

# Expansion Disorders Seminar

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Date: Monday, November 10

Venue: Bikuben (Auditoriet), Haukeland University Hospital

## Program Overview

**09:45-09:55** Coffee and fruit, registration

**09:55-10:00** Welcome

Speaker: committee (Siren Berland, Helle Høyer, Linda Mathiesen, Gry Hoem)

**10:00-10:15** Basic overview of expansion disorders

Speaker: Linda Mathisen, MD, Department of Medical Genetics, OUS

**10:15-10:35** Expansion analysis in Norway

Speaker: Helle Høyer, PhD, Department of Medical Genetics, STHF

**10:35-10:50** Coffee break

**10:50-11:40** Huntington Disease and Myotonic Dystrophy: Somatic expansion in driving disease and as a therapeutic target, part I

Speaker: Darren Monckton, Prof. in Genetics University of Glasgow, Scotland

**11:40-11:50** Kompetansebygging og kunnskapsspredning, inkl orphakoder

Speaker: Mari Bakken, Medisinsk rådgiver og barnelege, Nasjonalt senter for sjeldne diagnoser

**11:50-12:10** Myotonic Dystrophy, clinical overview

Speaker: Kristin Ørstavik, MD, PhD, Department of Neurology, Section for rare neuromuscular disorders, OUS

**12:10-13:00** Lunch Break

**13:00-13:35** Huntington and DM1, part 2

Speaker: Darren Monckton, Prof. in Genetics University of Glasgow, Scotland

**13:35-14:00** Treatment in Huntington Disease

Speaker: Lasse Pihlstrøm, MD and Prof. II at Dep of Neurology, OUS

**14:00-14:50 Detection of repeat expansions in patients with ataxia, neuromuscular disorders and neurodegeneration**

Speaker: Håkan Thonberg, Associate Professor in Medical Genetics at Karolinska Institute, Sweden

**14:50-15:05** Coffee break

**15:05-15:25 Clinical view on RFC1-CANVAS in Norway**

Speaker: Sjur Prestsæter MD, PhD fellow, dep of Neurology, OUS

**15:25-16:25 RFC1 genetics, clinical features and mechanisms**

Speaker: Andrea Cortese, MD and professional research fellow in Neurology, Department of Neuromuscular Diseases at University College London's Institute of Neurology (Queen Square), UK and at the University of Milan, C Besta Neurological Institute, Italy

**16:25-16:55 Fragile X Related Disorders**

Speaker: Gry Hoem, MD, PhD, Department of Medical Genetics, UNN

**16:55-17:00** wrap up and closure

Speaker: committee