Institution: De Montfort University

Unit of Assessment: 22

Title of case study: Ethnicity and Screening for Sickle Cell/Thalassaemia

1. Summary of the impact

Antenatal screening aims to identify genetic carriers of sickle cell/thalassaemia in order to provide prospective parents with "informed choice". Throughout the period January 2008-July 2013, the NHS in England has used a Family Origins Questionnaire in connection with sickle cell/thalassaemia screening derived from our research programme. The original policy issue concerned whether or not it is possible/desirable to target antenatal screening for sickle cell/thalassaemia by means of an ethnicity question. The policy problem was that socially constructed "ethnicity" categories correspond imperfectly and to an unknown degree with actual prevalence of genetic carriers. The screening question based on our research now guides the offer of initial screening and/or further laboratory tests for all pregnant mothers in England.

2. Underpinning research

The potential policy problem of the (mis)use of socially constructed ethnicity categories to target screening for the genetic carrier states for sickle cell and thalassaemia was first identified by Simon M Dyson (DMU 1991 -, Professor of Applied Sociology)[1] The possible solution, using UK census categories substantially adapted to reflect the requirement to identify particular ethnic groups at higher risk of carrying genes associated with sickle cell/thalassaemia, was also first identified by Dyson [2].

In 2002-3, Simon Dyson was principal investigator (with other DMU investigators: Lorraine Culley [DMU 1990 - , Professor Social Sciences in Health]; and Stephanie Hubbard [DMU 1986-2005, Senior Lecturer, Computing Sciences] on two linked research projects (£143k and £45k) for the NHS Sickle Cell and Thalassaemia Screening Programme. The large empirical study (of 5,211 pregnant women across London, Birmingham, Leicestershire and Devon in collaboration with 4 consultant haematologists, 4 laboratory officers, 298 community midwives, 7 sickle cell nurse counsellors and 6 research nurses), investigated the validity, reliability, practicalities and displacement effects of attempting to target antenatal screening for people at risk of carrying genes associated with sickle cell/ thalassaemia by means of an ethnic/family origins question.

A randomized controlled trial of two candidate ethnicity screening questions found that a category-based ethnicity screening question performed better than a binary plus open-ended question. The category-based question was more reliable (i.e. produced the same reply to the ethnic/family origins screening question when asked a second time, several weeks later by a different health professional) and took slightly less time to administer. This provided policy-makers with a measure of the degree of association between social constructs of ethnicity and actual genetic status. The conclusion was that the category-based question was recommended as the basis for the screening question to be adopted by the NHS Screening Programme [4].

The second part of the funded project – qualitative research with mothers and midwives – revealed problems on the ground with targeted screening. Some midwives used intuition to select/exclude clients from the screening questions rather than implement formal policy. The persistence of erroneous beliefs in 'racial' groups displaced correct understandings of the relation between ethnicity and risk of carrying genes associated with sickle cell/thalassaemia. We recommended that continuing professional education of midwives responsible for screening should include education around the concepts of ethnicity and the problems for sickle cell screening that arise if one thinks in terms of false notions of distinct racial categories [3].

In the low prevalence area of our study, use of our evidence-based ethnicity screening question (as opposed to locally devised "common sense" categories previously in use) increased the proportion of clients correctly identified as at risk of carrying genes associated with sickle cell or thalassaemia from 2.2% to 13.0%. Use of our screening question means that guidance can now be given to the 10.8% of women who were previously being denied his advice. Our research also found that, even where midwives were correctly identifying mothers as at-risk by the ethnicity-screening questions, only ten per cent of mothers were actually being offered a laboratory screen. In order to minimise the rates of failing to offer laboratory screening, it was pointed out that midwives require specific instructions on the screening question as to which risk groups to offer a laboratory screen [6], and this was a feature of the screening question adopted by the NHS

Screening Programme and used in practice from 2005 to date.

A major output of the research was the proposal of an ethnic/family origins screening questionnaire for use by the NHS Screening Programme [Appendix of reference 3]. With modifications made by the NHS Screening Programme itself, our screening question became adopted by the programme as the Family Origins Questionnaire (FOQ). The screening programme gradually brought all NHS Trusts into line with national screening policies between 2005-8 until all NHS Trusts in England were implementing screening using the FOQ by 2008.The National Institute for Clinical Excellence (NICE, 2008) drew upon our research in producing evidence-based guidelines for the implementation of antenatal screening in England.
Notes on non-DMU Contributors: David Rees (Consultant in Haematology, Kings College Hospital); Cynthia Gill (Sickle cell nurse counsellor, freelance consultant); Ann Kennefick (Research Nurse, Birmingham); Patsy Morris (Research Midwife, Kings College); Faye Sutton (Research Midwife, Exeter and Devon); Patricia Squire (Research Midwife, University Hospitals of Leicester); Keith Chambers (Senior Medical Scientific Laboratory Officer, University Hospitals of Leicester); Sue Gawler (Senior Medical Scientific Laboratory Officer, Exeter and Devon NHS Trust); Vanita Jivanji (Sickle Cell Nurse Counsellor, Leicester City PCT).

3. References to the research

Bold = DMU staff at time of the research *[Quality: all peer reviewed journals]*

[1]. **Dyson, SM**. (1998) 'Race', ethnicity and haemoglobin disorders. *Social Science and Medicine* 47 (1): 121-131. [ISSN: 0277-9536]

[2]. **Dyson, SM.** (1999) Genetic screening and ethnic minorities. *Critical Social Policy* 19 (2) 195-215. [ISSN 0261-0183]

[3]. **Dyson, SM** (2005) *Ethnicity and Screening for Sickle Cell/Thalassaemia* Oxford: Elsevier Churchill Livingstone [ISBN: 0-443-10232-5] 216 pages. *[Quality: Nominated for the Sociology of Health and Illness Book of the Year 2006 by Professor Gillian Hundt of University of Warwick]*

[4]. **Dyson, SM**; **Culley, LA**; Gill, C; **Hubbard, S**; Kennefick, A; Morris, P; Rees, D; Sutton, F; Squire, P (2006) Ethnicity Questions and Antenatal Screening for Sickle Cell/Thalassaemia [EQUANS] in England: A randomized controlled trial of two questionnaires. *Ethnicity and Health* 11 (2): 169-189. [ISSN 1355-7858]

[5]. **Dyson, SM**; Chambers, K; Gawler, S; **Hubbard, S**; Jivanji, V; Sutton, F; and Squire, P (2007) Lessons for Intermediate and Low Prevalence Areas in England from the Ethnicity Questions and Antenatal Screening for Sickle Cell/Thalassaemia [EQUANS] Study. *Diversity in Health and Social Care* 4 (2): 123-35. [ISSN 1743-1913]

4. Details of the impact

References refer to those listed in section 3

The research findings were presented to a wide range of stakeholder audiences, including the Department of Health, the NHS Screening Programme, the Sickle Cell Society, the UK Thalassaemia Society, NHS Scotland, the Royal College of Midwives, the UK Forum on Haemoglobin Disorders, the British Sociological Association (MedSoc), ESRC research seminar series, Sickle Cell and Thalassaemia Association of Nurse Counsellors, and the London Genetic Knowledge Park, and abroad to the Sickle Cell Disease Association of America, the Anemia Institute (Canada), and the Global Alliance for Nursing Education and Scholarship. The report to the NHS Sickle Cell and Thalassaemia Screening Programme proposed an evidence-based ethnic/family origins screening question [3], providing policy-relevant evidence of the validity and reliability of such questions [4]; the practicalities of implementing selective screening in busy practice [3] and the particular lessons for low prevalence areas [5]. It also raised the issue of negative reactions of ethnic majorities to screening results [3]. From 2005 onwards half of NHS Trusts in England (areas designated of low prevalence for sickle cell/thalassaemia) began to use the Family Origins Questionnaire (FOQ) to identify which women to offer a full laboratory screen for sickle cell/thalassaemia. From 2009 onwards, the remaining NHS Trusts in England (areas designated of high

prevalence for sickle cell/thalassaemia) also began to use the FOQ because (as pointed out in our research) ethnic/family origins helps laboratories target which laboratory tests to apply, especially in identifying potential cases of severe alpha-thalassaemia.

The *Family Origins Questionnaire* used by the NHS Screening Programme includes several specific features taken directly from our research, including:

- An evidence-supported, category-based question was chosen, not an open-ended one [3, 4], nor one based on locally devised, common sense racialised categories [1, 5].
- The categories used in the question departed from Census categories to amplify those ethnic groups at greater risk of sickle cell/thalassaemia [2].
- The fact that an "ethnic/family origins" screening would have utility in high prevalence areas (for assessing those at risk of severe alpha-thalassaemia) as well as in low prevalence areas [4].
- The screening programme commissioned an "ethnicity" screening question, but our research suggested an "ethnic/family origins" question and the policy-makers eventually picked up on and used the phrase "family origins". [4]
- The ethnicity screening question developed encouraged those of "mixed" heritage ethnicity to tick *any* combinations not just the restricted range of mixed options in the 2001 Census[4].
- The fact that midwives required prompts as to which ethnic categories constituted higher risk groups to whom an offer of a screen should be made [5].
- The fact that, if a woman ticked certain boxes, instructions for the midwife on what to do in terms of bottling, labelling and sending off blood for testing were required [5].

The **reach of the impact** is possible to quantify. All pregnant women in England (approximately 3 million between January 2008-July 2013) will have been administered the family origins questionnaire (the development of our ethnic/family origins screening question). Although the screening question was initially (2005-2009) used in only 50% of England (low prevalence areas) to identify pregnant women at higher risk of carrying genes associated with sickle cell/thalassaemia for the purposes of targeting an offer to screen in the laboratory for sickle cell, from 2009 onwards the questionnaire was also used in the rest of England designated as high prevalence areas (where all women are offered a laboratory test to screen for sickle cell/thalassaemia). This is because, as part of the research programme recommendations, it was pointed out by us that all screening laboratories made use of the ethnicity information to further target laboratory tests. For example, there are thousands of different types of mutations underlying thalassaemia, and if the laboratory knows a woman's ancestral origins they can more quickly and efficiently confirm thalassaemia by targeting their laboratory investigations to those genetic variations found more commonly in the woman's particular ethnic group. The programme therefore followed our specific recommendation that the same screening question would be appropriate in both low prevalence areas (for targeting whom to offer an initial laboratory screen) and in high prevalence areas (for targeting further laboratory tests).

The Family Origins Questionnaire as an ethnicity screening tool was part of the 2008 National Institute for Clinical Excellence (NICE) guidelines for antenatal care of women. These NICE guidelines cite the quality of the evidence provided by our reference 4 as Evidence Level 1+, the top level of evidence. The NICE guidelines summarize the evidence underpinning policy deriving from our research as "A fixed response question for screening for family origins is supported by findings from an RCT as being a useful screening test". (NICE, 2008: 132) and cite their Guideline Development Group interpretation of this evidence as "Screening for family origins using a fixed response tick box question is effective in identifying pregnant mothers at risk of haemoglobinopathy (i.e. sickle cell/thalassaemia). A validated family origin questionnaire has been developed for use (NHS Antenatal and Newborn Screening Programme). This is in line with National Screening Committee policy". (NICE, 2008: 132)

In summary, since 2009 all pregnant women in England (approximately 670,000 a year) are offered the Family Origins Questionnaire at their first antenatal appointment with their midwife as part of NHS Screening Programme policy. Eight key features of the design of this question derive directly from our research.

5. Sources to corroborate the impact

Annual reports from The NHS Sickle Cell and Thalassaemia Screening Programme.

The first Annual report to explicitly mention Dyson's research into a Family Origin Questionnaire is

the Annual Report 2003-4 (page 11). The combined reports from 2004-2007 state that "Based on the combined findings, the Family Origin Questionnaire was developed further and rolled out across England" (page 8). The report from 2007-8 lists the Family Origin Questionnaire under the development of laboratory services (page 15). Finally, the report from 2008-9 quotes Dr John Sentamu, Archbishop of York and Chair of the NHS Sickle Cell and Thalassaemia Programme Steering Group, who states: "This was the year in which we finally achieved our goal of rolling out antenatal screening across England. Today, every pregnant woman and every newborn baby in England is offered screening as part of mainstream maternity care."
http://sct.screening.nhs.uk/annualreport [accessed 12th August 2013]

National Institute for Clinical Excellence (NICE)
The NICE guidelines for Antenatal care are Antenatal care (CG62), which can be accessed from http://www.nice.org.uk/CG62 [accessed 12th August 2013]. This guidance was issued in March 2008, replacing guidance from 2003, and covers the routine care that all healthy women can expect to receive during their pregnancy. The new and updated recommendations are marked '**New**'.
Guideline 1.6.3 relates to Screening for haemoglobinopathies:
1.6.3.1 New Pre-conception counselling (supportive listening, advice-giving and information) and carrier testing should be available to all women who are identified as being at higher risk of haemoglobinopathies, using the Family Origin Questionnaire from the NHS Antenatal and Newborn Screening Programme.
1.6.3.2 New Information about screening for sickle cell diseases and thalassaemias, including carrier status and the implications of these, should be given to pregnant women at the first contact with a healthcare professional. Refer to 1.1.1 for more information about giving antenatal information.
1.6.3.3 New Screening for sickle cell diseases and thalassaemias should be offered to all women as early as possible in pregnancy (ideally by 10 weeks). The type of screening depends upon the prevalence and can be carried out in either primary or secondary care.
1.6.3.4 New Where prevalence of sickle cell disease is high (fetal prevalence above 1.5 cases per 10,000 pregnancies), laboratory screening (preferably high-performance liquid chromatography) should be offered to all pregnant women to identify carriers of sickle cell disease and/or thalassaemia.
1.6.3.5 New Where prevalence of sickle cell disease is low (fetal prevalence 1.5 cases per 10,000 pregnancies or below), all pregnant women should be offered screening for haemoglobinopathies using the Family Origin Questionnaire.

- If the Family Origin Questionnaire indicates a high risk of sickle cell disorders, laboratory screening (preferably high-performance liquid chromatography) should be offered.
- If the mean corpuscular haemoglobin is below 27 picograms, laboratory screening (preferably high-performance liquid chromatography) should be offered.

In March 2008, the National Collaborating Centre for Women's and Children's Health were funded to produce a Clinical Guideline for the NHS by NICE into Antenatal care: routine care for the healthy pregnant woman. This (more detailed) guidance also cites Dyson's work (See p 131, Section 8.3.5 paragraphs 1 and paragraph 4, and p132, lines 18-20 and lines 42-5). A hard copy is available upon request. http://www.nice.org.uk/nicemedia/live/11947/40145/40145.pdf [accessed 29th May 2013]

The **NHS Sickle Cell and Thalassaemia Screening Programme** became part of Public Health England on the 1st April 2013. They provide a comprehensive website for practitioners about how to use the Family Origins Questionnaire as an Ethnicity Screening Tool. From this site, all the necessary tools and guidance etc. necessary to undertake the questionnaire can be obtained.
http://sct.screening.nhs.uk/cms.php?folder=2506 [accessed 29th May 2013]