



11th
December
2023

PhD Colloquia 2023

**Rare genetic forms of
Amyloidopathies:
iPSC disease modelling
and RNA therapy
development**



A108



14:00



**Prof. Willeke
van Roon-Mom**

LUMC - Leiden University

In this seminar I will highlight the importance of iPSC disease modelling and disease modelling for very rare genetic disorders of the brain. As a first example I will outline the preclinical development of an RNA targeting therapy for a very rare familiar form of Cerebral Amyloid Angiopathy (D-CAA) caused by a point mutation in the APP gene. Results from traditional cell and mouse models will be discussed, followed by the characterization of a brain organoid model made from D-CAA patient derived induced pluripotent stem cells (iPSCs). Finally, I will outline results from a recently started project to use our knowledge of iPSC modelling and analysis to unravel disease pathology in a patient with a novel and unique APP mutation.