norskov Bak - "Targeted long-read capture is both scalable and economical for population and clinical genomics", Tina Han - "Visual browsing vs long-read SV-calling algorithms (read the small print)", Gregor Gilfillan *DNA extraction, choice of method, pitfalls in sample preparation, robotics, scalability in wetlab*	Ida Höijer, Anna Lyander, Aino Palva

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15:30 - 16:00	Coffee break and visit at sponsor booths	
16:00 - 16.15	**Summary of today's workshops**	Workshop coordinators
16:15 - 17:30	**Rapid-fire session on knowledge sharing** *Brief presentations (6+1 min)* - "Comparison of cancer gene panels for somatic mutation detection in solid tumors",Outi Monni - "Reducing turnaround times: optimising lab preparations and bioinformatics workflows", Beatriz Sá Vinhas and Marcela Gallardo - "Prenatal exome sequencing at Turku Genomics Laboratory", Minna Toivonen - "Genomic Medicine Sweden: Three national long-read sequencing pilot projects", Malin Melin - "Automations arrival to lab and it's effect to CNV analysis", Henrikki Almusa - "What it took to get an IVDR class C device approved for genetic diagnostics", Ben Liesfeld - "Experiences from implementing two long read platforms", Linnéa La Fleur, Felix Lenner and Susanne Månér - "Ring chromosome in exome sequencing", Salla Rusanen - "Features of gnomAD v4", Ronja Hotakainen	Dorte Launholt Lildballe
17:30-19:30	Time on your own – spa, sauna or free time	
19:30 – 20:00	Pre-dinner drink and social activity	Oslo University Hospital
20:00	Dinner	

27th September, Day 2

Time	Item	Responsible
09:00 – 10:00	**Bridging the gap – experiences with AI-solutions in clinical genomics** *In the 14th NACG workshop AI-solutions was the major theme.* *Brief presentations (8+2 min)* - "Introduction to AI in diagnostics", Lena Hausdorf - "Limitations of AI in genomics", Ben Liesfeld - "Benchmarking of AI-based variant prioritization tools: status update", Oleg Agafonov and Ksenia Lavrichenko - "How can AI help identify novel disease-causing mutations in diagnostics and research?", Rocío Acuña-Hidalgo - "An AI driven platform to support the diagnostic process for germline applications", Christophe Meynier - "EU AI Act and its consequences for users of AI-based IVD medical devices", Oleg Agafonov	Beate Skinningsrud
10:00 – 10:20	Coffee break and visit at sponsor booths	

10:20 – 11:30	**Long read seq; Clinical applications - sharing experiences** *Brief presentations (12+2 min)* - "Implementation of Long-Read Genomes for SV and Imprinting Disorders", Martin Larsen - "Identifying shared DNA methylation abnormalities in mendelian disorders of the epigenetic machinery", Katrin Möller - "Long read sequencing in hereditary cancer", Mev Dominguez Valentin - "From Symptoms to Diagnosis: Detection of Repeat Expansions with Nanopore adaptive sampling", Søren Færgeman, Simon O. Drue and Ebbe N. Bak - "Implementing PacBio sequencing in a clinical setting", Jakob Holm Dalsgaard Thomsen and Inge Søkilde Pedersen	Majbritt Busk Madsen, Ksenia Lavrichenko, Gry Asker
11:30 – 12:30	Lunch	
12.30 – 13:30	**Long read seq; Clinical applications - sharing experiences** *Brief presentations (12+2 min)* - "Rapid diagnosis of leukemic aberrations using nanopore sequencing", Ida Höijer - "Nanopore Sequencing in Functional Genetics – Experiences and insights two years later", Ulf Birkeland - "Testing different long-read platforms in a clinical setting", Majbritt Busk Madsen - "The clinical utility of whole genome long read sequencing in neonatal hypotonia diagnostics", Hans Matsson	Majbritt Busk Madsen, Ksenia Lavrichenko, Gry Asker
13:30 – 13:55	**Workshop** **Long read seq; Clinical applications - possibilities, limitations and scalability**	Majbritt Busk Madsen, Ksenia Lavrichenko, Gry Asker
13.55 – 14.15	Coffee break and visit at sponsor booths	
14:15 – 14.45	**Data sharing – Experiences from national data sharing of interpreted variants in Norway**	Dag Undlien
14:45 – 15:00	**Summary & farewell, announcement of next WS**	Dag Undlien

Participants

Last name	First name	Organisation/department	Country
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Thank you for attending the workshop!

More information about

Nordic Alliance for Clinical Genomics (NACG)

can be found at https://nordicclinicalgenomics.org/

A warm thank you to the sponsors:

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