PlaceME@SMI 2021: Sociological Studies / Wakefield Clinical Commissioning Group - Estimating service need among population groups practising close relative marriage

Project Leader / Supervisor	Sarah Salway / Duncan Cooper
Department / Organisation	Sociological Studies / Wakefield Clinical Commissioning Group
Project title	Estimating service need among population groups practising close relative marriage
Project reference	PMSMI – 04 - 2021
Location of placement	UoS / Wakefield CCG / remote working

Summary of research project

This research project will involve the development of a pre-existing model that estimates the risk of genetic disorders caused by close relative marriage (consanguinity).

The successful applicant will:

- Develop the model's accuracy – by identifying factors contributing to the population's risk of genetic disorders and, working alongside stakeholders, update the modelled estimates to reflect this research.
- Develop the model's accessibility – by producing a user friendly product that allows a wide range of stakeholders to investigate estimates of risk for their area.
- Disseminate the final product and present their findings to stakeholders via written and oral reports.

Background:
Close relative (consanguineous) marriage is widely practised globally with recognised benefits to couples and their families. However, this marriage pattern has often been stigmatised in the UK. Marriage between close blood relatives is linked to an increased risk of genetic disorders, particularly recessive genetic disorders. Population risk of any congenital anomaly is around 6% among cousin couples compared to around 3% among unrelated individuals. Around 90% of couples who are close blood relatives will not be affected. Studies show that populations practising close relative marriage often have inadequate access to accurate genetic information and low levels of knowledge. Furthermore, couples who have a child with a genetic condition may not gain access to genetic services and may therefore not have the information and

support needed to make informed reproductive decisions.

Accurate estimates of risk and need for services at population level are difficult to obtain directly from routine service data. Therefore, it is useful to produce modelled estimates. Bernadette Modell (Emeritus Professor of Community Genetics at UCL) developed a method for producing estimates of service need for haemoglobin disorders (Modell et al. 2007) and subsequently extended this method to estimate service needs for consanguinity-related genetic disorders. The method used 2001 census data on ethnicity (with a correction for increases in ethnic minority population size over time) and observations from the 5-year prospective Birmingham birth study (Bundey and Aslam 1993). A series of estimates were produced for England and Wales by 2006 electoral ward, primary care trust (PCT), and strategic health authority (SHA). In 2020, preliminary work has been undertaken to identify how the model can be updated in light of changes in ethnic make-up of local areas, other shifts in 'input variables' used in the model and new research data that can inform model parameters. The model generates a number of useful measures that can inform service planning e.g. annual births with consanguinity associated birth disorder; annual neonatal, infant and under-5 deaths due to consanguinity associated birth disorder

Links to external organisation(s), including international partners (if relevant)

West Yorkshire and Harrogate Health and Care Partnership will be a partner in this project, with Duncan Cooper Programme Analyst, who is based at Wakefield Clinical Commissioning Group acting as co-supervisor to the student.

The national Steering Group on Consanguinity and Unmet need for Genetic Services will be another partner, with various SG members, including Bernadette Modell also providing guidance as relevant and possible during the placement.

Tasks to be performed during the placement

- Familiarization with current model parameters and calculation steps
- Familiarization with subject matter
- Identification of parameters to update, in discussion with experts
- Searching/accessing relevant research evidence and routine statistics to inform new model parameters
- Discussion with stakeholders and experts regarding transfer of model into a new package, and potential for 'front end' development
- Updating of the model (new parameters and new format if deemed sensible)

- Discussions with stakeholders to identify useful outputs from the model
- Creation of model 'products' – presentation of outputs in text and visual formats
- Comparison of model outputs with other sources of relevant data ("validation" work)
- Preparation of report and oral presentation

Project's outputs

- Revised, updated model
- Presentation to the national Steering Group (which includes senior clinicians and policy makers)

Person specification

- Experienced user of Excel
- Experienced user of R
- High level of attention to detail
- High level of numeracy
- Interest in health and familiarity with common measures of mortality
- Literature searching, and evidence review skills
- Ability to work well with only remote supervision
- Good written and oral presentation skills

Terms and conditions

The placement will take place between the UoS / Wakefield CCG / and remote working (subject to covid restrictions), for 35 hours a week over a period of 6 weeks between the middle of June and end of September. The successful candidate will be awarded a £2012 bursary by SMI.

Students who will be awarded the placement are expected to write two blog posts (x 300 words) for SMI about their experiences.

How to apply?

Candidates should send a CV and a motivation letter in PDF formats by 19 April 2021 to Abigail Tazzyman a.tazzyman@sheffield.ac.uk in an email entitled 'PlaceME@SMI – Estimating service need among population groups practising close relative marriage.

Shortlisted candidates will be required to take a test and be interviewed by Sarah Salway and Duncan Cooper by the end of May 2021.

Contact details	Sarah Salway s.salway@sheffield.ac.uk Duncan Cooper duncan.cooper4@nhs.net

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