**Institution:** University of Aberdeen

**Unit of Assessment:** UoA1: Clinical Medicine

**Title of case study:** Transforming support for young people in families with Huntington's Disease

**Period when the underpinning research was undertaken:** 2003-2019

**Details of staff conducting the underpinning research from the submitting unit:**

| Name(s): | Role(s) (e.g. job title): | Period(s) employed by submitting HEI: |
|---|---|---|
| Karen Forest-Keenan | Research Fellow | 1999-2004; 2010-present |
| Zosia Miedzybrodzka | Head of Medical Genetics | 1998-present |
| Lorna McKee | Professor of Management | 1995-2016 (Emeritus 2016-2017) |
| Sheila Simpson | Clinical Geneticist (Hon) | 2004-2012 |
| Edwin van Teijlingen | Senior Lecturer Public Health | 1990-2009 |

**Period when the claimed impact occurred:** 2013-ongoing

**Is this case study continued from a case study submitted in 2014?** No

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

Prior to the Aberdeen research being carried out, the devastating impact of growing up in a family with Huntington's disease (HD) upon children and young people was largely unknown. Research by the team at Aberdeen explored different aspects related to the experiences of children and young people in families with HD, including conversations with parents, caring responsibilities and predictive genetic testing, demonstrating the need for specialised, age-appropriate services to provide young people with education and support. Research findings have informed the development of new support services and educational materials for children and young people in Scotland and around the world, as well as influencing the practice and training of healthcare professionals.

**2. Underpinning research** (indicative maximum 500 words)

Huntington's disease (HD) is an incurable hereditary neurodegenerative disorder, affecting approximately 3/100,000 individuals worldwide, and between 1/5000-1/6000 individuals in Scotland. The age of onset is usually between 30 and45 – often coinciding with parenthood – with symptoms including involuntary movements, changes in behaviour and personality, cognitive impairment, and increasingly debilitating symptoms. Because HD is due to an autosomal dominant gene, every child of an affected parent has a 50% risk of developing the illness in their adulthood. For children living in a family with HD, many witness the loss of a parent whilst also finding out that they are at high risk themselves. They may also face considerable stigma associated with the disease, higher rates of family breakdown, and secrecy within affected families about its existence. However, prior to 2001 little was known about children and young people's experiences of HD, and there were no services nor evidence-based educational materials specifically for children or young people impacted by this devastating disease.

The team of researchers at Aberdeen – initially led by Dr Sheila Simpson (Clinical Genetics) and later by Professor Miedzybrodzka (Medical Genetics) – sought to rectify this gap by creating an evidence base in five core areas.

First, they explored the difficulties patients had telling their children about the hereditary aspect of Huntington's disease. The research generated data about actual conversations using in-depth qualitative interviews with affected families in Scotland. The findings revealed that communication within a family about genetic risk is a complex issue and is influenced by both pre-existing familial and cultural factors and individuals' responses to risk information [**R1**].

The team used this knowledge to explore the trauma that children and young people can experience when growing up with an affected parent addressing – for the first time – how living within a family with HD was experienced from the perspective of the young person. Drawing on in-depth qualitative interviews, the research examined different types of disclosure experiences, along with the timing and style of disclosure and the subsequent impact on young people. Findings identified the extent to which living in a family with HD profoundly affects children and young people, evidencing that children worry about their own high risk, undertake caring roles, live with prolonged anxiety, experience multiple losses, and may be in need of child protection. A crucial finding was that young people who grow up knowing about the disease had been empowered to cope better, highlighting the importance of communication and the provision of age-appropriate information [**R2**, **R3**]. Findings have since been replicated by studies in other parts of the world (USA, Australia, Canada and Europe).

HD was one of the first conditions where predictive genetic testing for over 18s became available and as such is a paradigm for wider genetic testing guidelines and evaluation of how testing programmes impact upon individuals and families. The team's research on young people's experiences of predictive testing for HD provided much needed evidence and was the first to examine both prospective and retrospective accounts. The research demonstrated a need for additional support for young people around isolation, grief and loss, the waiting period, and particularly if the result contradicted what they had felt deep down [**R4**].

A further area of interest was related to understanding familial communications, specifically how individuals communicate with their partners about HD and the potential risks they face. Using qualitative interviews with partners of an individual at risk for HD, enabled the research team to identify and examine the efficacy of different disclosure experiences and develop recommendations based on the responses. Findings indicated that there could be significant benefit to the relationship if partners were encouraged to attend at least one separate appointment in pre- test counselling [**R5**].

Finally, the team sought to understand the variation in professional practice of genetic counsellors and clinical geneticists in Scotland shepherding patients through the process of communication with their family members. Through interviews with genetic health professionals, the research team was able to consider the efficacy of the different approaches that were taken – from a limited role to professional intervention, concluding that this remains a challenging and sensitive area, and one in which genetics professionals express a need for more resources and the clinical time to undertake this work [**R6**].

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

The quality of the research is deemed to be at least of 2* quality as corroborated by the following peer-reviewed, international publications (with Google Scholar **citations**) and funding from CSO and Wellcome Trust:

**[R1] Forrest Keenan**, **Simpson SA**, **Wilson BJ**, **Van Teijlingen ER**, **McKee L**, **Haites N**, **Matthews E**. 2003. 'To Tell or Not to Tell: barriers and facilitators in family communication about genetic risk'. Clinical Genetics 64: 317-326. DOI: 10.1034/j.1399-0004.2003.00142.x (**359**)

**[R2] Forrest-Keenan, K, Miedzybrodzka, Z, van Teijlingen, E, McKee, L & Simpson,** SA 2007, 'Young people's experiences of growing up in a family affected by Huntington's disease', *Clinical Genetics*, vol. 71, no. 2, pp. 120-129. DOI: 10.1111/j.1399-0004.2006.00702.x (**92**)(*This is a cornerstone paper which for the first time addressed by in depth interview how living within a family with Huntington's disease was experienced from the perspective of the young people.*)

**[R3] Keenan, K, van Teijlingen, E, McKee, L, Miedzybrodzka, Z & Simpson, SA 2009**, 'How young people find out about their family history of Huntington's disease', *Social Science & Medicine*, vol. 68, no. 10, pp. 1892-1900. DOI: 10.1016/j.socscimed.2009.02.049 (**48**)

**[R4] Keenan, K, McKee, L & Miedzybrodzka, Z** 2015, 'Help or hindrance: young people's experiences of predictive testing for Huntington's disease', *Clinical Genetics*, vol. 87, no. 6, pp. 563-569. DOI: 10.1111/cge.12439 (**17**)

**[R5] Keenan, K, Simpson, SA, Miedzybrodzka, Z**, Alexander, DA & Semper, J 2013, 'How Do Partners Find out About the Risk of Huntington's Disease in Couple Relationships?', *Journal of genetic counseling*, vol. 22, no. 3, pp. 336-344. DOI: 10.1007/s10897-012-9562-2 (**15**)

**[R6] Keenan, KF, McKee, L & Miedzybrodzka,** Z 2020, 'Genetics professionals' experiences of facilitating parent/child communication through the genetic clinic', *Journal of genetic counseling*. DOI: 10.1002/jgc4.1179 (**1**)

**Grants:**
Grant awarded to Van Teijlingen, E., 'To Tell or Not To Tell: Passing on Genetic Knowledge to Family Members' Wellcome Trust; 1999-2001; GBP59,176.

Grant awarded to Simpson SA, Forrest Keenan K, Miedzybrodzka Z, van Teijlingen E, McKee L. 'Growing up at-risk of late-onset familial disease.' Funded by Wellcome Trust Programme in Biomedical Ethics Grant. 2002-2004. GBP81,876.

Postdoctoral Training Fellowship for Dr Karen Keenan in Health Services and Health of the Public Research. 'Sharing information with children and young people about genetic risk: Using evidence to develop services for parents and practitioners.' Funded by Chief Scientist Office, Scotland. 2010-2014. GBP153,105

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

Young people in families living with HD face unique challenges: the burden of being a young carer; social isolation; learning about their own high risk and deciding whether to seek genetic testing. The research by the team at the University of Aberdeen has been instrumental in identifying a need for age-appropriate information and resources about HD, professional and peer support, as well as parental guidance about disclosure to children. The research built an evidence base that has been used to inform support services for young people in Scotland and around the world, and to influence training for health and social care professionals.

**Informing the development of new support services for young people in Scotland**
Early in the research, the Aberdeen team was able to develop a collaboration with the Scottish Huntington's Association (SHA). As well as a crucial avenue for recruiting research participants, the collaboration has informed the development of the SHA youth service (known as SHAYP), a dedicated service providing support to children and young people aged 8-25 in Scotland living in a family with HD. The CEO of SHA highlighted the Aberdeen research as key to SHAYP: "*A key element of the work is to support parents around the disclosure of HD in the family. The practice basis for this work is the research conducted by Keenan et al*" [**S1**]. Since August 2013, SHAYP has supported 163 young people and passed on the study findings to over 300 HD families, with four specialist youth advisors now employed to cover the whole of Scotland. Families attending a genetic clinic with children aged 8-25 in NHS Grampian and other Clinical Genetic Services in Scotland are told about the youth service, another route to reaching this vulnerable group. The SHA have also cited the research to access grant funding to support SHAYP to the value of GBP1,000,000 (Children in Need; Big Lottery) [**S1**].

SHA staff have drawn directly on the research to inform their professional practice and to support parents to start early conversations with children about HD [**S2**]. Aspects of the SHAYP approach that have been developed based upon the research include educational resources; one-to-one support; age-tailored group sessions; activity days; and residential breaks. Another key area of activity involves supporting young people through the predictive testing process. SHAYP has also reformulated its approach to working with partners of young people living with HD. Following publication of the research on disclosure to partners, SHA Specialist Youth Advisors included relationships and disclosure as part of their comprehensive assessment of needs, resulting in 70% of young people who have entered into serious relationships requesting at least one joint session with their partner and enabling them both to be able to plan better for the future. The SHAYP Children Services Manager said: "*The research… has positively influenced professionals' knowledge and understanding of living in families impacted by HD, whilst also affording organisational change in how services are tailored and delivered to this vulnerable client group*." [**S2**].

**Informing the development of new support services for young people around the world**

As well as informing support within Scotland, the research has also been pivotal in the services offered by the Huntington's Disease Youth Organisation (HDYO). HDYO was founded in 2011 by Matt Ellison, who first heard about the Aberdeen research through Dr Keenan at the European Huntington's Disease Network meeting in Prague, 2010 [**S3**]. Matt grew up with an HD parent and wanted to change the landscape for other children and young people. He founded HDYO in 2012, as a non-profit organisation providing online support and education for young people around the world aged up to 35 (HDYO.org). The organisation provides age-appropriate educational content including a genetic testing section for teens and young adults, an information pack for schools highlighting the needs of young carers, and information for parents about disclosure to children, all of which draw directly upon the Aberdeen research [**S3**]. For example, the genetic testing section specifically draws on findings about 'the waiting period,' the potential for isolation and the feelings young people may have when going through the testing process.

Since 2013, the website has had over 7,000,000 views, content has been translated into 13 languages and direct support has been provided to over 4,500 young people [**S4**]. Founder Ellison had said that the Aberdeen research "*provides an evidence base to underscore the important work of HDYO and to continue to obtain funding and support from partners.*" HYDO has secured approximately GBP500,000 in the REF impact period. Aberdeen researcher Keenan also served on the HYDO Board from 2012-2017 and is a member of the Feedback Team (hdyo.org/eve/about/105), supporting HYDO to use the research to understand the needs of young people and provide much needed support [**S3**].

**Influencing practice, training and support for health and social care professionals**

As well as providing support for young people directly, SHA also works with genetic professionals to improve their practice. Aberdeen's research on the variation in professional practice of genetic counsellors and clinical geneticists in Scotland, whose role is to facilitate communication within families about genetic risk, has directly influenced the development of a new strategic approach to supporting genetic professionals to hold such conversations. While this is a new development for the service, they have already received positive results, including one practitioner who said, "*My practice has been improved dramatically and I believe the benefit is to all family members now and not just the person receiving the genetic test. Had I not read the journal article, I would have continued with my old methods and would have been missing fully supporting families*" [**S2**].

Professional training has used the Aberdeen research to provide an evidence base. The Genetic & Genomic Counselling MSc at Cardiff University cites the Aberdeen research for subtopics in family matters and in predictive testing in their ethics module. This course has been delivered to 80 genetic counselling students since Aug 2013 all of whom have had the benefit of learning about the Aberdeen research [**S5**]. Manchester University also provides clinical training in genetic counselling citing the Aberdeen research in their counselling and communication skills module [**S6**].

The first-of-its-kind professional development course at Stirling University, 'Huntington's Disease: An Enabling Approach to Supporting Families' developed in conjunction with the SHA, cites the Aberdeen research 12 times in the 200-hour, SCQF level 10 course. Since its launch in 2015, 82 professionals from across the four nations of the UK have completed the CPD module [**S7**]. The CEO of SHA said of the Stirling course in relation to the Aberdeen research "*a key theme of the course is the impact of HD on the whole family, and recognition of the issues for young people and young adults is largely possible because of this body of work*" [**S1**].

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

[**S1**] Testimonial from SHA Chief Executive Officer
[**S2**] Testimonial Specialist Youth Advisor and manager of SHAYP
[**S3**] Testimonial from founder of HDYO; HDYO webpage feedback team: hdyo.org/eve/about/105; HDYO youth camp: https://www.youtube.com/watch?v=w7R_s83DiTo
[**S4**] Testimonial from Executive Director of HDYO
[**S5**] Cardiff University course: MSc in Genetic & Genomic Counselling information; Testimonial email from Programme Director
[**S6**] University of Manchester MSc Genomic Counselling information; Testimonial email from Pathway Lead
[**S7**] University of Stirling course on Huntington's Disease: an enabling approach for supporting families; FOI course metrics