**Department of Health Genetics Research Programme**
**Health Services Research projects**

**Final Report**

**Project code:** HSR07
**Date of report:** 31/12/09

**One signed copy and an electronic copy of this report should be sent to Dr Julian Hughes, LGC**

**1. Title**

Deaf individuals' understanding and perception of genetics and their needs from a genetic counselling service

**2. Principal Investigators and Research Team**

Principal Investigator (hearing):

- Dr Anna Middleton, Consultant Research Genetic Counsellor and Research Fellow, Institute of Medical Genetics, Cardiff University

Co-applicants (hearing):

- Prof Angus Clarke, Consultant Geneticist and Reader, Institute of Medical Genetics, Cardiff University
- Prof Graham Turner, Chair of Interpreting & Translation Studies, Department of Languages & Intercultural Studies, Heriot-Watt University
- Dr Maria Bitner, Consultant Geneticist and Reader, Clinical Genetics Department, Great Ormond Street Hospital, London

Research Associate (Deaf):

- Dr Steve Emery, Research Fellow, Department of Languages & Intercultural Studies, Heriot-Watt University

Steering Group (Deaf, hard of hearing and hearing):

- Prof Dai Stephens (hard of hearing), Retired Director of the Wales Institute for Hearing, University Hospital of Wales
- Susan Daniels OBE (Deaf), Chief Executive National Deaf Children's Society, London
- Paul Redfern (Deaf), Deafness and Disability Consultant
- Prof Srikant Sarangi (hearing), Director of the Health Communication Department, Cardiff University
- Prof Martin Richards (hearing), Retired Director of the Centre for Family Research, University of Cambridge

**3. Start date, duration and end date of the project**

1/11/05 – 31/12/09

The original project was 36 months but a no-cost extension was granted due to maternity leave and part time working

**4. Project summary**

The project aims to offer information to help design a genetic counselling service that is applicable, relevant and sensitive for deaf people and their families. Deaf adults are often interested to know why they are deaf and whether this can be passed on to their children – issues routinely addressed within genetic counselling. However, few deaf adults utilise this clinical service. There may be many complex reasons behind this – e.g. lack of information, assumptions about inheritance, mistaken beliefs of a link between present-day genetics services and eugenic practices of the past or fears about being told not to have children. This

project aims to gather the views of d/Deaf people with respect to their beliefs about inheritance, flow of information through families and communication of genetic risk. The study will also assess what attitudinal barriers may possibly be preventing access to genetic counselling. Deafness is a relatively common genetic condition, and the findings from this work may offer a model that is applicable to other genetic conditions where referral rates to genetic counselling are also low. The study will use both a quantitative and qualitative research approach, using postal questionnaires and interviews conducted with a Deaf researcher using British Sign Language.

**5. Aims and Objectives**

**Structured Questionnaire (mainly quantitative data)**

- To explore and analyse the experience of deaf people in relation to genetics issues (e.g to assess perception and understanding of genetic risk and genetic counselling).
- To find out whether deaf people would be interested in the questions that genetic counselling services can answer, such as is my deafness genetic? What is the chance of passing deafness on to children?
- To document the numbers of deaf people interested in genetic counselling but not currently accessing it and to understand why.

**Semi-Structured Interview (qualitative data)**

- To understand more about the fear of genetic technology including the historical and personal context to this.
- To explore further the reasons why deaf participants do not access genetic counselling.
- To explore the experience of those who have had genetic counselling.
- To explore the use of genetic terminology in signed language and the understanding behind this.
- To explore the communication of genetics issues within the family, e.g. by looking at explanations of deafness inheritance and expectations related to this (including hopes about having deaf or hearing children and reasoning behind this), flow of information through the family and communication of genetic risk.

**Workshop**

- To validate the quantitative and qualitative results via a national workshop involving deaf, Deaf and hard of hearing consumers of NHS services and health professionals and academics with an interest in deafness and genetic counselling.

**Produce Practical Outcomes**

- To use the above data to make an evaluation of how genetic counselling services should be delivered to deaf people and to understand and offer solutions to the barriers to effective service provision.
- To provide an evidence base for a model that may be applicable to other genetic conditions.

**6. Achievements of the project**

Please provide a summary (no more than one side of A4) setting out the achievements of the project. These should relate to the original aims and objectives of the project, but should not necessarily be limited by it.

A 32-item structured questionnaire was designed which explored the attitudes of deaf, Deaf[1] and hard of hearing people in relation to various issues relating to genetic counselling. For example, knowledge about what genetic counselling is, whether they would be interested in having it and their reasons for this. The questionnaire also analysed whether participants were aware of how to

[1] 'Deaf' written with an uppercase 'D' refers to people who use British Sign Language as their first or preferred language. 'Deaf' written with a lowercase 'd' is often used in this text to refer collectively to all people affected by deafness, including those who call themselves Deaf, hard of hearing, deafened or hearing impaired.

get genetic counselling and whether they felt they could easily explain their needs to their GP. Data from the questionnaire also showed the numbers of people interested in genetic counselling but not currently accessing it together with reasons why. More than 5000 questionnaires were sent out to potential participants via two magazines for deaf, Deaf and hard of hearing people and 1098 completed questionnaires were returned.

Participants completing the questionnaire could indicate if they were willing to be interviewed to explore their views further, 30 people were interviewed. These interviews were conducted by a Deaf Research Associate in British Sign Language (BSL). The interviews were video-taped and transcribed into written English for analysis. The semi-structured interview gathered in-depth information on views towards genetics and genetic counselling, fears of genetic technology including the historical and personal context to this as well as possible reasons why so few deaf people access genetic counselling. In addition to this participants were asked to share their own thoughts about having deaf children and how this was discussed in the family. Sections of interview were shared with other researchers from the University of Manchester to look at the use of genetic terminology in signed language and how genetic language was being used in the natural environment.

The questionnaire data underwent a descriptive analysis and the interview data a thematic analysis. The results were used to make an evaluation of how genetic counselling services should be delivered to deaf people.

The project identified that there were a set of barriers that deaf and hard of hearing people face in accessing genetic counselling services, these included:

- lack of understanding about what genetic counselling was
- fears about the perceived uses of genetic technology
- uncertainty about how to seek out a referral
- anticipated communication difficulties in explaining that a referral was required
- previous poor experience of using the NHS and an anticipated lack of Deaf Awareness from genetics professionals

Since relevant barriers were identified through the research it is now possible to begin to offer solutions to these barriers. Creating and distributing recommendations on best practice for working with deaf families has formed one way of addressing the possible lack of Deaf Awareness amongst genetics professionals. However, a much more proactive approach is needed to actively offer solutions to the barriers. This will be addressed in the next research project, for which funding has been sought via a project grant from NIHR Health Services Research programme.

It is likely that the research results will provide an evidence base that is applicable to other genetic conditions. For example, it is known that other patient groups such as adults with achondroplasia or Turner's syndrome rarely come for genetic counselling. The above barriers that have been identified are likely to be relevant to these groups too.

A booklet and DVD were produced which offered best practice recommendations for genetics professionals on the delivery of genetic counselling for deaf families. A book was also written for NHS professionals about how to communicate and work effectively with deaf, Deaf and hard of hearing patients. A Workshop was delivered to an audience of deaf, Deaf and hard of hearing lay members of the public as well as NHS health professionals and academics interested in deafness and genetic counselling. The aim of this Workshop was to deliver the research findings, debate these and thus validate the findings and recommendations.

The practical recommendations and information for genetics health professionals, based on the research findings, are provided in the next section.

**7. Achievements of the project: Lay summary**

Please provide a summary of the project (no more than 200 words) suitable for a lay audience, which can be

published and used by the Department of Health.

Genetic counselling is an NHS service which offers information about any condition that could be inherited. Deaf adults are often interested to know why they are deaf and whether this can be passed on to their children – issues easily addressed within genetic counselling. However, few deaf adults utilise this clinical service. The project aimed to identify the barriers faced by deaf people in accessing services; this was done via the analysis of questionnaires and interviews from 1098 deaf and hard of hearing individuals. Interviews were conducted in British Sign Language with a Deaf researcher. The results identified 5 key barriers preventing access to genetic counselling services:

- lack of understanding about what genetic counselling is
- fears about the perceived uses of genetic technology
- uncertainty about how to seek out a referral
- anticipated communication difficulties in explaining that a referral is required
- previous poor experience of using the NHS and an anticipated lack of Deaf Awareness from genetics professionals

The research findings have been used to create a set of recommendations for genetics health professionals on how to offer a genetic counselling service that is applicable, relevant and sensitive for deaf people and their families.

## 8. Research progress

Please give a detailed description of the research undertaken within this project.

In accordance with the aims and objectives of the project a structured questionnaire was designed (see Appendix A) and delivered to deaf/Deaf and hard of hearing people across the UK. In addition to this semi-structured interviews were conducted in British Sign Language to explore in-depth views. The following sections cover details of how each stage of the research was completed and delivered.

### 1. Questionnaire Design

The questionnaire was written by the PI, together with input from the co-applicants and steering group. The content was based on new, original work addressing issues specific to the research aims, as well as existing measures that had been used and thus validated with deaf and hard of hearing people in other research. A 32-item, non-standard questionnaire was designed for use with Deaf, deaf and hard of hearing participants, some of whom used spoken language and others sign language. The terminology ‘Deaf’ written with an uppercase ‘D’ refers to people who use British Sign Language (BSL) as their first or preferred language.

The questionnaire was introduced with a very basic overview of what genetic counselling is. The questions then ascertained views about new discoveries in genetics and knowledge of genetic counselling, including how to obtain a referral and thoughts on what happens in a consultation. Those who had previously had genetic counselling were able to give feedback on their experience of this and those who had not had genetic counselling previously were able to comment on whether they thought they would want it in the future giving reasons for their views. Participants could indicate on the questionnaire if they were willing to be interviewed in BSL to explore their views in greater depth.

The questionnaire was checked for face validity, readability and understanding and went through 17 drafts. The following groups were involved in this drafting process:

- Genetic counselling researchers offered input on question structure and face validity. One of these genetic counsellors is a specialist in working with deaf participants
- Deaf, hard of hearing and deafblind members of the public via the National Deaf Children’s Society and Sense (charity for deafblind people) offered input on the relevance of questions and ease of understanding, together with readability

- A teacher for the Deaf simplified the language, so that although it was still in written English, it also made sense in BSL
- Two Deaf BSL users from Deaf Association Wales critiqued the questionnaire language construction and cultural sensitivity and ensured that the language met 'Plain English for Deaf People' standards and also could be easily translated into BSL
- A statistician reviewed the questionnaire to assess how the data should be collated and analysed
- The readability function was calculated:- Flesch Reading Ease of 64.8 and a Flesch Kincaid Grade Level of 6.8. The higher the Flesch Reading Ease scale the better (this is subjectively associated with readers of national newspapers, e.g. a score of >80 = Sun Reader, > 70 = Express Reader a score of 65 is considered good in terms of understanding in Plain English), the lower the Flesch Kincaid Grade the better (7-8 = 13-14 year old child's reading).

Once the questionnaire was designed it was subjected to two pilot studies, these involved the following groups: 1) 11 teachers of deaf children, the teachers were all deaf themselves, 2) 2 Deaf members of the public whose first language was BSL. The questionnaire was again revised in light of how these participants completed the questions and their apparent ease of understanding the questions. The coding frame for data analysis was created and tested using the pilot data. The questionnaire structure was fine-tuned again in response to this exercise.

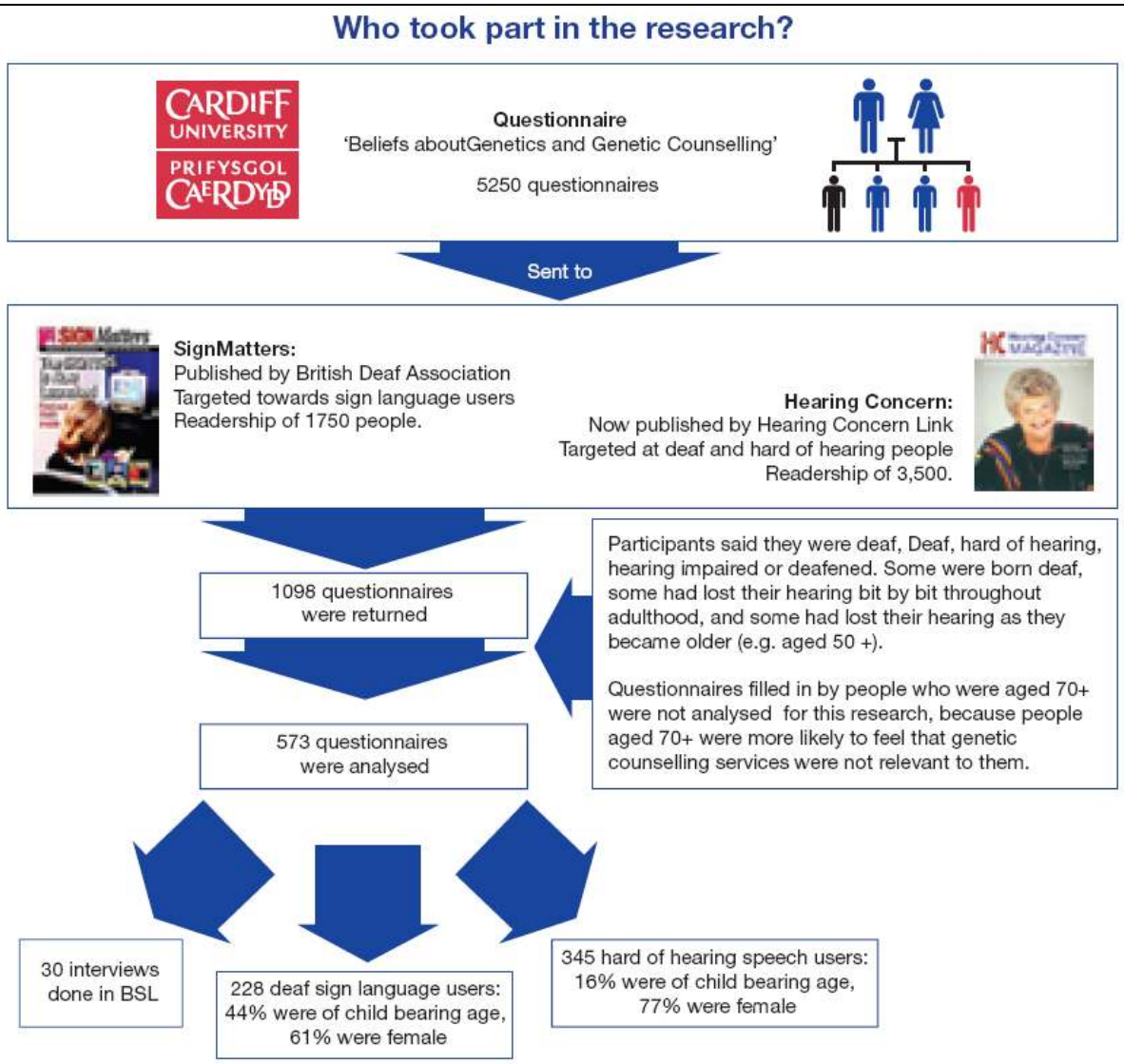
**2. Recruitment/Ascertainment**

Participants were ascertained for the study via the readership of two British magazines for Deaf and hard of hearing people, 'Sign Matters' (now named the British Deaf News) and 'Hearing Concern' (now named Hearing Matters). 'Sign Matters' is published by the British Deaf Association and is particularly targeted towards Deaf sign language users in the UK. In parallel, 'Hearing Concern' was published by a charity of the same name and targeted at deaf and hard of hearing people. In 2006, all copies of one issue of both magazines (i.e. n = 5250: 'Sign Matters' 1750; 'Hearing Concern' 3500) had a questionnaire included together with an invitation to participate in the study.

In the original project proposal we had planned to design a website that could deliver the questionnaire and capture data from participants completing it. There was extensive discussion with the IT department at Cardiff University about the logistics of doing this and the ability to deliver the questionnaire online via podcasts in BSL. However, after exhausting all practicalities on how this could be achieved, in a computing sense, the decision was made to drop this idea, due to the extensive (uncosted) time that the IT department would have to contribute. This decision was also based on discussions held with the Centre for Deaf Studies (CDS) at the University of Bristol. CDS is one of the leading, international centres for Deaf Studies research involving both Deaf and hearing academics. At the time (in 2006) they felt that there wasn't the IT expertise to adequately capture BSL data via an online questionnaire. It is worth adding that since 2006 IT technology has improved extraordinarily and in 2009 an excellent questionnaire was produced by the University of Manchester which allows participants to complete an online questionnaire via webcams and deliver their answers in BSL. A similar format will be used by the research team in future projects.

In the original proposal we explored many different possible recruitment methods that could be utilized. Using the two magazines for ascertainment of participants was very successful and also time consuming (questionnaires were still being returned 6 months after they had been disseminated). Considering the fact that large numbers of questionnaires had already been returned (1000+) and the timeline for gathering questionnaire data had been completed, we decided, in consultation with the steering group and a lead statistician from the Clinical Epidemiology Interdisciplinary Research Group at Cardiff University, to limit the recruitment methods to just the magazines.

**3. Sample Size and Structure**




1098 people returned a completed questionnaire. Participants defined themselves as deaf, Deaf, hard of hearing, hearing impaired, deafened or having a specific level of hearing loss, and included those who were born deaf, those who had lost their hearing in childhood, those who had progressive, adult onset hearing loss, as well as those who had lost their hearing as they approached their older years (e.g. aged 50 +). No hearing people participated. Participants could indicate whether they identified more with the Deaf community, Hard of Hearing community, mainstream hearing society, all communities or did not feel part of any community.

99 questionnaires were discounted from the analysis either because they were unintelligible or returned too late for coding. 426 questionnaires were returned by participants who were over the age of 70 when the questionnaire was completed. This group were the most likely to give incomplete or missing answers and were also the most likely to indicate a level of uncertainty of the relevance of the study to them. In reality attendance at genetic counselling clinics is unusual for people over the age of 70. We therefore excluded the over 70's from some of the analysis; the remaining sample size for the study was 573.

### 4. Ethical Approval and Monitoring

Multi-centre Research Ethics Committee approval was granted for the research (with limited amendments required), so too was permission to carry out the study from the Cardiff and Vale NHS Trust and the Cardiff University R+D department. The PI submitted periodic progress reports to the

local NHS Research and Development Ethics committee as well as to the Welsh Assembly Government via the Trust R+D department. Also quarterly progress reports were submitted to the Health Services Research team at the Department of Health.

### 5. Analysis of Quantitative Data

All questionnaire data was coded by a statistician and entered onto the software package SPSS 14.0. Descriptive statistics were used involving cross-tabulations and chi-squared analysis. Within the quantitative data analysis, comparisons were made between participants who used sign language as their first or preferred language and those who used spoken language as their mode of communication. The descriptive analysis of the quantitative data was managed by the PI. Four different statisticians were consulted at different time points in the project to offer advice on data analysis and to check methods for interpretation of data.

Participants were able to give open, free text comments in the written questionnaire; these were coded and ordered into different themes. The analysis was initiated by open coding and then further refined by axial coding.

### 6. Semi-Structured Interviews

The interviews were conducted with the Deaf Research Associate in BSL throughout the UK in 2007-2008, and were digital video-recorded. 75 participants indicated on the written questionnaire that they would be willing to be interviewed in BSL. 64 participants were selected for interview; they were chosen if their questionnaire answers indicated that they were likely to have a genetic deafness (and thus the issue of genetic counselling could be deemed of most relevance). Of the 64 invited for interview, 30 were actually interviewed (the others later declined, were not contactable or withdrew). Given the fact that a large proportion of the interview cohort declined to participate (after originally agreeing) the Research Associate's contract had to be extended by 12 months. This was because so much time was spent chasing up potential participants as well as travelling to those who consented to participate. We are unsure as to the reason why so many declined participation after an initial enthusiasm, but postulate that it might be related to the media coverage of the Human Fertilisation and Embryology Bill (HFEB) which was being discussed in Parliament at the same time as participants were being contacted about the interviews. The media at the time were discussing Clause 14 of the HFEB which covered issues relating to pre-implantation genetic diagnosis for deafness and the restrictions on the use of this technology by Deaf people. There was a large backlash from the Deaf community in the UK about this Clause [see publications 3, 11-13, 20, 28 in the section on dissemination] and it is possible that this played a part in the decision from people to pull out of the study. It is impossible to know more about whether this is indeed the case as it would have been unethical to push potential participants to explain their reasons for declining participation.

A hearing person familiar with the professional requirements of translating signed data into written text from academic projects produced written transcripts from the video interviews. This work was funded by the government Access to Work programme via Heriot-Watt University.

### 7. Content of the Semi-Structured Interviews and Pilot Work

The interview schedule was designed with input from researchers at CESAGen (Centre for the Economic and Social Aspects of Genomics) at Cardiff University. This centre specialises in social science research on issues surrounding genetics and has experts in qualitative data collection and interview delivery. A seminar was delivered by the PI on the research project to CESAGen and several members of the centre gave specific input on the interview structure. The content was checked for face validity and relevance.

The interview schedule was piloted on the Deaf Research Associate, delivered in speech by the PI and interpreted by a BSL interpreter. This was done so that the Deaf Research Associate would experience the interview as a 'participant', thus helping him to work out how we might deliver the questions in BSL and how participants might answer these. This was also done so that the PI

could carefully demonstrate how the interview questions should be delivered. The Research Associate then piloted the interview with him as interviewer with a Deaf colleague as interviewee. This interview was conducted entirely in BSL and was video-recorded. The video was also interpreted live so that there was a voice-over on the film in spoken English. The Research Associate and PI discussed how the interview appeared from the interviewer's perspective and some of the question structures were changed in light of this discussion. The RA then conducted two more pilot interviews with Deaf members of the public. These were also filmed and used as a practice run for the interview data analysis and to check the question delivery, participant response and to also to practice the video data capture, filming and editing for dissemination. The pilot interviews were transcribed into written English and a preliminary analysis done by the PI with extensive discussion about potential thematic analysis with researchers from CESAGen.

**8. Consent to Share Video Data**

An extensive consent procedure was created which allowed ample 'cooling off' time, in case participants decided that they didn't want to be interviewed, or didn't want their interviews to be video-taped. On completion of the questionnaire, participants could indicate if they were willing to be contacted about having an interview, thus consent was deemed implicit if they filled in their contact details and returned these to the research team. The participants were then contacted about the interview and given a booklet about the study and what to do if they wished to pull out or make a complaint. They were also sent a consent form to read ahead of the interview. Once the RA arrived at the participant's house for the interview, the consent form and process was carefully discussed in BSL. Participants could choose to be filmed and also receive a copy of their interview on DVD, so they could check that they were happy with what they discussed. On receipt of their DVD they had a month in which they could inform the research team that they did not wish to be involved further in the study, in which case their video would be destroyed and not used in the research. They could also choose the level of involvement that their interview would have in the dissemination process and were informed that their video image could be used in a public setting, e.g. at a conference. This meant that clips of their interview could be shown to an audience of people that they might know. Participants then filled out the consent form giving details of how they wanted their interview data to be utilised by the research team (see Appendix G for consent form).

**9. Qualitative Data Analysis**

A 'pragmatic variant' grounded theory approach was used to guide the data analysis. Codes and subsequent themes were assigned. The data was coded in the first instance by the PI and secondly by the RA, thus enhancing reliability. We used an iterative approach, with the analysis of the first interviews offering additional lines of enquiry in later interviews. 30 semi-structured interviews were completed and saturation of themes achieved.

**10. Analysis of Interview Data**

The interviews were analysed two-fold. The first analysis was conducted by the hearing PI using the written translated transcripts. Here a thematic analysis was performed, starting with an open coding procedure and then refining with axial coding [as shown in publications 2, 5-7 in the dissemination section below]. The second analysis was by the Deaf researcher who conducted the interviews using the original BSL data [see presentations 20-21 in the dissemination section].

Two examples are provided in Appendix B of papers that have been accepted in peer reviewed journals for publication [publications 2 and 4].

**11. Results and Recommendations from Research**

A list of recommendations as well as statements of information are given in a booklet entitled: 'Working with deaf people: a handbook for genetics professionals'. This booklet has also been translated into BSL and appears as a DVD in the backcover (see Appendix H). A book has also been written, published by Cambridge University Press, entitled: 'Working with deaf people: a handbook for health professionals', which contains an extended version of the recommendations for genetics professionals (see Appendix F). The content of both the booklet and book contain

evidence-based findings from the research, they have also been checked for face validity with the audience of the Workshop. A précis of the recommendations are included here:

- Deaf sign language users and hard of hearing speech users may have completely opposite views towards deafness.
- Genetics professionals need to be aware how modern genetics services relate to history. Some deaf and hard of hearing people may link genetic counselling to eugenic practices of the past, when people tried to stop more deaf people being born.
- Genetics professionals need to be aware that there are often misunderstandings about what genetic counselling is. Deaf and hard of hearing people may think that one of the key aims is to find out if babies have inherited deafness before they are born and abort them if the baby is found to be deaf. It can be helpful to reassure deaf clients that this is not a key aim of genetic counselling.
- Genetics professionals need to be aware that deaf and hard of hearing people may have a whole variety of attitudes towards genetics services. Some may feel positive, some neutral and some negative.
- Genetics professionals need to be aware that hard of hearing speech users often hope that genetic technology will help to find a cure for deafness, but some Deaf sign language users feel threatened that deafness may be wiped out by the use of genetic technologies.
- Deaf people do not always understand what genetic counselling is, so genetics professionals should focus on explaining what can be offered in a consultation and find out what their deaf and hard of hearing clients expect.
- Deaf and hard of hearing clients may not be keen to talk to their GP about issues linked to genetics. This is because they expect there will be communication difficulties, or they wrongly think that their GP won't know about genetic counselling.
- Genetics professionals need to be aware that some deaf people are sensitive towards genetic language. Avoid words like 'mutant', 'mutation', 'abnormal', 'normal', be sensitive to words like 'risk'.
- Genetics professionals may need to contact other relatives to find out more information about the family history of a deaf or hard of hearing person as some deaf individuals are unaware of their family history due to communication difficulties in the family.
- Deaf people are interested to know why they are deaf and whether this can be passed on to their children or not. Deaf people are interested in having genetic counselling for other things as well as deafness.
- Some deaf people prefer to have deaf children, some don't mind whether their children are deaf or hearing, and others prefer to have hearing children.
- Genetics professionals need to be aware that some deaf people may rather not focus on deafness within a consultation, especially if they have been referred to talk about something else.
- Genetics professionals need to explain carefully why they might need to ask questions about a family history of deafness (for example, to find out if there is a syndrome linked to the deafness), because deaf people may not understand the aims of these questions.
- At least one member of administration staff and one member of clinical staff in each genetics department should have Deaf Awareness training.
- All genetics professionals who regularly see deaf and hard of hearing clients (for example, on a monthly basis) should have Deaf Awareness training.
- At least one member of the genetics team which often sees Deaf sign language users (for example as part of a specialist deafness clinic, perhaps on a monthly or even weekly basis) should have training in BSL; at least to a basic level.
- Genetics professionals who specialise in working with deaf clients and who frequently see Deaf sign language users should aim to become fluent in BSL and be able to give a consultation in BSL.

In addition to the above recommendations, the research explored the preferences that participants

might have for communication in a hospital (e.g. genetic counselling) consultation (see Appendix B for details). The results showed that deaf and hard of hearing participants who use spoken language preferred to have a consultation in speech, but only if the health professional had a good level of Deaf Awareness, in that they could be lip-read easily and took time to make allowances for good communication. Those participants who preferred to use BSL were very unlikely to accept a consultation in speech, most were content with an interpreter but some said they would far prefer to have health consultations directly in BSL with signing health professionals. The hospital communication preferences for most people with deafness could be met by increasing Deaf Awareness training for health professionals, a greater provision of specialised sign language interpreters and of health professionals who can use fluent sign language directly with clients in areas where contact with deaf people is frequent.

The study explored the views, knowledge and beliefs that some Deaf and hard of hearing people have about genetics and genetic counselling. Our results showed that 50% of the sample indicated they knew what genetic counselling was before they read the questionnaire but 80% did not know how to get genetic counselling. Thus there are a set of barriers that are preventing deaf and hard of hearing people from accessing services, we have identified the following:

- lack of understanding about what genetic counselling was
- fears about the perceived uses of genetic technology
- uncertainty about how to seek out a referral
- anticipated communication difficulties in explaining that a referral was required
- previous poor experience of using the NHS and an anticipated lack of Deaf Awareness from genetics professionals

**12. Validation of Research Findings**

We ran a Workshop entitled 'Deafness and Genetics: What do deaf people want? Public consultation for deaf/hard of hearing people and health professionals', this was on 18 September 2008 11-5pm at the Millennium Centre, Cardiff Bay (see Appendix C for flyer). This was attended by 120 people including deaf and hard of hearing members of the public as well as NHS health professionals interested in deafness (e.g audiologists, nurses, genetic counsellors, GPs) and academics (e.g. from the Centre for Deaf Studies at University of Bristol, from University of Manchester, Cardiff University) as well as support groups for deaf people (e.g. British Deaf Association, Royal National Institute for Deaf People, National Deaf Children's Society, Deaf Education through Listening and Talking and other members of the UK Council on Deafness). The aim of the Workshop was to disseminate the project results and check if these seem reasonable (validation of results) as well as to offer information about genetic counselling. The list of recommendations that were to appear in the booklet summarising the project findings were also double checked with the Workshop audience, again as a form of validation of the project findings. The research team also performed two role plays – 'the genetic counsellor from hell' and 'the genetic counsellor from heaven', which were a fun way of getting the message across about deaf awareness (how it should and shouldn't be done) within a genetic counselling consultation. The full Workshop was filmed by the Media Resources team at Cardiff University and an hour long edited version of the day was put on DVD to accompany the booklet of recommendations from the project.

**13. Consultation with the Research Team**

The co-applicants, Research Associate and steering group met for one whole day on a yearly basis to discuss the project progress and offer advice on direction of the research. This was an important opportunity for input from the Deaf lay members of the group (consumers of NHS services and lay members of the public). An agenda and post-meeting minutes for each meeting were circulated to the Department of Health.

**14. Feedback of Study Recommendations to Research Participants**

Participants completing the study questionnaire could indicate at the end if they wished to receive more information on genetic counselling. They had the option of receiving this in written English (a

leaflet, see Appendix D) and/or in BSL on a DVD. Approximately 500 participants indicated that they wanted to receive such information and so this was sent out.

At the end of the study, a booklet and DVD on the research findings and recommendations to genetics professionals was produced (see Appendix H). The DVD also contained an edited hour from the Workshop at the Millennium Centre in Cardiff on the research. This was sent out to every participant who was interviewed for the study; as well as to participants who took part in the research and attended the Workshop (approximately 50 of these individuals were sent the booklet and DVD; additional booklets and DVDs were sent elsewhere, see later for details).

Articles on the project results were also written for the British Deaf News (formally Sign Matters – one of the magazines in which the study questionnaire was disseminated) and Hearing Matters (formally Hearing Concern – the other magazine that the study questionnaire was disseminated in). Therefore, there were several steps taken to inform the people who participated about the outcomes from the study in which they took part.

**15. Feedback of Study Recommendations to NHS and Academia**

The booklet and DVD on the study results and recommendations for genetics professionals was sent out to every genetics department in the UK as well as to geneticists and genetic counsellors known to the PI (100 individuals). The booklet and DVD was also sent to every member of GENDEAF, a European group of health professionals and academics who work with deaf patients and have a particular interest in the genetics of deafness (32 individuals across Europe). They were also sent to Deaf Studies academics in the UK and health professionals and academics from the US who have a particular interest in genetic counselling research for deafness and have published in this area (10 individuals).

**16. Feedback of Study Recommendations to Deaf and Hard of Hearing Consumers of NHS Services**

The booklet and DVD on the study results and recommendations for genetics professionals was sent out to every member of the UK Council on Deafness, an umbrella organisation for each charity and support group in the UK that has an interest in deafness (100 organisations).

**17. Dissemination**

Extensive effort has been made to disseminate the project findings in various different forms. This has included publication in peer reviewed journals, 3 papers have been accepted for publication [2-4 in list of publications] and 3 have been written and will be submitted shortly [5-6 in list of publications]. A book has been written [1 in list of publications] and distributed to libraries across the UK as well as to health professionals with a specific interest in deafness. Several book chapters have been written [8-10, 14-15] and conference presentations delivered in speech [16-18] and British Sign Language [19-21] and via posters [22-24]. We have made a specific attempt to also reach practicing genetic counsellors and geneticists who work clinically and may not be able to regularly attend conferences nor read academic journals, this was done via the delivery of seminars [25-26, 28-29, 31-32, 36]. We have also delivered seminars to academic institutions with a particular interest in deafness and/or genetic counselling research [27, 30, 33-36]. Finally, we have also disseminated the research results to deaf and hard of hearing members of the public [37-40].

**18. Additional Funding Awarded**

Three additional grants were awarded to support the printing costs of the booklet and DVD and filming costs of the Workshop on the study. These grants also helped with additional costs relating to the Workshop, allowing us to run this from a prestigious venue (Millennium Centre in Cardiff Bay) rather than within unsuitable (but free) rooms at the university. The Royal National Institute for Deaf People awarded £1650, the Community Engagement Fund at Cardiff University awarded £1212 and Deafness Research UK awarded £4050.

**19. Collaboration with Other Researchers**

Ethical approval was gained to involve the qualitative interview data in other relevant research studies. Consent was given by participants to have their interviews involved in further research as long as additional local ethical approval was granted for these studies. Consequently, interviews from those participants who gave consent, were shared with a doctoral student from Manchester University. A contract was set up between Cardiff University and Manchester University governing the way the interview data would be used and who it would be shared with. Sections of interviews were chosen by the PI and sent to the doctoral student who will be analysing the data to look at genetics terminology used in BSL by participants.

**20. Networking within Cardiff University**

The PI was able to mix and network with social science researchers from CESAGen (Centre for the Economic and Social Aspects of Genomics), the Psychosocial Research group at the Institute of Medical Genetics, Cardiff University and the Health Communication Department at Cardiff University, this proved fruitful in terms of enabling discussion about research methodology as well as intellectual and academic issues relating to social science research in genetics. The PI also mixed with the Wales Gene Park, writing articles on the deafness project for the Wales Gene Park newsletter and also presenting at one of their conferences.

**21. Additional Public Engagement Event**

The PI was approached by the Wales Gene Park and also the Progress National Trust to organise an evening debate: 'Debating Deafness and Embryo Selection: Are We Undermining Reproductive Confidence in the Deaf Community?' (see Appendix E for flyer). This was scheduled for 9th April 2008 and was attended by approximately 150 members of the public. Three members of the research team (PI – Dr Anna Middleton, Research Associate- Dr Steve Emery and Co-applicant – Prof Graham Turner) all presented at this event, providing information about the research project and also philosophical and ethical discussion about the Human Fertilisation and Embryology Bill.

**22. Importance of this Research**

The reaction of the Deaf community to the Human Fertilisation and Embryology Bill and Clause 14, which could potentially have an impact on the reproductive liberty of deaf people in the UK, provides evidence that deaf people are VERY sensitive to issues surrounding genetics. We have already shown this in previous research, however, this arose again in the present study. This illustrates how important it is that Clinical Genetics services engage appropriately with this group of people and make particular attempts to address the fears and difficulties they have with genetics. As we have shown, Deaf, deaf and hard of hearing people have particular unique needs, which are different to those of hearing people.

## 9. Dissemination of results

Please report and provide information on the dissemination and translation of the results of the project during the entire project under the following headings.

**Patentable results including any patents submitted**

Not applicable

**Publications, conference presentations etc**

Book

1. Middleton A (Ed) (2009) Working with deaf people – a handbook for health professionals. Cambridge: Cambridge University Press

Papers in Peer Reviewed Journals

2. Middleton A, Emery SD, Turner GH (2010) Views, Knowledge and Beliefs about Genetics and Genetic Counselling amongst People with Deafness. Sign Language Studies (in press)
3. Emery SD, Middleton A, Turner GH (2010) Whose deaf genes are they anyway? The Deaf community challenge to legislation on embryo selection. Sign Language Studies (in press)

4. Middleton A, Turner GH, Bitner-Glindzicz M, Lewis P, Richards M, Clarke A, Stephens D (2010) Preferences for Communication in Clinic from Deaf People: a Cross-Sectional Study. Journal of Evaluation in Clinical Practice (in press)

Forthcoming Papers

5. Middleton A, Turner GH, Bitner-Glindzicz M, Lewis P, Richards M, Clarke A, Stephens D (2010) Are deaf people interested in genetic counselling? Forthcoming, written for the Journal of Genetic Counseling
6. Middleton A, Turner GH, Bitner-Glindzicz M, Lewis P, Richards M, Clarke A, Stephens D (2010) Barriers to accessing genetic counselling amongst deaf people. Forthcoming, written for the European Journal of Human Genetics
7. Middleton A, Turner GH, Bitner-Glindzicz M, Lewis P, Richards M, Clarke A, Stephens D (2010) Research with the deaf community - methodological issues relating to definitions of deafness and culture, participant collection and questionnaire preparation. Forthcoming, written for The Application of Clinical Genetics

Invited Book Chapters, Papers and Letters That Have Been Editor Reviewed

8. Middleton A (2009) General themes to consider when working with deaf and hard of hearing clients. In: A Middleton (Ed) Working with deaf people – a handbook for health professionals. Cambridge: Cambridge University Press, p29-83.
9. Middleton A (2009) Specialist issues relevant to working with d/Deaf clients. In: A Middleton (Ed) Working with deaf people – a handbook for health professionals. Cambridge: Cambridge University Press, p129-138.
10. Middleton A, Neary W, Moller K (2009) Facts and figures about deafness, NF2 and Deafblindness. In: A Middleton (Ed) Working with deaf people – a handbook for health professionals. Cambridge: Cambridge University Press, p1-28.
11. Blankmeyer Burke T, Emery S, Belk R, Middleton A, Turner G (2008) Response to Editorial Change and Stallworthy Letter. British Medical Journal. Letter to the Editor, published online 22 May 2008
12. Emery S, Blankmeyer Burke T, Middleton A, Belk R, Turner G (2008) Reproductive Liberty and Deafness: Clause 14(4)(9) of the UK Human Fertilisation and Embryology Bill should be amended or deleted. British Medical Journal. Letter to the Editor, published online 21 April 2008, published in paper copy: 336:976 (3 May)
13. Blankmeyer Burke T, Belk R, Middleton A (2008) Legislation, Deafness and Reproductive Rights. Comments on Clause 14 of the Human Fertilisation and Embryology Bill. Bionews. Progress Educational Trust. 5 Feb 2008. http://www.bionews.org.uk/commentary.lasso?storyid=3721
14. Middleton A (2007) Attitudes of deaf people and their families towards issues surrounding genetics. In: A Martini, D Stephens and AP Read (Eds). Genes, Hearing and Deafness. From Molecular Biology to Clinical Practice. London: Informa Healthcare, p163-172.
15. Middleton A (2006) Genetic Counselling and the d/Deaf Community. In: D Stephens and L Jones (Eds). The Effects of Genetic Hearing Impairment in the Family. London: Wiley, p257-284.

Peer Reviewed Conference Presentations – Spoken Presentations

16. Middleton A, Emery S, Turner GH, Clarke A, Sarangi S, Bitner-Glindzicz M, Richards M, Stephens D (2009) 'They just take these genes and fix 'em together and create a fake human' – lay beliefs about genetic counselling from people with deafness and impact on access to services. Communication, Medicine & Ethics (COMET) Seventh Interdisciplinary Conference 25-27 June 2009, Cardiff University
17. Middleton A (2008) Working with Deaf People. Invited speaker at the British Society of Human Genetics Conference 15-17 September 2008, York. Journal of Medical Genetics 45 suppl 1 page, Abstract number: SP28, spoken presentation

18. Middleton A, Emery S, Turner GH, Clarke A, Sarangi S, Bitner-Glindzicz M, Richards M, Stephens D (2008) Why don't Deaf People come for Genetic Counselling? Association of Genetic Nurse Counsellors Spring Meeting 15th May 2008, St Albans Conference Centre, London, spoken presentation

Peer Reviewed Conference Presentations - Delivered in British Sign Language

19. Middleton A, Emery S, Turner GH, Clarke A, Sarangi S, Bitner-Glindzicz M, Richards M, Stephens D (2008) Use of genetics services in the NHS: thoughts from deaf and hard of hearing people. UK Council on Deafness Conference, 6th November 2008, London,
20. Emery S, Middleton A, Turner GH (2008) The End of Deafness? Transition in times of Genetic Legislation. Deaf and Other Lives: Living in Multiple Cultures Conference. September 25 - 26 2008, Amsterdam, The Netherlands
21. Emery SE, A Middleton, GH Turner (2007) I was told I had a moral responsibility to society to abort my son as he would be deaf. Society for Social Studies of Science Annual Meeting. The Deaf/genetics interface. 14.10 Science Technology Studies, Disability Studies and Deaf Studies. 10-13 October 2007, Montreal, Canada

Peer Reviewed Conference Presentations – Poster Presentations

22. Middleton A, Emery S, Turner GH, Clarke A, Sarangi S, Bitner-Glindzicz M, Richards M, Stephens D (2008) Why don't Deaf People come for Genetic Counselling? Quantitative and qualitative findings from a UK study. American Society of Human Genetics Conference 11-15th November 2008, Philadelphia, 300/W
23. Middleton A, Emery S, Turner GH, Clarke A, Sarangi S, Bitner-Glindzicz M, Richards M, Stephens D (2008) Why don't Deaf People come for Genetic Counselling? Quantitative and qualitative findings from a UK study. British Society of Human Genetics Conference 15-17 September 2008, York. Journal of Medical Genetics 45 suppl 1 page. Abstract number: 4.08,
24. Middleton A, Emery S, Turner GH, Clarke A, Sarangi S, Bitner M, Richards M, Stephens D (2008) Why don't Deaf People come for Genetic Counselling? European Psychosocial Aspects of Genetics Conference, Barcelona 31st May – 3rd June. European Journal of Human Genetics, Volume 16, Supplement 2, EP05.1

Invited Conference Presentations/Seminars across the NHS and Academia

25. Middleton A (2009) 'They just take these genes and fix 'em together and create a fake human' - views from Deaf people about genetics. Invited seminar at the Deafness Cognition and Language (DCAL) Centre and UCL Ear Institute at University Central London. 8th October 2009
26. Middleton A (2009) 'They just take these genes and fix 'em together and create a fake human' – views about genetic counselling; barriers to accessing services. Invited seminar at the Department of Clinical and Molecular Genetics, Institute of Child Health, London, 21st July 2009
27. Middleton A (2008) 'They just take these genes and fix 'em together and create a fake human' – views from deaf people about genetics. Invited seminar at the Centre for Family Research, University of Cambridge, 25th November 2008
28. Middleton A (2008) Choosing deaf embryos and the law: the UK Deaf community challenges the UK government. Invited seminar to the University of California in Los Angeles, UCLA Center for Society and Genetics, LA, USA, 30th October 2008
29. Middleton A (2008) Barriers to accessing genetic counselling: thoughts from deaf people and recommendations for genetics professionals. Invited seminar to the Wessex Regional Genetics Laboratory, Salisbury District Hospital, 15th October 2008
30. Middleton A (2008) Deafness and genetics: what deaf people want from genetic services. Invited presentation to the GenCAG group, Department of Health, 8th October 2008, London
31. Middleton A (2008) Why don't Deaf People come for Genetic Counselling? Invited seminar at the Clinical Genetics Department, Birmingham Women's Hospital, 8th May 2008, Birmingham.

32. Middleton A (2008) Human Fertilisation Embryology Bill. Invited seminar at the All Wales Medical Genetics Service, 24th January 2008, Cardiff.
33. Middleton A (2007) Genetic counselling in silence: attitudes of the Deaf community towards genetic counselling. Invited seminar at University of Glamorgan, 25th January 2007, Wales.
34. Middleton A (2006) Deafness, genetics and attitudes. Invited seminar at Health Communication Research Centre, Cardiff University, 11th July 2006, Cardiff.
35. Middleton A (2006) Deaf perceptions about genetics and genetic counselling. Invited seminar at Centre for Deaf Studies, Bristol University, 28th April 2006, Bristol.
36. Middleton A (2006) Deaf perceptions of genetic counselling – setting up a research project. Invited seminar at Institute of Medical Genetics, Cardiff University, 7th April 2006, Cardiff. Also invited to deliver this seminar to CESaGen, Department of Social Sciences Seminar programme, Cardiff University, 21st March 2006.

Dissemination to the Deaf and Hard of Hearing Lay Public

37. Middleton A (2009) Beliefs about Genetics and Genetic Counselling. British Deaf News. October issue. ISSN: 1756-4514
38. Middleton A (2009) Working with deaf people: a handbook for genetics professionals. Booklet and DVD targeted towards genetics professionals and deaf/hard of hearing people who. Printed by Clements Pope, Cardiff, distributed by Cardiff University
39. Middleton A, Emery S, Turner G (2008) Deafness and Genetics: What do deaf people want? Public Consultation for deaf/hard of hearing people and health professionals: 18 September 2008, Millennium Centre, Cardiff Bay 11am-5pm
40. Middleton A (2006) Beliefs about Genetics and Genetic Counselling. Sign Matters. September issue. British Deaf Association: Coventry

**Application of the results and outcomes of the project to the clinic, including benefits of the project outcome to the NHS and clinical practice**

The booklet and DVD on the study results and recommendations for genetics professionals was sent out to every genetics department in the UK as well as to geneticists and genetic counsellors known to the PI (100 individuals); so too was a flyer for our book, 'Working with Deaf People- a Handbook for Health Professionals' (see Appendix F). The same information was also sent to every member of the GENDEAF Group, which is a European group of clinicians, health professionals and academics with a specialist interest in the genetics of deafness and working clinically with deaf patients (32 individuals were sent this work). Finally, the same information was sent to clinicians working with deaf families and specifically interested in genetic counselling for deafness in the US (and who have published on this area, 10 individuals).

The study recommendations are of practical importance to the genetic counselling clinic. This work has been disseminated widely amongst NHS professionals and to NHS organisations. There is limited evidence to support best practice within genetic counselling for deafness and thus the findings from our work is new, innovative and more importantly highly relevant for improving clinical practice. The project therefore has extreme relevance and also benefit to the NHS and clinical practice.

**Interactions with NHS staff not involved directly with the project**

The study results were presented to NHS professionals attending conferences for clinicians, e.g. British Society of Human Genetics conference, the Association of Genetic Nurses and Counsellors conference, the European Psychosocial Aspects of Genetics conference, the American Society of Human Genetics conference. Seminar presentations were also given to clinicians at the Joint Committee on Medical Genetics, GENCAG (group of commissioners of genetics services) as well as numerous NHS regional clinical genetics services.

The PI sought out a meeting with Dr Mike Warburton, GP Access Programme lead, Department of Health to present the research findings and discuss how these may be used by the Department of Health in practice. Dr Warburton is involved in restructuring access to GP services across the whole of the UK (e.g. by increasing opening hours and services available) and as part of this programme is implementing changes that improve access for deaf and hard of hearing people. He was very interested in our research results and has made our book available to every GP practice in the UK.

**10. Matters relating to consumer involvement**

Involvement of Deaf and Hard of Hearing People in the Research Design, Construction and Delivery

Consumers of NHS service, i.e. Deaf and hard of hearing people, have been involved in the design, delivery and dissemination of the research. The Research Associate is a Deaf sign language user; he played a pivotal role in guiding the research so that it was sensitive and appropriate for Deaf participants. He has also delivered the research findings in BSL to Deaf academics and lay members of the public. The Steering Group consisted of Deaf and hard of hearing lay members of the public, one of whom was an ex-health professional. This group have been particularly important in helping to ensure that the research was relevant and fitting for Deaf and hard of hearing users of NHS services. Careful attention has been given to making sure that deaf consumers of NHS services have been involved at every stage of the research process. Our research team has been invited to offer advice on best practice in involving deaf and hard of hearing people in research by the NIHR Primary Care Research Network.

Dissemination of Results to the Lay Deaf and Hard of Hearing Community

The booklet and DVD on the study results and recommendations for genetics professionals was sent out nationally to every support group and charity involved with deaf and hard of hearing people. So too was a flyer for our book, 'Working with Deaf People- a Handbook for Health Professionals'. These were sent to 60 different groups.

As already discussed, the national Workshop run by the research team offered a forum for disseminating the study results to a large audience of deaf and hard of hearing lay public.
The PI sought out the Public Relations department at Cardiff University to discuss how to engage with the media on the findings of the project. A press release about the findings of the project was consequently written and disseminated to the media. The PI was invited to discuss the implications of the research project as well as national issues relevant to deaf and hard of hearing people linked to the Human Fertilisation and Embryology Bill on the radio (e.g. BBC Radio Wales, BBC World Service, Radio Cardiff), TV (e.g. BBC See Hear, Channel 4 V-TV, CNN news, ITV Look North) and the PI was also interviewed for articles published on issues about deafness (e.g. Nature, Telegraph, Western Mail).

**Technical, clinical and/or economic potential for exploitation of the project results and outcome**

There is no obvious potential to exploit the project findings in an economic or technical sense. However, the project findings can definitely be used to improve clinical services for deaf and hearing patients.

**11. Future research**

Please describe future work which arises from this project. This could include further basic research, clinical research, health services research, clinical application, consumer research etc.

Deaf adults, who use sign language, face barriers in accessing genetic counselling services. Our next grant proposal, submitted to the NIHR Health Services Research programme (HSR001), intends to explore effective methods for addressing such barriers. This new project, working with additional partners, examines whether the use of genetics services can be increased by the implementation of an education programme about genetic counselling and a specialist genetic counselling clinic. The new, innovative clinic (which will be the first in the UK) will be based at Great Ormond Street Hospital as part of the NE Thames Regional Genetics Service and will be for Deaf adults to discuss any genetic counselling issue, to include those not necessarily related to deafness. The next project will also link up with DCAL (Deafness, Cognition and Language) Research Centre at UCL, an internationally recognised research centre involving Deaf and hearing academics. The goal of this proposal is to build on the existing research done for the Department of Health by our team and address the access issues identified in our previous research. The ultimate aim is to provide evidence to improve health care delivery of genetic counselling services for Deaf people.

**Finances**

To be sent by the Research and Commercial Division (RACD) office at Cardiff University

Signed:…….
Dr Anna Middleton, Chief/Principal Investigator

Date: 31/12/09