

Stellingen behorende bij het proefschrift:

Genetics of Cardiovascular Disorders

1. A negative family history should not preclude cardiovascular screening in first-degree relatives of patients with thoracic aortic disease. *(this thesis)*
2. Inflammation and mitochondrial dysfunction play an essential role in the pathogenesis of thoracic aortic disease, including heritable disorders such as Marfan syndrome. *(this thesis)*
3. Hypoplastic left heart syndrome may result from abnormal myocardial development caused by the absence of plakophilin-2. *(this thesis)*
4. Biallelic variants in *ALPK3* cause a syndromic form of pediatric cardiomyopathy, that shows unique evolution from dilated to hypertrophic cardiomyopathy. *(this thesis)*
5. Tail-anchored membrane protein insertion is essential for vertebrate cardiac function. *(this thesis)*
6. Discovering a gene-disease relationship (and getting it published) is easier than disproving it.
7. Even researchers who do not explicitly provide diagnosis to patients should be aware that their published findings may be used as support for decisions made in clinical settings. *(Daniel MacArthur, Nature 2014)*
8. No disease will be cured without proper understanding of the molecular function of the causative gene. *(Frank Kooy and Rob Willemsen, Neuroscience 2017)*
9. Germline genome editing is the ultimate form of genetic medicine.
10. This is just the beginning of the era of polygenic risk scores. *(Robert Plomin, Blueprint 2018)*
11. On ne voit bien qu'avec le coeur. L'essentiel est invisible pour les yeux. *(Le Petit Prince)*