Application number/Title: 33519 - Phenome-wide association study of the C282Y mutation in HFE

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Keywords provided by the Applicant PI to describe the research project:
hfe, phenome-wide-association-study

Application Lay Summary:

The C282Y mutation (rs1800562) in HFE has a frequency of about 6% in Northern European populations and is the main cause of hereditary hemochromatosis. The excess stored iron can have serious health consequences, e.g. liver cirrhosis at young age. The UK Biobank provides a unique opportunity to investigate disease prevalences in relation to genotype status under consideration of relevant life style factors. Given that more than 1,500 homozygous and almost 50,000 heterozygous carriers can be expected, moderately increased prevalence rates in carriers can be identified. The project will provide new insights into the clinical consequences of increased iron levels caused by the C282Y mutation in HFE. The results have the potential to improve diagnosis and treatment of disease. Based on the genotype data, three groups with a genetic predisposition for different iron levels are defined. The prevalence of a variety of health conditions will be compared between these groups. Conditions will be defined based on ICD9/ICD10 and surgery codes from hospital episodes and the self-reported medical conditions. Other biochemical and environmental factors important for iron status (serum levels, alcohol, meat, fruit and vegetable consumption) will be incorporated in sub-group analyses. Furthermore, we plan to study whether additional genetic risk factors play a role for clinical consequences associated with the C282Y mutation. We plan to study the full cohort.