

<b>Institution:</b> University of Sheffield		
<b>Unit of Assessment:</b> C-21 Sociology		
<b>Title of case study:</b> Developing appropriate policy and practice in response to addressing the genetic risk associated with customary consanguineous marriage		
<b>Period when the underpinning research was undertaken:</b> Jan 2013–2019		
<b>Details of staff conducting the underpinning research from the submitting unit:</b>		
<b>Name(s):</b>	<b>Role(s) (e.g. job title):</b>	<b>Period(s) employed by submitting HEI:</b>
Sarah Salway	Professorial Research Fellow	Jan 2013–present
<b>Period when the claimed impact occurred:</b> 2014–2020		
<b>Is this case study continued from a case study submitted in 2014?</b> N		
<p><b>1. Summary of the impact</b> (indicative maximum 100 words)</p> <p>Though repeatedly acknowledged as a public health issue, local and national policymakers have struggled to respond to the increased genetic risk associated with close relative (consanguineous) marriage among some UK ethnic minority populations. Salway's research has impacted on health policy, practice, and investment across the UK. Salway's innovative and influential programme of research including her four-stranded service model, has reframed and increased understanding amongst professionals, building their confidence to act. Salway's research has challenged and reframed understanding amongst policymakers and public health practitioners, leading to changes in practice at the national, regional, and local levels across the UK. The research has improved community understanding of the impacts of consanguineous marriage, with more than 1,000 individuals in Sheffield alone benefiting from face-to-face conversations with public health officials. Online materials based on Salway's research reached 4,594 users with learning resources being adapted for the NHS e-Learning for Health.</p>		
<p><b>2. Underpinning research</b> (indicative maximum 500 words)</p> <p>Close relative (consanguineous) marriage can result in increased risk of genetic morbidity, particularly recessive conditions including severe metabolic and neurological disorders. Despite being recognised as a public health issue that has significant impact on families and services, local and national policymakers have struggled to respond to the issue. Salway's innovative and influential programme of sociologically-informed research has improved understanding of the social and political factors that shape risk patterns, service access and experiences, and policy responses. The co-designed research with practitioners, policymakers, community organisations, and local people, has developed, evaluated, and supported the implementation of culturally appropriate and effective responses.</p> <p><b>Research findings 1: unmet demand for genetic literacy and services</b></p> <p>Between 2013 and 2016 Salway conducted a series of participatory research projects examining understanding and experiences among community members affected by this health issue. Findings demonstrated an appetite for better knowledge; low awareness of services; poor understanding of risk; previously inaccurate messages from healthcare professionals; suspicion of motives behind interventions; importance of lay, as well as professional, sources of</p>		

information; and the dominance of word-of-mouth messaging as the preferred method of communication. Findings showed that local people wanted to co-design genetic literacy approaches, and that materials should include narrative, visual methods; non-stigmatising content; facts and figures from credible sources; acknowledgement of moral and religious concerns; and encouragement to access genetic services (unfamiliar and frightening to many). Evaluation findings confirmed public acceptability of developed materials and the importance of opportunities for face-to-face discussion to clarify complex information [R1, R2].

### **Research findings 2: partial professional understandings and patchy local responses**

Salway's formative evaluation examined local services and professional understandings of consanguineous marriage and approaches to address genetic risk around England. Findings showed variable responses, mixed success, and some alienation of affected communities. The predominant focus from public health services was on educating community members. Services did not include more holistic approaches that aimed to also equip healthcare professionals and increase access to genetic services for affected families. Unrealistic policy expectations of short-term reductions in infant mortality, and problematic 'invest to save' justifications for service developments were highlighted. These resulted in activity not being sustained. Inadequate local approaches reflected the wider commissioning system that struggles to accommodate diverse needs and questions ethnic minority healthcare entitlement. The research provided important insights into inappropriate policy and practice approaches, obstacles to progress, as well as positive elements to be replicated [R3, R4].

### **Research findings 3: opportunity to develop a national policy framework**

In 2018, Salway conducted a study with healthcare professionals and the public to explore their views on how to respond to this health need. She employed the Delphi method to build consensus in expert opinion in an iterative and structured way over three rounds. Following this consensus conference discussions resulted in a final set of 148 agreed statements, providing direction for both policymakers and healthcare professionals. These included principles of approach and specific expectations for services. The study produced important new insights into how a national response could be crafted. It highlighted the importance of responding to health needs that disproportionately affect ethnic minority groups without racialisation. Findings helped to frame the issue as a problem of fundamental inequitable access to healthcare. From this Salway developed a 'four-stranded' service model incorporating the four priorities: enhancements to clinical genetic services; training for healthcare professionals; increasing genetic literacy within communities; and equitable access to genetic technologies [R5, R6].

### **3. References to the research** (indicative maximum of six references)

University of Sheffield researchers in **bold**

- R1. Ali, P. A., Salway, S., Such, E.,** Dearden, A., & Willox, M. (2018). Enhancing health literacy through co-design: development of culturally appropriate materials on genetic risk and customary consanguineous marriage. *Primary Health Care Research & Development*, 20, e2. <https://doi.org/10.1017/s1463423618000038>
- R2. Salway, S., Ali, P., & Ahmed, S.** (2014). Towards enhanced community genetic literacy among a minority ethnic community: a participatory action research project. *The Lancet*, 384, S7. [https://doi.org/10.1016/s0140-6736\(14\)62133-4](https://doi.org/10.1016/s0140-6736(14)62133-4)

- R3. Salway, S., Ali, P., Ratcliffe, G., Such, E., Khan, N., Kingston, H., & Quarrell, O. (2016).** Responding to the increased genetic risk associated with customary consanguineous marriage among minority ethnic populations: lessons from local innovations in England. *Journal of Community Genetics*, 7(3), 215–228. <https://doi.org/10.1007/s12687-016-0269-1>
- R4. Salway, S., Mir, G., Turner, D., Ellison, G. T. H., Carter, L., & Gerrish, K. (2016).** Obstacles to “race equality” in the English National Health Service: Insights from the healthcare commissioning arena. *Social Science & Medicine*, 152, 102–110. <https://doi.org/10.1016/j.socscimed.2016.01.031>
- R5. Salway, S., Yazici, E., Khan, N., Ali, P., Elmslie, F., Thompson, J., & Qureshi, N. (2019).** How should health policy and practice respond to the increased genetic risk associated with close relative marriage? results of a UK Delphi consensus building exercise. *BMJ Open*, 9(7), e028928. <https://doi.org/10.1136/bmjopen-2019-028928>
- R6. Salway, S. (2018).** Working ‘with and against’ ethnic categories to understand and tackle health inequalities: illustrations from research and practice relating to infant mortality in England. *European Journal of Public Health*, 28(S1), cky047.036. <https://doi.org/10.1093/eurpub/cky047.036>

#### 4. Details of the impact (indicative maximum 750 words)

##### **Challenging and reframing understandings among practitioners and decision makers at national and local levels**

Salway drew on research findings [R1- R4] in oral and written policy briefings to policymakers and healthcare professionals to effectively improve knowledge and reframe understanding away from a focus on marriage practices towards enhancing informed choice and service equity.

In 2018, Salway established a national Steering Group, with Public Health England, NHS England/Improvement (NHS/I), Department of Health and Social Care (DHSC), plus eight professional bodies. This was ‘*completely new and desperately needed*’ [GP with Special Interest (GPwSI) in Genetics and Primary Care Adviser, HEE Genomics Programme, S1]. Salway used the Steering Group to issue policy briefings to key professional networks (maternity, general practice, genomics). Salway was subsequently asked to present at the Maternity Transformation Programme (MTP) Inequalities Roundtable chaired by Chief Midwife, Jacqueline Dunkley-Bent in November 2019. Salway’s research is credited with “*challenging earlier approaches*” [Principal Genetic Counsellor, S1] and generating new understanding of need and appropriate action [GPwSI in Genetics and Primary Care Adviser, HEE Genomics Programme, S1].

Learning events across England (September 2013-2020) drew on Salway’s research findings [R3, R4] to inform course content. Over 470 healthcare professionals representing, amongst others, public health, midwifery, clinical genetics, and health visiting attended these events, which were held in every ‘high need’ area in England [Learning event feedback, S2]. Event evaluations demonstrate impact on knowledge and awareness. Events were “*thought provoking, helping to shift attitudes around the issue and the role of professionals in addressing unmet need*” [Director of Public Health, Rotherham, S3] and delivered important learning [S2]. Online materials were promoted reaching 4,594 users and learning resources are being adapted for the

NHS e-Learning for Health *“attesting to their recognised importance and quality”* [GPwSI in Genetics and Primary Care Adviser, HEE Genomics Programme, S1].

Locally, Salway established and chaired the Sheffield multi-agency community genetics group 2013-2016 and remains a member, enabling sustained research-informed dialogue. The research *“helped to significantly raise awareness and shift understanding of the issue among key professional stakeholders”* [Director of Public Health, Sheffield, S3]. In 2019, Salway gave *“invaluable”* guidance to a BBC Newsnight feature shaping the first mainstream media coverage to highlight *“the important issue of improving access to genetic services”* [Producer, S4]. In Birmingham, this coverage created *“a more receptive atmosphere”* amongst local politicians [Interim Assistant Director of Public Health, S4], prompting a presentation to the council’s Health Overview and Scrutiny Committee. Salway has briefed public health officials working in communities with England’s highest rates of consanguinity: Rotherham, Leicester, Bradford, West Midlands, and several London boroughs.

### **Directly influencing new policy, practice, and investment**

In 2019, the national Steering Group was formally incorporated into the MTP Reducing Inequalities work plan. In 2020, Salway’s findings [R5, R6] *“directly shaped the development of the first national policy proposal in this area”* [Consultant Geneticist, S5]. Salway worked with clinical stakeholders to make a case for government funding. Her work was used to design a proposed new model to incorporate national, regional, and place-based solutions. *“This represents a significant new national policy development since all prior action has been locally led and often not sustained”* [Consultant Geneticist, S5]. The submitted proposal includes multi-million-pound investment which would flow to eight areas of highest need and includes newly developed specialist posts. This constitutes a significant policy innovation. *“Reducing genetic risk amongst ‘at risk’ populations is necessary to help achieve the ‘halve it’ aim and to reduce health inequalities [...] Therefore, the work of Professor Salway and others aligns closely to the MTP’s strategic aims. The work is valuable because it identifies not only the issues but also clear, practical, evidence-based solutions [...] The quality of the research enabled us to gain support for the proposal from senior colleagues, both internally and externally”* [Programme Manager MTP, S6].

Salway’s research has been instrumental in informing the development of the Clinical Genomics Service specification through the incorporation of explicit attention to unmet ethnic minority needs. Without Salway’s work the Principal Genetic Counsellor, Manchester, states *“the progress that has been made at local and national level...would not have been possible”* [S1]. Salway was an invited expert in the West Yorkshire and Harrogate Local Maternity System Prevention Workstream (2020). Her Delphi research [R5] directly informed the development of the expert group recommendations and is now shaping action to enhance service access and increased genetic literacy at the community level. *“The recommendations are now guiding local action, particularly in Bradford and Kirklees, two areas with high numbers of families at risk and in need of enhanced services. Further, Sarah has supported both these areas via the adaptation of evidence-based genetic literacy materials”* [Programme Manager for Improving Population Health & Public Health Lead for Maternity, S7].

At a local level Salway’s research [R1-R4] directly shaped Sheffield’s new response to the issue. This included the creation of a novel Community Genetics Outreach Worker post and use of co-produced genetic literacy materials at community and health professional levels [Community Outreach Worker, S8; Director of Public Health Sheffield, S3].

Seven of the Local Authorities with the highest need have adopted the genetic literacy materials to support genetic literacy strategies [Programme Manager for Improving Population Health & Public Health Lead for Maternity, S7; S9].

#### **Enhanced information and service access**

Enhancing informed choice among even small numbers of individuals has huge benefits at family and health service level due to the high social, emotional, and financial costs associated with severe conditions. Families raising a disabled child face extreme economic pressure; it is estimated that it is three times more expensive to raise a child with disabilities than without. The emotional and social impact of unanticipated affected births can also be significant for parents and wider families. Therefore, access to accurate, sensitive, and trusted sources of information is essential. Salway's research has impacted over 1,000 people in Sheffield via face-to-face conversations amongst practitioners and community members and through the 5,000 distributed genetic literacy leaflets [Director of Public Health Sheffield, S3]. The genetic literacy materials have been "*extremely important in engaging community members and helping them to understand the patterns of risk and gain confidence to discuss the issue*" [Community Outreach Worker, S8]. Case studies developed by Sheffield City Council, documenting their outreach work, demonstrated that families were better informed and better supported to access genetic services [Director of Public Health Sheffield, S3].

Additionally, health and social care costs can be very high due to multiple hospital appointments, repeated hospital stays, care from multiple specialities and community teams, medication, home adaptations, and treatment. For a child with severe learning disabilities alone, the average care package cost per annum is estimated at £70k. Thus, a clear pathway of support and collaborative working amongst health and social care services is vital. Salway's research played a crucial role in developing more joined-up working [Programme Manager MTP, S6] and creating and sustaining this work [Director of Public Health, S3; Community Outreach Worker, S8].

#### **5. Sources to corroborate the impact** (indicative maximum of 10 references)

- S1.** Testimonial Evidence from senior genetic service professionals.
- S2.** Learning event feedback September 2013-October 2020.
- S3.** Testimonials from local directors of public health.
- S4.** Impact stemming from Newsnight feature including testimonial from the interim Assistant Director of Public Health, Birmingham.
- S5.** Testimonial from consultant geneticist and ex-chair of the National Clinical Reference Group for Clinical Genetics.
- S6.** Evidence related to the development of national proposal for new service investments including testimonial from the Maternity Transformation Programme Manager.
- S7.** West Yorkshire and Harrogate Local Maternity System Public Health Recommendations July 2020 (Salway's research referred to on p.25) and testimonial from programme manager.
- S8.** Testimonials from patient and community perspectives.
- S9.** Evidence showing the genetic material leaflets used by seven local authorities.