Protein aggregation disorders: from clinic to therapy

Organizers: Willeke Roon-Mom[1], Ronald Buijsen[1], Wim Mandemakers[2], Vered Raz[1]

[1] Human Genetics, Leiden University Medical Centre

[2] Erasmus Medical Centre

Date: **once every two years**

Location: LUMC

Registration

Note: Friendly request to fill in 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.

The course is free of charge for all personnel of MGC associated institutes. Participants from outside these organizations pay € 200 for this course.