



Applicant Dr. Connie Bezzina, Amsterdam UMC

E-mail Address c.r.bezzina@amsterdamumc.nl

Project Title

Identifying novel genetic factors predisposing to inherited cardiac disorders

Project Summary

Inherited cardiac disorders arise from genetic variations and can manifest as cardiomyopathies or arrhythmic disorders. For example, hypertrophic cardiomyopathy (HCM) results from abnormal heart muscle thickening, which can cause complications like arrhythmias or sudden death. Long QT Syndrome (LQTS), an arrhythmic disorder, shows prolonged intervals on an ECG, increasing the risk of fainting or severe events. The Mendelian inheritance model has deepened our understanding of genes underpinning these disorders. Yet, phenotypic variability in those with the same genetic anomaly suggests the influence of 'modifier genes'. To investigate, we're launching Genome Wide Association Studies (GWAS) to pinpoint genes that affect susceptibility or severity of these conditions. We've collected samples from Europe but need matching control samples, especially those tested on the GSA chip, and basic demographic data. Through GWAS, we aim to enhance our understanding and patient care.

Keywords

genome-wide association, phenome-wide association, interactive browser, data sharing