

WORKSHOP

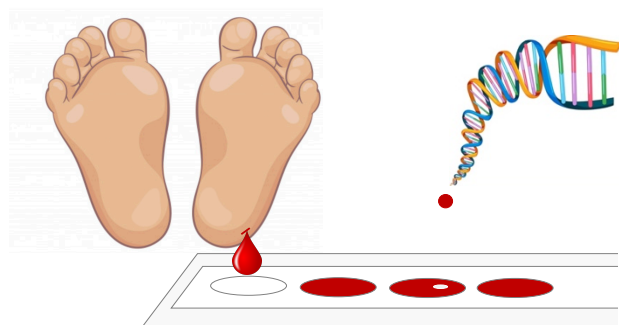
Next Generation Sequencing in Newborn Screening

Gene testing and use of NGS technologies in the NBS laboratory

Auditorium: Forskningsveien 2B, Oslo, Norway

DAY 1 - Wednesday June 21, 2023:

Time	Programme Item
11.00 – 12.00	Registration/Tour in the lab
12.00 –13:00	Lunch
13.00 –13:10	Welcome, introduction of participants and practical info
13.10 – 13.20	DNA-analyses in Newborn screening
13.20 – 13.40	Cystic fibrosis and NGS, Emma Lundman
13.40 – 14.00	NGS in SCID-screening, Asbjørg Stray-Pedersen
14.00 – 14.15	Still a role for Sanger sequencing in NBS, Silje Hogner
14.15 – 14.30	Coffee/Tea
14.30 – 15.00	Slovenian experience with DNA analyses in NBS, Jernej Kovač
15.00 – 15.45	When can it go wrong? What have we learned so far? Interpretation of screening results with cases.
15.45 – 16.00	Q&A, wrap up
16.15	Transport to the hotel
19.00	Get-together Dinner, Scandic Holmenkollen Park



WORKSHOP



Network
Hereditary Metabolic
Disorders (MetabERN)

Member
Oslo University
Hospital – Norway

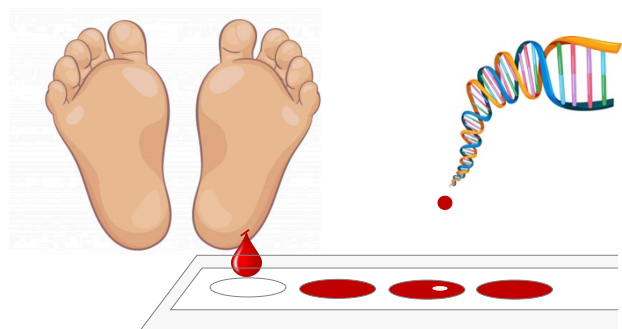
Next Generation Sequencing in Newborn Screening

Gene testing and use of NGS technologies in the NBS laboratory

Auditorium: Forskningsveien 2B, Oslo, Norway

DAY 2 - Thursday June 22, 2023:

Time	Programme Item
09.00 – 09.40	DNA analyses in NBS in Denmark and presentation of new projects, Alberte Aspaas Lundquist and Jonas Bybjerg-Grauholm
09.40 – 10.00	Biochemical screening by LC-MS/MS including 2.tier methods, Ingjerd Sæves
10.00 – 10.10	Post-analytical bioinformatics tools and 2.tier DNA analyses in IEMs, Alex Rowe/Trine Tangeraaas
10.10 – 10.30	Coffee/Tea
10.30 – 10.45	qPCR combined with ddPCR in SMA screening, Janne Strand
10.45 – 11.00	Whole genome sequencing (WGS) performance on DBS. Bioinformatic tools and filtering. NBS variant database. Alex Rowe/Janne Strand
11.00 – 11.10	Long reads with Nanopore using NBS filter cards, William Tourniaire
11.10 – 11.20	RNA seq using NBS filter cards, Mari Ytre-Arne
11.20 – 11.40	Future perspectives: NGS/WGS as 1.tier in NBS, Lennart Hammarström
11.40 – 12.40	Lunch
12.40 – 13.00	Ethical considerations, carrier testing, Asbjørg Stray-Pedersen
13.00 – 14.00	To report, or not report, that's the question. Interpretation of screening results with cases.
14.00 – 14.15	Q&A, wrap up
14.15 – 15.00	Tour in the lab



The workshop venue:

Forskingsveien 2B, Oslo

<https://goo.gl/maps/p5DZDgKyjihUqzww9>,

Room: Seminarrom CU1065

How to get there:

From the airport (Oslo airport Gardermoen) you may travel by bus or train/metro

Bus: From Oslo airport Gardermoen direction Bekkestua (Bus Plattform 11) go off at Rikshospitalet, then 2 min walk over the bridge to Forskningsveien.

Train: airport express train (Flytoget) to Oslo Central station, and then travel with the metro (T-bane) to a station near Forskningsveien: either use line 5 direction Sognsvann and go off at Forskningsparken station, or use line 1 direction Frognerstøya and go off at Gaustad station.

Metro info: <https://ruter.no/globalassets/rutetabeller/t-bane/t-banen-linjekart-03042016.pdf> and <https://ruter.no/en/>

You may store the luggage safely at the workshop venue, we will travel to the hotel together after the workshop.

The hotel:

Scandic Holmenkollen Park

Kongeveien 26, 0787 Oslo

<https://goo.gl/maps/F4kGS6NjezXMdLjMA>



WORKSHOP - REGISTRATION

Next Generation Sequencing in Newborn Screening

Gene testing and use of NGS technologies in the NBS laboratory

Oslo, 21.-22.June 2023:

Course fee: EUR 200 includes:

- Workshop “NGS in NBS”
- Hotel 1 night: 21.June-22.June Scandic Holmenkollen Park
- **Get-together Dinner 21.June at 19:00**

(Payment for the workshop only is: 50 EUR)

Registration: Pay the course fee, and send an email to astraype@ous-hf.no

Last day of registering: June 8th 2023

Payment:

- IBAN-number: NO8381014818847
- Swift/BIC: DABANO22
- Bank address: Danske Bank, Søndre Gate 15, 7466 Trondheim, Norway.
- Please pay in EUR to avoid extra transfer fees
- And write the name of the workshop participant in the comment field

Looking forward to seeing you in Oslo!

On behalf of the organizers,

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Chair, Norwegian Society for Medical Genetics.

18. June 2023