

# Genomic Medicine in Action symposium

May 28<sup>th</sup> 2019  
BioClinicum aula, Solna

9.00-9.30	Registration and coffee
9.30-12.00	<b>SESSION 1</b>
9.30	<i>Welcome words</i> <b>Jenni Nordborg</b> , Life Science office, Sweden
9.50	<i>100,000 Genome Project – Transforming Healthcare</i> <b>Mark Caulfield</b> , Chief Executive and Chief Scientist, Genomics England
10.30-11.00	<b>Coffee break</b>
11.00	<i>Genomic Medicine Sweden – building a national infrastructure</i> <b>Richard Rosenquist Brandell</b> , Karolinska Institutet
11.30	<i>Deployment of Genetic Data Management System in Cloud Platform</i> <b>Mikko Rotonen</b> , IT-Development Director, Helsinki University Hospital, Finland
12.00-13.00	<b>Lunch</b>
13.00-15.10	<b>SESSION 2</b>
	<b>Track 1 – Rare diseases</b> Room: Torsten N Wiesel J3:04
13.00	<i>NGS innovations to uncover new genes for intellectual disability: from research to diagnostics and back</i> <b>Lisenka Vissers</b> , Dept of Human Genetics, Radboud UMC, Nijmegen, The Netherlands
13.50	<i>Clinical Implementation of Genomics for Rare Disease</i> <b>Marc LePage</b> , Genome Canada
14.15	<i>Whole genome characterization of germline chromosomal rearrangements reveals underlying disease mechanisms</i> <b>Anna Lindstrand</b> , Karolinska Institutet

14.40	<p><i>Long read sequencing for clinical applications</i></p> <p><b>Lars Feuk</b>, Department of Immunology, Genetics and Pathology, Uppsala University, Sweden</p>
	<p><b>Track 2 – Cancer</b></p> <p>Room: Sune Bergström aula</p>
13.00	<p><i>Feasibility and impact of whole genome and whole transcriptome sequencing in the diagnostic work-up of hematological neoplasms</i></p> <p><b>Claudia Haferlach</b>, MLL Munich Leukemia Laboratory, Munich, Germany</p>
13.40	<p><i>Integrative sequencing and target identification in acute leukemia</i></p> <p><b>Thoas Fioretos</b>, Lund University</p>
14.05	<p><i>Molecular Pathology 3.0: complex biomarkers enter clinical stage</i></p> <p><b>Albrecht Stenzinger</b>, Institute of Pathology, University Hospital Heidelberg, Germany</p>
14.45	<p><i>Genomic tools for diagnosis and immune characterization of cancer of unknown primary and rare cancers</i></p> <p><b>Jonas Nilsson</b>, Gothenburg University</p>
	<p><b>Track 3 – Infectious Diseases</b></p> <p>Room: Ulf von Euler J3:06</p>
13.00	<p><i>Clinical Metagenomics and bioinformatics pipelines</i></p> <p><b>Natacha Couto</b>, Department of Medical Microbiology and Infection Prevention, University Medical Center Groningen, University of Groningen, Netherlands</p>
13.40	<p><i>Neisseria gonorrhoeae - Rapid antibiotic resistance predictions using Nanopore technology</i></p> <p><b>Magnus Unemo</b>, Department of Laboratory Medicine, Microbiology, Örebro University Hospital, Sweden</p>
14.10	<p><i>NGS in clinical microbial diagnostics</i></p> <p><b>Hedvig Engström Jakobsson</b>, Clinical Microbiology, Sahlgrenska University Hospital, Gothenburg</p>
14.40	<p><i>Shotgun metagenomics of the human microbiome</i></p> <p><b>Fredrik Boulund</b>, Center for Translational Microbiome Research, Department of Molecular, Tumour and Cell Biology, Karolinska Institutet</p>
15.10-15.40	<p><b>Coffee break</b></p>
	<p style="text-align: center;"><b>SESSION 3</b></p>

15.40-16.20	<p><b>SME presentation and discussion</b></p> <p>Moderated by Frida Lundmark, Vinnova</p> <p><b>1928</b>, Kristina Lagerstedt, Gothenburg</p> <p><b>SAGA Diagnostics</b>, Lao Saal, Lund</p> <p><b>Moleculink</b>, Mathias Howell, Uppsala</p> <p><b>CartaNA</b>, Malte Kuhnemund, Stockholm</p> <p><b>Genagon Therapeutics</b>, Simon Fredriksson, Stockholm</p>
16.20	<p><i>Rare Diseases Sweden – challenges and opportunities for the rare disease field</i></p> <p><b>Beata Ferencz</b>, Rare Diseases Sweden</p>
16.40	<p><i>Patient-customized Oligonucleotide Therapy for an Ultra-Rare Genetic Disease</i></p> <p><b>Timothy Yu</b>, Division of Genetics &amp; Genomics, Boston Children’s Hospital and Department of Pediatrics, Harvard Medical Medical School, USA</p>
17.10	<p>Closing words</p> <p><b>Mats Ulfendahl</b>, Director Research Region Östergötland and <b>Hannie Lundgren</b>, Director Research &amp; Development Region Skåne</p>
17.20-19.00	Mingle