Application number/Title: 34904 - Meta-analysis of genome-wide association studies for chronic venous disease

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Keywords provided by the Applicant PI to describe the research project: snps, genetics, susceptibility-loci, varicose-veins

Application Lay Summary:

This project aims at the detection and characterization of common and rare risk variants and hence genes associated with chronic venous disease (CVD). CVD is a multifactorial condition, defined as ‘morphological and functional abnormalities of the venous system of long duration manifested either by symptoms and/or signs indicating the need for investigation and/or care’. The identification of genetic susceptibility factors in the aetiology of CVD is fundamental for future translational and clinical research in order to understand disease mechanisms and to be able to translate knowledge of involved genes into better prevention and/or treatment of patients. CVD represents one of the most common disorders among populations of Western countries. Prevalence of CVD in Germany and the USA was estimated at ~30%. CVD has a major socioeconomic impact. The financial burden of CVD treatment on the health-care system is immense with an estimated cost of US$3 billion per year in the USA and up to 2% of the total health-care budget of all Western countries. Despite the frequency of CVD and its’ considerably high impact on health-care budgets and on patients’ quality of life the underlying aetiology and pathophysiology are still poorly understood. We will perform a genome-wide association study with 4,000 German CVD cases and 4,000 non-CVD controls using Illumina’s GlobalScreeningArray, followed by imputation, to detect/characterize common and rare genetic risk variants/genes associated with CVD. To increase our sample size, we want to perform a meta-analysis with imputed genotype data of the UK Biobank. We will perform state-of-the-art association analyses for a near complete catalogue of variants with a frequency >0.1% in Europeans to identify statistically convincing causal genetic variants and to provide a powerful substrate for experimental elucidation of disease mechanisms. We would like to include genotype data of the full cohort or at least of all samples for which information about CVD/varicose veins is available. Individuals having CVD/varicose veins will be included in our genetic
study as cases (C2-C4 according to CEAP classification) and individuals without CVD/varicose veins as controls.