Skien 7.-8. november 2017

Variantvurdering og variantdatabaser

Det 9. nasjonale fagmøte i humangenetikk

Variantvurdering og variantdatabaser

7.-8.november 2017
Ibsenhuset, Skien

Norsk forening for medisinsk genetikk
DEN NORSKE LEGEFORENING

NSHG

Generell informasjon:

WiFi: Ibsenhuset, passord: Ibsen203

Vi anbefaler å ta en prat med utstillerne ute i foajeen.

Middag serveres i Olavsalen på Thon Hotel Høyers.
Aperitif kl 19.30, Middag kl 20.00.
Ta med telefon!
Etter middagen fortsetter vi i peisestua og baren.

Har du praktiske spørsmål angående fagmøtet, ta kontakt med sekretariatet eller arrangører.

Vi ønsker alle et lærerikt og spennende møte!

Hilsen lokal arrangementskomité, Medisinsk genetikk, STHF Styrene i NFMG og NSHG

Variant interpretation and sharing of genetic data

November 7-8 2017

Ibsenhuset, Skien

Tuesday November 7th

0830-0930	**Registration**
0930-0935	**Welcome**
	Asbjørg Stray-Pedersen, Chair, Norwegian Society for Medical Genetics (NFMG)
0935-1050	**Deciphering the Genome through Community-Driven Approaches**
	Heidi Rehm
1050-1110	**Break**
1110-1210	**Challenges in variant interpretation – how to minimize inter and intra-laboratory inconsistencies**
	Teresa Neuhann
1210-1315	**Lunch**
1315-1415	**Parallel workshops** (Variant interpretation)
1415-1445	**Break with poster viewing**
1445-1530	**Presentation of submitted abstracts** ("frie foredrag")
1530-1545	**Nytt om genetikkportalen** v/ Gunnar Houge
1545-1600	**Closing remarks**
1600	General Assembly, Norwegian Society for Human Genetics (NSHG) Årsmøte for NSHG (In Norwegian)
1700	General Assembly, Norwegian Society for Medical Genetics (NFMG) Årsmøte for NFMG (In Norwegian)
1930	**Dinner** (Thon Hotel Høyers)

Wednesday November 8th

0830-0840	**Registration**
0840-0845	**Welcome**
	Wenche Listøl, Chair, Norwegian Society for Human Genetics
0845-0945	**Nucleotide and copy number variant detection by array and NGS in constitutional genome diagnostics**
	Nicole de Leeuw
0945-1030	**Presentation of submitted abstracts** ("frie foredrag")
1030-1100	**Break with poster viewing**
1100-1200	**Parallel workshops** (HTS and Prenatal diagnostics / CNVs)
1200-1300	**Lunch**
1300-1345	**Can IVF influence human evolution?** (In Norwegian)
	Hans Ivar Hanevik
1345-1405	**Break**
1405-1505	**Parallel workshops** (Rare syndromes and inborn errors of metabolism and Cancer genetics)
1505-1520	**CLG** v/ Olaug Rødningen
1520-1530	**Closing remarks**
	- Best presentation award

Parallel workshops

Tuesday November 7th 13:15-14:15

Variant interpretations	**Peer Gynt**
A set of variants has been sent Norwegian laboratories for interpretation according to the ACMG guidelines. The results will be presented.	

Wednesday November 8th 11:00-12:00

HTS	**Peer Gynt**
- Using the DDG2P gene panel to diagnose developmental disorders (Tine Prescott, Department of Medical Genetics, Telemark Hospital, Skien) - Detection of disease-relevant CNVs in NGS data (Hanne Sorte, Department of Medical Genetics, Oslo University Hospital, Oslo) - BigMed and practical implications for diagnostics of rare genetic diseases (Sharmini Algaratnam, Life Science Group Technology and Research, DNV GL, Høvik and Tony Håndstad, Oslo University Hospital, Department of Medical Genetics)	
Prenatal diagnostics / CNVs	**Anitra**
1) Norwegian guidelines (Mona Nystad) 2) The Nijmegen experience (Nicole de Leeuw) Why Genotype Information Matters in Prenatal SNP-based Array Diagnostics Since 2010, we routinely perform genome wide SNP-based array analysis after a normal QF-PCR test result in prenatal diagnosis in case of structural ultrasound anomalies or intra uterine foetal death. The genotype information from the SNP probes not only significantly improves the diagnostic yield, but it also enhances the quality of the diagnostic laboratory workflow. A brief overview will be given, including a few illustrative examples demonstrating the reliable detection of CNVs, more mosaic imbalances as well as copy neutral changes of homozygosity leading to the subsequent identification of pathogenic mutations in recessive disease genes or uniparental disomies (UPD). Exome sequencing for prenatal diagnostics 3) Reporting of low penetrance variants (Ragnhild Glad) 4) Expanding indications for prenatal CNV analysis? (Ragnhild Glad)	

Parallel workshops

Wednesday November 8th 14:05-15:05

Rare syndromes and inborn errors of metabolism	**Peer Gynt**
Daily life with variant interpretation – learning by doing. Six groups will each share 1-3 illustrative cases from their own experience – for example, variants that were interpreted incorrectly initially or problems with a pipeline.	
Cancer genetics, inherited cancer	**Anitra**
- Mev Dominguez Valentin: The Prospective Lynch Syndrome Database - Acetylsalisylbehandling ved Lynch syndrom - Retningslinjer for oppfølgning av Li-Fraumeni syndrom - Nasjonale handlingsprogrammer for arvelig kreft. Nasjonalt nettverk for arvelig kreft (under Helsedirektoratet).	

Presentation of submitted abstracts

Tuesday November 7th 14:45-15:30

14:45-15:00	Ny kopitallsanalyse ved AMG, St. Olavs Hospital	**Trine Vold** Seksjon for medisinsk genetisk laboratorium, APMG, St Olavs Hospital HF
15:00-15:15	Three faces of a unique CDKN1C-mutation in a boy: BWS, IMAGe and developmental delay, and confirmation of a novel transcript CDKN1C_A6NK88	**Siren Berland** Center for Medical Genetics and Molecular Medicine, Haukeland University Hospital, Bergen, Norway
15:15-15:30	An electronic clinical decision support system for actionable pharmacogenomics: Prototype demonstration with azathioprine and thiopurine methyltransferase	**Sjur Urdson Gjerald** Oslo University Hospital, Department of Medical Genetics

Wednesday November 8th 09:45-10:30

09:45-10:00	Homozygous KIDINS220 loss-of-function variants in fetuses with cerebral ventriculomegaly and limb contractures	**Inger-Lise Mero** Department of Medical Genetics, Oslo University Hospital, Oslo, Norway
10:00-10:15	Clonal hematopoiesis in donor cells developed into acute myeloid leukemia in the recipient after allogeneic stem cell transplantation	**Randi Hovland** Center for Medical Genetics and Molecular Medicine, Haukeland University Hospital, Bergen, Norway
10:15-10:30	The Northern Norwegian KBG syndrome series including a patient with normal ANKRD11 sequencing, but impaired gene expression	**Marie Smeland** Dpt of Medical Genetics, University Hospital of North Norway, Tromsø, Norway

Poster presentations

Tuesday November 7th 14:15-14:45

Guro Meldre Pedersen	Building trust in precision medicine
Mev Dominguez-Valentin	Potential implication of ATM c.3806A>G in familial breast cancer
Gregor D. Gilfillan	Determining the bacterial microbiome with a novel ultra-high-throughput 16S rDNA sequencing method.
Jonas Langerud	Risk assessment of BRCA1 variants with unknown clinical significance
Trine Prescott	A year with DDG2P
Cecilie Rustad	SPARC-related osteogenesis imperfecta with a myopathy-like presentation
Marie Smeland	Angiokeratomas and severe behavioural disturbances as the clue to Beta-mannosidosis – caused by an intragenic copy number variant in MANBA
Bente A. Talseth-Palmer	After a decade of searching for modifier genes affecting disease expression in Lynch syndrome – Where are we now?

Wednesday November 8th 10:30-11:00

Sharmini Alagaratnam	Variant databases and data sharing in the BigMed project
Siren Berland	HUWE1 variants cause dominant X-linked intellectual disability: a clinical and genetic study of 21 patients
Øyvind Evju	Norvariome
Asgeir Lande	Temple Syndrome as a Differential Diagnosis to Prader-Willi Syndrome. Identifying three New Patients.
Mariève J. Rocque	Investigating the mutation frequencies of a PPAP associated POLE variant
Wenche Sjursen	Uten tittel
Marie Smeland	A novel PIK3CA mutation in Megalencephaly – capillary malformation syndrome with extreme brain overgrowth and 45,X mosaicism: related or coincidental phenomena?