

llse Luyckx

In-depth characterization of mice lacking SMAD6 expression using an immune-histological approach

Duration	6 weeks
Short Bio	I'm a molecular biologist, trained in cardiogenetics. During my PhD, I focussed on the identification of genetic causes for (bicuspid aortic valve)-related aortopathy, and provided insights into expanding the phenotypic spectrum of patients with thoracic aortic aneurysmal disease. More recently, I complemented my genetic and molecular skills with the study of cellular and murine disease models to understand how these genetic aberrations cause and/or influence disease. I focussed primarily on phenotyping of genetically-modified mouse models for (bicuspid aortic valve)-related aortopathy during development and adulthood using (imaging) techniques including echocardiography and histology.
Home Institution	University Hospital Antwerp (UZA), Antwerp, Belgium
Host Institution	Leiden University Medical Center (LUMC), Leiden, The Netherlands
Project Description	My project aims to understand the impact of SMAD6-deficiency on human congenital genetic disorders, with a primary focus on SMAD6- induced bicuspid aortic valve-related aortopathy. With this research stay, I expanded my expertise in the phenotypic characterisation of genetically-modified embryonic pups using histology. The identification of the involved structures, and cell types at the crucial developmental time point allowed me to further pinpoint the underlying processes causing disease. Additionally, the ability to use an alternative imaging tool, i.e. light-sheet imaging, to screen for bicuspid aortic valves in embryonic pups was explored.
Personal Statement	With this fellowship, my inspiration to improve the life of patients with congenital cardiovascular disease got even more boosted.

