

*Stellingen behorend bij het proefschrift:*

## **CLINICAL AND GENETIC STUDIES IN INHERITED CARDIOVASCULAR MALFORMATIONS**

NPHP4 variants contribute to a wide range of congenital heart malformations and more complex defects within the heterotaxy spectrum (this thesis)

Left ventricular outflow tract obstructions can be accompanied by thoracic aortic aneurysm, right-sided heart defects and septal defects (this thesis)

Aneurysms-osteoarthritis syndrome predisposes patients to aggressive and widespread cardiovascular disease, and early elective surgical treatment is warranted (this thesis)

A molecular diagnosis of aneurysms-osteoarthritis syndrome will allow early and reliable identification of patients and family members at risk for major cardiovascular complications (this thesis)

It is important to distinguish hereditary cardiovascular malformations from sporadic forms to identify potential associated morbidities, family members at risk and to otherwise provide appropriate counseling (this thesis)

Syndromic and non-syndromic forms of thoracic aortic aneurysms can not be strictly separated but comprise a clinical continuum

Estimating true aortic size (and reconciling discrepant reports) can be difficult since no consensus document or uniform definition of normal diameters of the thoracic aorta have been published in recent years (Larsson, Annals of Surgery 2011)

We invariably overestimate the short-term impacts of new technologies and underestimate their longer-term effects (The First Law of Technology Adoption)

Samenwerking resulteert in een geheel dat groter is dan de som der delen. Dit geldt voor de TGF- $\beta$  en Ang II type 1 receptor pathway, maar ook voor wetenschappers

Kennis is nog geen wijsheid (Arthur Schopenhauer)

Een eigen gezin lijkt soms een proeftuintje voor een geneticus

*Ingrid van de Laar,  
27 juni 2012*