Institution: Plymouth University

Unit of Assessment: A3

Title of case study: Genetic counselling: developing European wide standards of professional competence

1. Summary of the impact (indicative maximum 100 words)

This case study demonstrates the impact of research focussed on establishing a set of European core competences in genetics for health professionals. Prior to the research, there were clear inequities and inconsistency of practice regarding genetic health services for patients across Europe, in particular in the fields of genetic counselling and genetic nursing. The core competences for genetic nurses and counsellors are now being used as the foundation for genetic nursing and genetic counselling profession in Europe, to improve standards of patient care. Standards of practice and education, curricula for Master's degree training programmes and a formal European registration process for genetic counsellors and nurses are all based on the outcomes of the initial study at Plymouth. These measures have resulted in professionalization of genetic nursing and genetic counselling in the European context and consequently improved consistency of care.

2. Underpinning research (indicative maximum 500 words)

The study to develop a set of core competences in genetics for health professionals was led by Professor Heather Skirton at Plymouth University (Reader in Applied Health Genetics, 2004-2008, Plymouth University; Professor of Applied Health Genetics 2008-present, Plymouth University) and European colleagues as part of the (Framework 7) EuroGentest project to standardise and harmonise genetic testing in Europe. The initial work was undertaken between 2007- 2010.

Research indicated that individuals in different European countries were not being treated equitably regarding genetic testing, nor were they having access to appropriate genetic counselling to support informed decision making and adjustment to test results (Mehta, 2005; Cordier et al, 2010; Skirton et al, 2013). In some cases, patients were being asked to make decisions about genetic tests (including prenatal testing of the fetus) that could affect the life of the entire family, without discussion with a trained professional. In other cases, they received genetic test results without any explanation of the implications or options for future management. Many of those offering 'genetic counselling' were not trained in communication skills. Setting minimum standards of professional care was integral to harmonising genetic testing and ensuring greater equity for patients affected by or at risk of genetic conditions.

Establishing a set of agreed professional competences was adopted as both a theoretically sound and practical way to establish minimum standards of care across countries with differing health and educational systems; the aim of the study was therefore to systematically develop a set of genetic competences for European health professionals. As part of a team comprising Coviello (Milan, Italy) and Lewis and Kent (from the patient group Genetic Alliance UK), Skirton led the work between 2005 and 2009 to develop a set of core competences that could be applied to health professionals in primary, secondary and tertiary settings in all European countries.

Initially a systematic literature search was undertaken to identify peer-reviewed and grey literature on professional education and/or competence in genetics (Coviello et al, 2007). This was followed by a Delphi study conducted in five stages to ascertain the views and opinions of experts in the field of genetics and genomics from a wide range of European countries. In 2005, an initial two-day workshop was organised with 30 participants from professional and service user groups to encourage production of ideas and exchange of views on relevant competences. Following this, the draft competences were circulated to a larger group of experts for further comments. In the third stage, another workshop attended by 55 participants from 16 European countries was held in 2006 to refine the competences and develop corresponding learning outcomes for each. The resulting competences were submitted for scrutiny by members of the Board of the European Society of Human Genetics (ESHG) and subsequently all members of that organisation, before

being endorsed by the ESHG Education Committee in 2008.
In the fifth and final stage, the competence framework, comprising competences and learning outcomes in genetics for each professional group were presented to another set of key informants, representatives of 43 national genetics professional societies from 37 European countries, for feedback. The members of these societies are all professionals who would be directly involved in the standards, training and assessment of the competence of health professionals. The study report documents (1. Executive Summary, 2. Background, 3. Core competences for genetic specialists and 4. Core competences for primary care professionals and non-genetic specialists) were then published on the websites of the ESHG and the EuroGentest project. A peer-reviewed publication summarising the work was published (Skirton et al, 2010).

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

Three of the relevant papers were published in the *European Journal of Human Genetics, which* is the official journal of the European Society of Human Genetics, with an IF of 4.4, ranking 30/157 journals on genetics and heredity. It has an international editorial board and serves the entire academic, clinical and research communities in the field of human genetics in Europe.

1. Coviello DA, **Skirton H**, Ceratto N, Lewis C, Kent A. (2007) Genetic testing and counselling in Europe: health professionals current educational provision, needs assessment and potential strategies for the future. *European Journal of Human Genetics* 15: 1203 – 1204.
Original report of the literature review.

2. **Skirton H**, Lewis C, Kent A, Coviello D. (2010) Genetic education and the challenge of genomic medicine: development of core competences to support preparation of health professionals in Europe. *European Journal of Human Genetics* 18(9):972-7. Available at:
http://www.nature.com/ejhg/journal/v18/n9/pdf/ejhg201064a.pdf
Paper reporting the method and results of the core competences framework

3. **Skirton H**, Barnoy S, van Kessel I, Patch C, O'Connor A, Serra C, Stayner B, Voelckel MA. (2013) A Delphi study to determine the European core curriculum for Master programmes in genetic counselling. *European Journal of Human Genetics* DOI:10.1038/ejhg.2012.302
This paper demonstrates that the core curriculum was mapped against the core competences to ensure educational programmes were satisfactory in preparing competent practitioners.

4. **Skirton H**, Voelckel MA, Patch C. (2010) Using a community of practice to develop standards of practice and education for genetic counsellors in Europe. *Journal of Community Genetics* 1: 169-173. Paper reporting the further development of standards, based on core competences.

5. **Skirton H**, Lewis C, Kent A, Ceratto N, Coviello D. (2007) EuroGentest Unit 6: Patient and Professional Perspectives of Genetic Information/Education in Europe. Unit 6.2. Professional Perspective. Core competences in genetics for health professionals in Europe. Background document: development of a set of core competences in genetics for health professionals. Available at:
https://www.eshg.org/fileadmin/www.eshg.org/documents/CoreCompetence02Background.pdf

6. **Skirton H**, Lewis C, Kent A, Coviello D. (2007) EuroGentest Unit 6: Patient and Professional Perspectives of Genetic Information/Education in Europe. Unit 6.2. Professional Perspective. Core competences in genetics for health professionals in Europe. Core competences for health professionals who are specialists in genetics. Available at:
http://www.eurogentest.org/fileadmin/templates/eugt/pdf/CoreCompetence04GeneticSpecialists.pdf

4. Details of the impact (indicative maximum 750 words)
The impact of the research onto the core competences for genetic counsellors and specialist genetic nurses led directly to impact on professional standards and on guidelines and training throughout Europe, resulting in greater consistency in the quality of genetic healthcare services for

patients.

Genetic counselling involves providing support and information giving for patients who are affected by or at risk of a genetic condition. At the time this project began, provision of genetic counselling in Europe was patchy. In the majority of countries it was provided by a range of professionals, many of whom had no specific educational preparation or assessed competence for the role. This had a potential impact on standards of patient care. The status of genetic nurses and genetic counsellors was ill-defined, due to lack of professional regulation; in some cases this resulted in a reluctance to utilise these professionals within health services (Skirton, Cordier et al, 2013). However, with increasing burden on genetic health services, there was pressure to recruit appropriately trained practitioners to enable patients to make informed decisions about genetic tests, to help them to understand the results and the implications of those results, and to support them to make decisions about future management of the condition. Therefore, to achieve safe care for patients, appropriate education and a system of assessing competence of practitioners was needed.

A national registration system for genetic counsellors and nurses exists in the UK (Skirton et al, 2003), but no similar formalised system exists in other European countries. However, due to low populations and practitioner numbers, it is unfeasible to introduce registration systems in every country. The European Society of Human Genetics (ESHG) recognised the need for a concerted approach to genetic nurse and counsellor accreditation and requested that Skirton lead a new Ad Hoc Committee in 2010 [5] . This was subsumed in 2012 into the European Board of Medical Genetics (EBMG), of which Skirton is now the inaugural Chair [https://www.eshg.org/ebmg.0.html]. The Board is responsible for setting up systems of accreditation for genetic specialists: genetic counsellors, genetic nurses, medical geneticists and laboratory scientists.

The focus of this case study, the research to establish agreed core competences in genetics for genetic nurses and counsellors, has been used to underpin the establishment of genetic counselling and genetic nursing as recognised professions in Europe. Initially, the work had an impact on professional standards, as the competences were used as the basis for the professional standards of practice and education for the genetic counselling profession in Europe (Professional and Educational Standards for Genetic Counsellors, 2010) [Section 5, Item1]. These standards and a Code of Practice for genetic counsellors were prepared by a Working Group of experts in 2010 and approved by 156 members of the European network of genetic nurses and counsellors (Skirton et al, 2010) [Section 3, item 4]. Subsequently they had an impact on education in several ways. The core competences were used by a number of European universities to guide the curriculum of profession specific courses. The curriculum of the Master degree programme in genetic counselling at Porto University] was based entirely on the core competences (evidence available in course documentation) [Section 5, items 2 and 3]. They were used directly to inform new Master's degree programmes in genetic counselling established in Hungary, Greece, Romania and Sweden (evidenced by correspondence with Skirton) [Section 5, Item 4]. Professor Bela Melegh, Chairman of the Department of Medical Genetics at Pécs University, Hungary, wrote' …there has been a significant progress in the development of the new MSc course curriculum …special thanks for providing the core competences for genetic counsellors'. In a separate educational initiative, the research was used by the EBMG to define a core European curriculum for Master's degree in genetic counselling and genetic nursing. The President of the ESHG acknowledged the importance of this work to developing the profession, writing 'our next aim is to achieve European-level recognition for genetic counsellors/nurses …. the Ad-hoc Committee for Certification of Genetic Nurses/Counsellors …. have gone deeply into developing core curricula as a basis for European-level recognition' [Section 5, item 5]. The core curriculum has now been used by the EBMG as the standard for assessing suitability of European master's degree programmes to train genetic counsellors and genetic nurses, prior to them being awarded registration [Section 5, Item 6]. The impact of the work on professional registration and recognition is evidenced by the establisghment of a formal European registration system for genetic nurses and genetic counsellors, which was launched in June, 2013. Registration is now open to those practitioners who can demonstrate their competence, and all practitioners must demonstrate (through submission of a portfolio) that they fulfil each of the core competences relevant to the profession before being awarded registration status [Section 5, Item 7]. Joerg Schmidtke (ESHG Past President) wrote 'Skirton was instrumental in this process…her studies on the core competences were ground- breaking.. for the

development of the profession of genetic counsellors in Europe' [Section 5, Item 8].

As demonstrated above, the establishment of European core competences for genetic nurses and genetic counsellors has provided a foundation for the construction of a stronger, safer profession. Standards of practice, the Code of Practice, educational curricula and the registration system have all emerged from the original set of competences [Section 5, Items 9 and 10]. It is now possible for genetic nurses and counsellors to demonstrate their competence within a coherent framework across the entire European genetics community, encompassing 43 countries. Although the system is at present voluntary, forty-four genetic nurses and counsellors from 13 countries have indicated they wish to register in the first cohort and the EBMG is seeking to make registration mandatory through the expected EU professional qualifications directive. The impact of this work is ultimately greater consistency and quality of care for patients seeking genetic counselling.

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

1. Evidence that the core competences were an integral part of the standard setting for the profession in Europe: Professional and Educational Standards for Genetic Counsellors in Europe (2010), available at: https://www.eshg.org/fileadmin/www.eshg.org/documents/committees/ProfessionalAndEducationalAtandardsForGeneticCounsellors.pdf

2. Evidence that the core competences have been used to underpin curricula for Master's degree programmes across Europe: Course documentation for Master's degree in genetic counselling, University of Porto (relevant documents available in English from Heather Skirton). http://sigarra.up.pt/icbas/pt/cur_geral.cur_view?pv_curso_id=1287&pv_ano_lectivo=2012&pv_origem=CUR

3. Copy of the formal approval document to the Ministry of Science, Technology and Higher Education applying for establishment of the MSc in Genetic Counselling at University of Porto, which cites the core competences as a basis for the programme and on pages 7 and 8 names Professor Heather Skirton from Plymouth University as the expert contact. https://www.researchgate.net/project/Core_Competences/bench/Core_competences/?ev=proj_det_bnch_addf).

4. Letters from Course Director of European master course, confirming the use of the core competences as a basis for the Master's courses in genetic counselling.

5. Copy of the President's report in the European Society of Human Genetics Newsletter, which names Skirton and emphasises the importance of the core curriculum to development of the genetic counselling profession in Europe. https://www.eshg.org/fileadmin/eshg/newsletter/Newsletter_No21_2012.pdf (p2)

6. Evidence that the core curriculum has been used to assess European Master degree courses to determine their fitness as training programmes for genetic counsellors: European Board of Medical Genetics website [https://www.eshg.org/408.0.html]

7. Evidence of the European Registration Process, based on the core competences: European registration process for genetic nurses and counsellors. All details posted on EBMG website, hosted by ESHG at https://www.eshg.org/471.0.html

8. Letter from Past President of the ESHG on the importance of establishing the standards for European genetic counsellors and the relevance of the core competences and core curricula developed by Skirton to this work.

9. Evidence of the use of the core competences as a foundation for the educational recommendations and registration process: National Human Genetics Societies Meetings. Minutes and agendas for past six years, available at: https://www.eshg.org/nhgs.0.html (e.g. 2007 minutes, page 32011 minutes, Item 9).

10. Report of the Committee Chairs of the Ad Hoc Genetic Nurse and Counsellor Accreditation Board. Available at : https://www.eshg.org/fileadmin/www.eshg.org/documents/committees/ESHGGNCComMay2010Report.pdf