Protein aggregation disorders: from clinic to therapy

Organizers:

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Date: **30-31 October, 2019**

Location: LUMC

Registration: send an email to Nivard@lumc.nl

Note: Friendly request to mention also the name of your department and institute at the registration!

Aberrant protein homeostasis can lead to accumulation and aggregation of proteins in insoluble cellular structures. Protein aggregation is a hallmark feature of many adult neurodegenerative and neuromuscular disorders although the genetic cause and symptoms of these diseases highly varied. Despite varied genetic causes for the protein aggregation disorders, many of these diseases sharing common cellular and molecular mechanisms. Accumulation of proteins and subsequently its aggregation is caused by malfunctioning of protein homeostasis and protein folding machineries. Also aberrant protein homeostasis is a hallmark of aging tissues, in neurodegenerative and neuromuscular disorders the effect is more pronounced and is associated with pathology. Some disorders, such as OPMD, FXTAS and polyglutamine disorders are rare and have a clear genetic component while for more common disorders such as Alzheimer and Parkinson, there is often no clear genetic cause. Thus, genetic variants of protein aggregation disorders are important in the unravelling of the molecular and pathological processes of more common protein aggregation disorders.

This course will discuss the main molecular mechanisms regulating protein homeostasis and protein folding with a focus a mechanistic foundation in protein aggregation disorders. We will focus on four selected diseases and will discuss genetic, clinic, and current knowledge of disease mechanisms and models for examples. The end point of this course will be development of novel therapeutic strategies to combat protein aggregation in pathological conditions.

There is a minimum of 12 and a maximum of 30 places. Deadline for registration 14 October, 2019.

The course is free of charge for all personnel of MGC associated institutes and for participants working in a research group of one of the course teachers. Participants from outside these organizations pay € 250 for this course. In case of no show without any notification you will be charged 100 euro.

Programme

Day 1, 30 October 2019:
Introduction: an overall concept for protein aggregation and pathological conditions: similarities and variations between condition (spectrum of conditions, genetics, causes). And the path to therapeutics.

Morning session:
09:00-09:15 Registration and coffee/tea: (Restaurant building 2 – ground floor)
Molecular Mechanism regulating protein accumulation and folding: molecular regulators of protein aggregation.
LUMC room P5-34

09:30	Lecture 1: Mitochondrial dysfunction	- *Pier Mastroberardino* (Erasmus MC)
10-:15	Lecture 2: Autophagy	- *Fulvio Reggiori* (Utrecht MC)

11:00-11:15 coffee/tea

11:15	Lecture 3: Modifiers of protein aggregation and toxicity'	- *Ellen Nollen* (UMCG)
12:00	Lecture 4: The UPS	- *Huib Ovaa* (LUMC)

12:45-13:30 Lunch

***Afternoon session*:** Disease examples: genetics, symptoms, models

13:30	Parkinson	- *Wim Mandemakers* (ErasmusMC)
14:15	SCA3	- *Ronald Buijsen* (LUMC)

15:00-15:15 coffee/tea

15:15	OPMD	- *Vered Raz* (LUMC)
16:00	CADASIL	- *Saskia Lesnik-Oberstein* (LUMC)

17:00 drinks and meet with the speakers.

Day 2, 31 October 2019, LUMC room P5-34

***Morning session*:**

09-:00	Lecture 1: Clinics: a neurologist and a patient	- *Suzanne de Bot* (LUMC)

Translational session - examples of therapeutic developments:

10:00	Lecture 2: Gene therapy development in the lab	- *Willeke Roon-Mom* (LUMC)

10:45-11:00 coffee/tea

11:00	Lecture 3: Therapeutic development: technology transfer	- *Frits Fallaux (*Luris, LUMC*)*
11:30	Lecture 4: Therapeutic development in a company	- *Thomas de Vlaam* (Amylon)

12:30-13:15 Lunch

13:15-17:00 Afternoon session: Translational development: an active session and presentations
Ronald Buijsen, Willeke Roon-Mom, Vered Raz (LUMC)
17:00 drinks