Focus of research group (I)

Name PI: Mirjam Langeveld, MD PhD
Department: Endocrinology and Metabolism, Amsterdam UMC

Size of research group
My research group is embedded in SPHINX, the Amsterdam Lysosomal Center. The research is headed by two PI’s: Carla Hollak and Mirjam Langeveld. Team members are Simon Korver (PhD student), Sanne van der Veen (PhD student), Mohamed El Sayed (PhD student), Shirley Klein (research nurse), Rika Rhee-Martha (research nurse), Laura van Dussen (epidemiologist) and Mareen Datema (datamanager).

Current mission, vision and aims
My long term research goal for Fabry cardiomyopathy is to:
• understand the pathophysiology and identify potential treatment targets
• develop new diagnostic strategies for early disease detection
• define relevant functional outcome parameters of treatment
Focus of research group (II)

Current expertise
My research focusses on inherited metabolic diseases in adults, with an emphasis on lysosomal storage disorders (LSD’s) and fatty acid oxidation disorders (FAOD). The LSD research is embedded in SPHINX, a collaboration of the departments of Radiology, Internal Medicine and Paediatrics and the Laboratory of Genetic and Metabolic Diseases. With regard to LSD my main focus is Fabry disease, in particular Fabry cardiomyopathy.

Current funding
• F20-Study of the Safety and Efficacy of PRX-102 Compared to Agalsidase Beta on Renal Function (BALANCE)
• F30-Extension Study of 1 mg/mL Pegunigalsidase Alfa in Patients With Fabry Disease (BRIDGE)
• International Fabry Genotype Fenotype Study and Open Access Registry Development (ZonMW VIMP round GGG1)
• Stofwisselkracht
Future plans

**Short-term plan (1-2 year)**
- Study exercise intolerance in Fabry disease and its relationship with early signs of cardiac dysfunction and/or skeletal muscle pathology
- Perform longitudinal study on imaging and electrophysiology characteristics and biomarkers during the development of Fabry cardiomyopathy

**Long-term plan (>2 year)**
- Develop disease models to study pathophysiology and potential treatment targets for Fabry cardiomyopathy
- Test pharmacological interventions

The necessary infrastructure is available

**Collaboration in ACS**
- Cardiology (AMC): Matthijs Boekholdt and Pieter Postema
- Physiology (VUMC): Jolanda van der Velden