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| Host department: Nottingham |
| Project Title: |
| Improving the diagnosis of Haemochromatosis in primary care through development and validation of a case-finding algorithm in primary care electronic health records |
| Proposed supervisory team: |
| Professor Nadeem Qureshi, MRCGP DM, University of Nottingham –GP & primary care health service researcher with expertise identify common inherited disorders in primary care  Dr Mathew Grainge, PhD, University of Nottingham, statistician with experience of risk prediction modelling  Dr Barbara Iyen, MRCGP PhD, University of Nottingham –GP & primary care epidemiologist, with expertise in risk prediction modelling  Susan Hancock, FRCGP, University of Central Lancashire – GP educator with special interest in  Haemochromatosis  Julie Brown, Lecturer in Nursing and Public Health, Queen’s University Belfast- Involved in setting up primary care case-finding services in primary care |
| Potential for cross consortium networking and educational opportunities: |
| This project will be co-designed with Haemochromatosis UK ([*https://www.haemochromatosis.org.uk*](https://www.haemochromatosis.org.uk)*)* , in particular with their Clinical Advisory Panel[[1]](#footnote-1), representing a range of clinical specialities, including primary care and nursing. We will work with the charity’s clinical advisors work with a range of academic institutions including University of Exeter Medical School, University of Central Lancashire Medical School, St George’s Medical School (London) and Queen’s University (Belfast) and clinical organisations including Addenbrookes Hospital, Great Ormond Street Hospital, NHS Glasgow & Clyde NHS Trust and others. We anticipate that there would be a wide range of opportunities for collaboration and networking internationally e.g. Western Australia.  The research team have experience of developing and translating risk prediction algorithms for identifying common inherited condition in primary care and developed national networks for implementation e.g. with Academic Health Science Networks. (1,2) |
| Project description: |
| Genetic haemochromatosis is the UK’s most common genetic condition. A recent study by York Health Economics Consortium, based upon UK Biobank data, suggests that up to 1.2 million people are at risk.(3) There has been similar work in the CPRD primary care database.(4)  Untreated, the condition can cause liver cancer, diabetes, liver disease, arthropathy, cardiomyopathy and chronic fatigue. Treatment is simple (regular phlebotomy) and can greatly reduce the incidence of co-morbidities.  Yet, the condition is significantly under-diagnosed – just 20,698 people are known to be undergoing NHS treatment. Low rates of diagnosis are attributable to the high degree of variability of disease expression, lack of reliable symptoms to aid diagnosis and poor clinician knowledge of the condition.  This project aims to develop an algorithm which can probabilistically identify patients at risk within primary care, based upon pre-existing blood test results and clinical characteristics. This will involve similar methodology to the approach we have developed to identify familial hypercholesterolaemia in primary care, using the FAMCAT algorithm  Phase 1: Systematic review of identification of Haemochromatosis in non-specialist setting  Phase 2: Consensus meeting to consider other predictors of Haemchromatosis  Phase 3: CPRD GOLD database analysis to develop the Haemochromatosis algorithm in primary care  Phase 4: Validation of the algorithm in the CPRD Aurum database |

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| Training and development provision by host: |
| *Formal training:*  JB Systematic Review course, University of Nottingham  Clinical epidemiology module of the MPH, University of Nottingham  Also PhD students will be encouraged to explore training opportunities in other academic institutes |
| *Informal training:*  A range of training course available through host institution, through University graduate school and School of Medicine research skills training opportunities |
| *PPIE:*  The study will be co-designed with Haemochromatosis UK , with active support for study implementation and dissemination/engagement  Also the PRISM research group has an established PPIE group that will support development of the PhD. To ensure we engage underserved communities, we will work with Nottingham Healthwatch |

References

1. Weng S, Kai J, Akyea R, Qureshi N. Detection of familial hypercholesterolaemia: external validation of the FAMCAT clinical case-finding algorithm to identify patients in primary care. *Lancet Public Health*. 2019;4(5):e256-e64.
2. Qureshi N, Akyea RK, Dutton B, et al. Case-finding and genetic testing for familial hypercholesterolaemia in primary care. Heart. 2021;107(24):1956-1961. doi:10.1136/heartjnl-2021-319742
3. Evaluating the Cost of Illness of Genetic Haemochromatosis in the UK, January 2022 – York Health Economics Consortium, ISBN 9781399907361
4. Crooks, C., West,J., Solaymani‐dodaran, M., Card, T."The epidemiology of haemochromatosis: a population‐based study." Alimentary Pharmacology & Therapeutics 2009;29.2.

1. [↑](#footnote-ref-1)