Norsk forening for medisinsk genetikk
DEN NORSKE LEGEFORENING

NSHG
NORSK SELSKAP FOR HUMANGENETIKK

Standardization in genetic testing

Wednesday, November 6th 2019

08.30-09.30	Registration
09.30-09.35	Welcome
	Asbjørg Stray-Pedersen, Chairperson of the Norwegian Society for Medical Genetics (NFMG)
09.35-10.30	Standardizing genomic analysis and interpretation – lessons from the UK 100,000 genomes project
	Dominic McMullan, West Midlands Regional Genetics Laboratories (WMRGL), Birmingham
10.30-11.30	The contribution of non-coding variation to rare disease
	Bill Newman, Division in Genomic Medicine Manchester University NHS Foundation Trust
11.30-12.30	Lunch break
12.30-13.15	Sudden cardiac death in Medico-Legal cases - experiences from Sweden
	Gisela Pettersson, Division for Legal Medicine Umeå
13.15-14.05	Presentation of submitted abstracts (“frie foredrag”)
	Christa Schmidt: Detection of a new AluY insertion in the NF1 gene
	Kine H. Kristiansen og Kristin S.Steinsbekk: Høykapasitetsanalyser innen medisinsk genetikk – Nye tider, nye kunnskapsbehov!
	Cecilie Rustad: Sometimes all it takes is a second patient (and international collaboration)
14.05-14.15	“Elevator pitch” – Posters (uneven numbers)
14.15-14.45	Break with posterviewing
14.45-16.15	Workshop “Bioinformatikk”, Trondhjemssalen 1
	Workshop “Arvelig kreft”, room Møllenberg
16.15-16.30	Closing remarks
16.30-	General Assembly Norwegian Society of Human Genetics (NSHG), room Møllenberg
17.15-	General Assembly Norwegian Society of Medical Genetics (NFMG), room Møllenberg
19.00-	Dinner, Trondhjemssalen 1

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Thursday, November 7th 2019

08.15-08.50	Registration
08.50-08.55	Welcome
	Elen Siglen, Chairperson of the Norwegian Society for Human Genetics (NSHG)
08.55-09.45	Ethical and legal aspects of genomic data storage, sharing and interpretation in research clinical setting
	Mahsa Shabani, Assistant Professor in Privacy Law, Ghent University
09.45-10.10	Two-dimensional variant classification: an ESHG proposal
	Gunnar Houge, Department for Medical Genetics Bergen
10.10-10.20	"Elevator pitch" – Posters del 2 (even numbers)
10.20-10.50	Break with posterviewing
10.50-11.10	Helsedirektoratets laboratorieveileder for genetiske undersøkelser (på norsk)
	Laboratory guidelines from the Norwegian Directorate of Health (in Norwegian)
	Grethe Foss, Helsedirektoratet (Norwegian Directorate of Health)
11.10-11.30	Helseplattformen i Midt-Norge - Fordeler og ulemper med strukturert journal (på norsk)
	Health platform in Middle Norway – Advantages and disadvantages with a structured patient journal (in Norwegian)
	Kurt Krogh, Helseplattformen (Health platform)
11.30-12.00	Direktoratet for eHelse: Standardisert rekvirering og svar av molekylærgenetiske analyser i NLK (Norsk Laboratoriekodeverk) og oppdatering på internasjonalt arbeid for datautveksling av genetiske analyser (på norsk)
	Directorate for eHealth: Standardized requisitioning and reporting of molecular genetic analyses in NLK (Norwegian Laboratory Code registry) and update on international work on data exchange of genetic analyses (in Norwegian)
	Evita Maria Lindholm, Direktoratet for eHelse (Directorate for eHealth)
12.00-13.00	Paneldebatt med foredragsholderne og deltaker fra medisinsk genetiske avdelingene om ønskesituasjon for «digital genetikk» (på norsk)
	Debate with speakers and representatives from medical genetic departments about the perfect situation in "digital genetics"
13.00-14.00	Lunch

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13.00-14.00	Lunch
14.00.14.45	Presentation of submitted abstracts (“frie foredrag”)
	Asbjørg Stray-Pedersen: Second-tier next generation sequencing integrated in newborn screening provides rapid molecular diagnostics of severe combined immunodeficiency
	Siren Berland: The potential of array- and NGS-based diagnostics: a FOXP1 mutation, a distal 16p11.2 microdeletion and 45,X/46,XX mosaicism explaining the phenotype, and a pre-CLL as an incidential finding
	Siri Briskemyr: 28-year-old female with familial adenomatous polyposis (FAP) caused by a de novo triple translocation
14.45-15.00	Break
15.00-16.00	Workshop “Labteknisk”, Trondhjemssalen 1
	Workshop “Svarrapportering”, room Møllenberg
	Workshop “NGS/dypsekvensering”, room Munkholmen
16.00-16.10	Awards
16.10-16.15	Closing remarks