



**Ethnic Monitoring in Clinical Genetics
Project Report
July 2003**

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Forward

As the UK alliance of charities and support groups for individuals and families affected by or at risk from genetic disorders, the Genetic Interest Group (GIG) represents the point of view of all sections of our community. Whether you are affected by a condition which is caused by a mutation in a single gene or at risk from one where genetic and environmental factors interact in complex ways, and whichever group in Britain's multi-ethnic, multi-cultural society you identify with, you need to know that the genetic advice you receive from the NHS is scientifically accurate and delivered in a way which is sensitive to your needs and which comes to you in a form you can use. Indeed, a commitment to equity in practice that this would imply is a fundamental premise on which the NHS was founded.

Whilst few would argue with the rhetoric of equity, as the research highlighted in this report indicates, we are some way from achieving this in practice. There are significant varieties across the UK and between different minority groups. Systems and procedures need to be developed to improve access and to maintain the uptake of services by all who need them, and training schemes introduced for staff if the NHS is to be confident that the interactions between relevant genetic factors, cultural norms and societal expectations are to be taken into account, and services provided that are of high quality, timely, accessible and appropriate as a result.

Alastair Kent
Director

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Finally, I would like to thank the Department of Health for funding this project.

1. Project overview, findings and recommendations

1.1 Project aims

- 1) To explore and identify issues around ethnic monitoring within a clinical genetics context
- 2) To assess the feasibility of ethnic data collection within clinical genetics departments
- 3) To pilot Department of Health (DH) ethnic categories, refine them in the light of the results of the pilot processes and thus develop an ethnic-monitoring framework relevant to clinical genetics.
- 5) To liaise with other professionals to ensure that the system is practical and wide ranging in its scope
- 6) To produce reports and other forms of dissemination that will ensure uptake and adoption of the systems thus developed.

This project was funded by the Department of Health for a period of two years, commencing on the 1st July 2001.

1.2 The report

This report is intended to give an overview of the study findings and is divided into 6 main chapters. This chapter presents the main aims of the project, the summary and project findings/outcomes and recommendations. The second explores issues around ethnicity and ethnic monitoring within a clinical genetics context. The third chapter reviews survey findings of the regional genetics centres (RGC-s) and reports on the stakeholder consultation (Workshop on 'Ethnicity Profiling in Clinical Genetics'). The fourth provides the background and results of the multi-centre pilot study, the aim of which was to test the feasibility and range of ethnic data collection within clinical genetics and pilot the Department of Health ethnic categories. The fifth and sixth chapters discuss a proposed ethnic monitoring framework developed for clinical genetics departments, with guidance and recommendations on how to implement it.

1.3 Summary

A multi-ethnic society creates a challenge for genetics services. Population genetics, genetic epidemiology, culture, religion and the social structure of individual communities all play significant parts in defining a persons' particular health needs.

Ethnic monitoring is an essential mechanism for ensuring equity of access to all sections of society, and in identifying and addressing variation across ethnic groups in the uptake of services. Although it has been introduced by the NHS it is not routinely carried out in most clinical genetics centres. This is

partly due to attitudinal problems, and partly because the standard categories used to record ethnicity, are not clinically useful.

This 2-year study funded by the DH was undertaken to examine issues relating to ethnic monitoring within clinical genetics, assess the feasibility and range of ethnic data collection, and pilot and refine DH ethnic categories with a final aim of formulating an ethnic category framework that is useful in terms of clinical management, whilst still maintaining consistency with the DH ethnic categories. The creation of an appropriate framework and relevant supporting materials will help in the planning and delivery of genetic services to a multiethnic society and as a result improve uptake of genetic services among minority ethnic communities.

Research methods were literature survey, surveys of regional genetics centres, consultation with relevant stakeholders (Ethnicity Profiling in Clinical Genetics Workshop), and a multi-centre pilot study.

1.4 The main findings and outcomes of this project were as follows:

- Ethnic monitoring is still a seriously neglected component of genetic service delivery; survey findings show that only 5/24 centres routinely collect ethnicity data on patients. However, as a result of this project, there is now a strong and growing consensus amongst stakeholder groups to address this shortfall.
- The feasibility of gathering both client and parental ethnic origin data within clinical genetics departments has been clearly demonstrated through this project.
- The DH ethnic category framework has been reviewed, piloted and refined in light of these processes. The final template presented in chapter 5 has thus been specifically developed for use within clinical genetics departments. It is based on the DH framework, but also includes certain other clinically relevant categories that are not found within the original classification. The report argues that this framework will be more clinically useful whilst still maintaining consistency with the DH ethnic categories.
- The project has highlighted a significant level of variability in staff member's skill and attitudes towards ethnic data collection, thus emphasising the need for staff training in this regard. The results of the pilot study also clearly suggest that barriers to ethnic data collection exist more strongly within the minds of the health professionals than in the clients' unwillingness to provide such information.
- During the pilot period there was seemingly proportionate uptake of services from the main ethnic groups characterised in this study ('*White*', '*Mixed*', '*Black*', '*Asian*' and '*Other*'). Analysis of cancer referrals across these groups showed that there was significant under-representation across the minority groups ('*Mixed*', '*Black*', '*Asian*' and '*Other*'). It suggests that although the rate of service uptake may be proportional across the five main ethnic groups, access by diagnosis is not. Further

investigation of this result will be required, however it could suggest that there are a large numbers of patients with genetic disorders not being diagnosed.

- 6 % of the total number of patients/clients sampled during the pilot study indicated that their preferred language during the consultation was not English. This statistic translates into 24% when given as a percentage of the total minority ethnic groups (all groups except '*White British*'/ '*White English*') seen during the study period. This figure suggests that at least 24% of minority ethnic groups seen in clinic might be in need of interpreter services.

1.5 Recommendations

1) Every genetics centre should gather client ethnic origin data. This project also recommends that centres simultaneously record client and parental ethnic origins details, as this information will lead to the collection of higher quality data.

2) Centres should begin by piloting the template framework for a period of 1-2 months. Material to assist with this will be available from GIG. Centres may then wish to make subsequent changes to the framework to reflect local population variations.

3) The report recommends that this data be gathered during a face-to-face consultation, as part of a wider information gathering process, such as when family history pedigree is being collected. Exactly which point in the patient pathway will most likely be determined locally, however centres must require a high level of recording and that the responsibility be spread across health care groups in genetics.

4) It is the responsibility of every RGC to ensure that all staff groups are trained in obtaining such data. This report provides background material to support such training, however guidance material is also available from the Department of Health website. Some of the key issues are around understanding the different concepts of ethnicity, allaying some of the sensitivity around the gathering of such information, and in communicating why it is necessary to collect such data.

St Mary's RGC in Manchester has already expressed an interest in piloting the template ethnic framework. We hope that other centres will follow suit after publication of this report.

This work will be discussed during a platform presentation at 2003 Annual British Society Human Genetics (BSHG) conference.

GIG will also be seeking further funding to support the implementation of this ethnic framework and for providing ongoing support. We welcome the commitment in the White Paper¹ to equity and equality in access (section 5.17).

2. Background

British society is made up of many ethnic groups. The 2001 National census revealed that 12.5% of the population of *England and Wales* did not classify themselves as 'White British'. This subset of the population includes 'White Irish' (1.2%) and 'White Other' sub-group as well as all the other subgroups normally classified as minority ethnic groups. These other groups form 9% of the population of *England*. (National statistics Online – census 2001)

A multi-ethnic society creates a challenge for genetic services, its diversity generating a complex array of biological and sociological possibilities. Apart from population genetics and genetic epidemiology, history, demography, and the social structure of individual populations, all play a part in defining an individual's particular health needs.

2.1 Why Monitor Ethnicity?

Ethnic monitoring of service users is essential for a number of reasons.

- Enabling equity of access to all sections of the community,
- Identifying variations in uptake of services across different ethnic groups,
- Determining risk, improving and informing the clinical management of patients and in gathering data on genetic epidemiology.
- Identifying areas for service improvement or development of appropriate services.

Although the NHS has recorded ethnic data on inpatients since April 1995, ethnic monitoring is still not routinely carried out in most Regional Genetics Centres (RGC-s). A recent survey showed that only 5/24 RGC-s currently monitor the ethnicity of their patients and only 3/5 keep electronic records of such information (Mehta P. unpublished data).

This shortfall is partly because the previous ethnic categories (based on the 1991 national census) have been too broad, with limited use value in terms of health service provision and patient clinical management. The familial nature of genetics means that finer cultural and ethnographic distinctions must be drawn, because these may have a bearing on clinical advice that is given. Apprehension on the part of the health care professional might have also resulted in reduced ethnicity recording amongst RGC-s. Of equal or greater significance however, is that the DH does not currently sponsor the collection of outpatient ethnicity data; there are therefore no national standards or guidelines across regional genetic services. The Department has however said that should the NHS wish to implement such a change, it would be supported.

2.2 Ethnicity and Clinical Genetics

One of the key aims of this project is to develop an ethnic monitoring framework for genetics services that is clinically useful. Here the report considers some of the background issues leading to its development, including a brief discussion around the meaning of ethnicity and how it

impacts on clinical genetics. Through these discussions the report attempts to define which ethnicity variables might be most relevant in this context. The last section introduces the 2001 national census categories, as any system that is developed must map back to these census categories.

2.21 The conceptual basis of ethnicity

- Ethnicity is a complex, multi-dimensional, variable term encompassing language, history, culture, upbringing, religion, nationality, geographical and ancestral origins and place. There is a clear need to define its precise meaning in the context of clinical genetics.
- 'Ethnic questions' have utilised a range of terms to define ethnicity, each with an underlying conceptual base e.g. ethnic group, ethnic origin, and ancestry.
- There is a conceptual distinction between human genetic variation and variation based on socially defined measures of ethnicity like language, culture and religion. However ethnicity also encompasses variables of biological importance such as geographical origin that are amongst the forces that drive human genetic variation.

2.22 The impact of ethnicity on clinical genetics

- *The clinical perspective – genetics and disease*

Any population can be regarded as a microcosm of the global population, carrying with it 85% of total genetic variation^{2,3}. Population specific differences are rare representing only a small fraction of the global genetic diversity. However such differences are significant and can lead to variation in susceptibility to disease and pharmacogenetic response. Indeed, these are the suggested findings of two recent studies on single base variants in DNA, Single Nucleotide Polymorphisms (SNP-s) that show that SNP-s most likely to influence disease/pharmacogenetic response are found at a lower frequency than silent substitutions and tend to cluster within specific subpopulations^{4,5}. Certainly, populations do exhibit diversity in the types of genetic diseases that are expressed. Classic examples include Thalassaemia, Sickle Cell Disease, and Tay Sachs Disease, which are respectively more common in people of Asian, African and Ashkenazi Jewish descent. Heterogeneity is also observed at the molecular level, in the mutations associated with a given genetic disease. Post genomic studies are progressively identifying more allelic variants, and it is not surprising to find that they often cluster within different subpopulations.

A population-based approach will become increasingly important in medicine and clinical genetics. Such an approach is essential for a better understanding of genetic and molecular epidemiology and for the development and evaluation of molecular diagnostics. In the future gene tests and drugs may be designed to take account of a patient's ethnic background. This leads to the question of how ethnicity should be monitored in the context of clinical genetics. Which type of ethnicity data is relevant to clinical genetics and how should such data be measured and collected?

- *The social perspective – the impact of culture, religion and other sociological factors*

Culture and religion play an important role in determining to what extent and how members of different ethnic communities express their needs for genetic services and the way in which those services respond in providing culturally competent care. Specific inter-relationships between genetics, culture and kinship patterns need to be appreciated, including issues raised by consanguinity and endogamy.

- *Language and communication.*

During genetic counseling there is a need for clear, accurate communication of genetic information. It is implicit that this process occurs in a language that is understood by the service user. Significant problems arise when the user is not fully conversant with the English language. There is a need to improve equity of access through;

- Provision of multiethnic staff speaking some of the key languages,
- Access to interpreters and translation services, and
- Patient advocacy outreach programmes.

2.3 Which ethnicity data should be recorded in the context of clinical genetics?

The discussions above show that the interplay between genetics and ethnicity is a complex one. Besides population genetics and genetic epidemiology, knowledge of culture, religion and the social structure of individual communities also contribute significantly to a better understanding and appreciation of their health needs.

The following variables are likely to be most relevant for clinical genetics.

- *Ethnic origin or family ethnic origin*
- *Where applicable, membership of specific sub-communities, especially important for endogamous communities*
- *Consanguinity*
- *Religion born into as well as current religion*
- *Preferred language if not English*

2.4 2001 National census/ DH ethnic categories

The newly revised 16-category ethnic group question set for the England and Wales 2001 national census forms the basis of the DH national mandatory standard for the collection of ethnicity data. These categories are divided into 5 main ethnic sub-groups (*'White', 'Mixed', 'Asian', 'Black, Other'*) and are listed in **annex 1**. The DH have also included a 17th category "not stated" to cover situations where information is genuinely lacking for some reason, including "data refused" or data not asked for". A more detailed DH

framework consisting of 62 categories is listed in **annex 2**. The DH have stated that local and national ethnic data collection can be more detailed than the minimum standard provided that the resulting data can be grouped consistently with the 16+1 main categories.

2.5 The development of an ethnic framework for clinical genetics

An ethnic framework developed for genetic services must have clinical relevance whilst still maintaining consistency with DH/census ethnic categories.

Both the national census minimum standard and the detailed DH ethnic framework provide a basis on which to design such a classification. However there are some important criticisms of these classifications that must be considered. Firstly, categories based on skin colour have limited value in the context of health. Secondly, certain clinically relevant groups are absent or invisible such as *Arab, Middle-eastern, Mediterranean, Southern European, and Jewish*. These points illustrate that the framework will need to be evaluated and additional categories considered. A separate section indicating membership of specific sub-communities (e.g. *Mirpur*) might also be necessary.

The development of an ethnic framework also requires consideration of other factors such as wording and structure of the ethnic question, and the method of assignment. There are also some overall requirements, such as the need for simplicity, reliability and acceptability to the workforce⁶.

3. Survey of ethnic monitoring across clinical genetics departments and Report on stakeholders' consultation

3.1 Survey results

At least 14 of the 24 Regional Genetics Centres (RGC-s) across the UK serve regions with a high prevalence of minority ethnic populations. In order to assess how many centres routinely gather ethnicity data, a questionnaire was sent to each one. In summary, only five centres routinely record the ethnicity of their patients, and only three collect such data electronically. One centre routinely records data on religion and most centres keep information on consanguinity within the patient's notes.

A second survey attempted to examine whether, ethnic background/family ethnic origins details were discussed as part of the clinical consultation, to inform the clinical management of the patient. Interestingly, the majority of RGC-s (17/18) do discuss such details when it is considered relevant, and these are recorded within the patient notes.

These findings confirm that although ethnicity data is not routinely gathered, such information is sometimes sought during clinical practice. This indicates that a strong context might already be set for the development of a national framework for recording clinically relevant ethnic data across all RGC-s.

3.2 Stakeholder Consultation – 'Ethnicity Profiling in Clinical Genetics Workshop' April 2002

This workshop was held to consult with professional groups over some of the key issues outlined in the previous chapter and develop a consensus approach to working in the future. The full report can be downloaded from the GIG website (www.gig.org.uk). Web statistics show that this report has been downloaded 1561 times (to date; 10 July 2003) since it was first posted early in 2003.

3.21 The main aims of the workshop were as follows;

1. To understand the principals and objectives of gathering ethnicity data within the context of Genetic Services.
2. To examine how ethnicity data is currently collected across Regional Genetics Centres (RGC-s).
3. To discuss proposals towards the development of an ethnic question for Clinical Genetics, and consider what other kinds of ethnicity data should be gathered within this context.

4. To achieve consensus amongst professional groups on how best to improve current practice across RGC-s and develop strategies for future development with respect to ethnic monitoring.
5. To foster partnerships and collaborations to take this work forward.

This event brought together genetic consultants, counsellors, and specialist nurses from 9 RGC-s (apologies from 2 centres), haemoglobinopathy counsellors, a member of the haemoglobinopathy steering committee and researchers involved in health service and ethnicity and health research. The group reflected a wide range of specialist knowledge, experience and insight and represented a spectrum of ethnic backgrounds.

3.22 The main points of the discussion were as follows:

The first issue raised was the need to be clear about the objectives of gathering ethnicity data for genetic services.

3 main purposes for gathering ethnicity data were highlighted:

- Monitoring and addressing variation in uptake of/access to genetics services across different ethnic groups;
- Development and provision of appropriate services; and,
- To inform the clinical management of the patient (evaluating risk, determining appropriate molecular diagnostic test and gathering data on genetic epidemiology)

There was a broad consensus amongst the group that collection of ethnicity data across RGC-s is essential for the development and delivery of appropriate genetic services, in monitoring service usage, and in addressing variation in access. There was also agreement across the group that these objectives could be achieved using minimum ethnic data sets.

In relation to the clinical management of the patient, the group agreed that more detailed ethnicity data might be required, (e.g. finer demographic detail, membership of specific sub-communities etc).

The group agreed that the minimum ethnic categories should be based upon the DH/ national census 16 (+1) ethnic categories, and that the more detailed DH list could be used as a baseline for more detailed data collection. However the group agreed that certain clinically relevant groups were absent or not clearly represented in these lists and would need to be considered.

In relation to *how* and *when do you ask* the ethnic question, the group highlighted that such information could be gathered in various ways and at a number of different stages during the clinical consultation episode. The group therefore concluded that there was a strong need to develop a standard protocol for clinical practice.

The group discussed which other types of ethnicity data should be routinely collected in the context of clinical genetics.

- With regard to **religion data**, two main questions arose: 1) whether you need to know a person's religion in the context of genetic service delivery, and 2) whether you need to record it? With reference to the first question, the group agreed that there should be greater awareness of different religious viewpoints. Certain members also argued that when dealing with sensitive issues such as prenatal diagnosis and termination it is necessary to be aware of a person's religion, as this may have a bearing on the advice that is given, or on the way in which such advice is delivered. However, other members vehemently opposed this idea and argued that knowledge of a person's religion should not have a bearing on the advice that is given. They felt that all options should be offered to every individual, irrespective of religion, allowing them to make informed decisions.

The group did not reach a consensus over whether to record religion, as approximately half were in favour and half were opposed. Those in favour argued that religion should be recorded as it helps to define particular groups in more detail, and thus distinguish between groups that would otherwise be invisible. They argued that if religion is not recorded resources allocated to that particular group might be less than justified. Those against the recording of religion argued that it is a further act of intrusion into a person's personal life and does not, or should not have any bearing on the clinical advice that is given.

- There was a general consensus amongst the group that **consanguinity** data should be routinely collected across RGC-s.
- With regard to **preferred language** data, many group members argued that this information is necessary in defining the needs of the population and in delivering appropriate services.

In principal the group agreed that, it is necessary to gather different types of ethnicity data across RGC-s. However, in light of the fact that only 5 RGC-s currently record the ethnicity of their patients, the primary objective should be in getting this standardised across all RGC-s. Then over a period of time each centre could begin to develop more detailed recording systems.

3.23 Summary and recommendations of the workshop

The group agreed that there was an urgent need to improve upon current practice in RGC-s. The group thought that this should begin by the recording of minimum ethnic data sets, which could initially be based upon the DH ethnic categories. However the group agreed that this framework would need extending to include clinically relevant groups. The group felt that there was a strong need to establish a national standard for collection of ethnicity data across all RGC-s.

Present:

Mrs Asfa Ahmed, Genetic Associate, B'ham Womens Hospital
Mr Mushtaq Ahmed, Genetic Counsellor, St James Hospital, Leeds
Dr Peter J Aspinal, Senior Research Analyst, Centre for Health Services Studies, University of Kent at Canterbury
Dr Margaret Barrow, Consultant Clinical Geneticist, Leicester Royal Infirmary
Ms Alyson Bradbury, Clinical Nurse Specialist in Genetics, North Trent Clinical Genetics
Ms Tara Clancy, Genetic Counsellor, St Marys Hospital, Manchester
Dr Aamra Darr, Sociologist, Open University
Dr Simon Dyson, Director TASC Unit De Montfort University, Leicester, Lead Researcher in 'the development of an appropriate ethnic question in relation to antenatal screening' for haemoglobin disorders
Ms Lucille Fifield, Haemoglobinopathy Councillor, Sickle & Thalassaemia Centre Charnwood Health Centre Leicester
Ms Carol Giblin, Genetic Counsellor, St Marys Hospital, Manchester
Dr Ian Hopkinson, Senior Lecturer, Primary Care, Genetics Medicine, ICH, UCL Oxford
Ms Vanita Jivangi, Haemoglobinopathy Councillor, Sickle & Thalassaemia Centre Charnwood Health Centre Leicester
Ms Gulshan Karbani, Genetic Counsellor St James Hospital, Leeds, Member of Haemoglobinopathy steering group
Mr Alastair Kent, Director, Genetic Interest Group, London
Dr Carole McKeown, Consultant Clinical Geneticist, B'ham Womens Hospital
Dr Pritti Mehta, Action Researcher, Genetic Interest Group, London
Mrs Shanta Patel, Multilingual Specialist Genetic Nurse, Leicester Royal Infirmary
Ms Nicola Phelan, Clinical Nurse Specialist in Genetics, Northampton General Hospital
Mrs Margaret Ponder, Chair, Genetic Interest Group, London
Mrs Sherlene D Rudder, Genetic Nurse Counsellor, Northwick Park Hospital
Dr A K Saggar, Consultant in Clinical Genetics, St Georges Medical School, London
Ms Jayne Shakespeare, Genetic Counsellor, City Hospital, Nottingham

Apologies:

Professor Elizabeth Anionwu Head of the Mary Seacole Centre for Nursing Practice, Thames Valley University
Mrs Judy Holroyd Genetic Counsellor, University Hospital of Wales, Cardiff
Dr Jane Hurst, Consultant Clinical Geneticist, Radcliffe Hospital
Mrs Susan Lewis Executive Director, Gauchers Association
Professor B Modell Emeritus Professor of Community Genetics, UCL

4. Pilot study to assess the feasibility and range of ethnic data collection required for clinical genetics.

One of the principle outcomes of the workshop was in establishing greater links between GIG and different RGC-s. It enabled this multi-centre pilot study to take place, which assessed the feasibility and range of ethnic data collection required for clinical genetics. The following chapter provides an overview of the methodology, results and discussion.

4.1 Primary aims of study

- To assess the feasibility of ethnicity data collection within clinical genetics.
- To identify if the new DH categories are useful in the context of clinical genetics.
- To record the number of patients of different ethnic origins that use clinical genetic services across three different centres.
- To identify common service users preferred language(s) for the consultation.

Secondary aims

- To test the feasibility of gathering parental ethnic origin details from service users and to examine whether the information obtained is consistent with service user's own ethnic origin choices.
- To link ethnic origin data with medical history information and highlight any pattern that emerge.

The research objectives were to provide information and analyses that might contribute to the development of an ethnic monitoring framework across RGC-s, which is workable and useful in the context of service provision and patient management, whilst still maintaining consistency with DH requirements.

4.2 Methodology

This pilot was a prospective, multi-centred study involving South -West Thames Regional Genetics Service (centre 1), North -West Thames Regional Genetics Service (centre 2), and Leicestershire Clinical Genetics Service (centre 3). The study was carried out as an internal audit over a period of 1 month, all data was processed thereafter on site through collaboration with GIG.

Centre Coordinators

Margaret Barrow (Leicestershire Clinical Genetics Service)
Sherlene Rudder (North West Thames Regional Genetics Service)
Anand Sagar (South West Thames Regional Genetics Service)

The main steps were as follows:

1. Ethnic data was recorded on all service users and their birth parents for a period of 1 month.
2. The recording took place during the face-to-face consultation between the genetic consultant/counsellor/nurse (service provider) and service user.
3. The service user's ethnicity was recorded on a pre-printed form (see page 20).
4. Service user's details were anonymised prior to analysis using a PC database, on site (by Pritti Mehta).

Both the standard DH 16(+1) ethnic category framework (**annex 1**) and the detailed DH ethnic framework (**annex 2**) were used in this study. The standard framework consists of 5 main subgroups; '*White*', '*Mixed*', '*Asian*', '*Black*', and '*Other Ethnic groups*'. For the purpose of this study this framework was presented on a light green laminated card. The detailed DH ethnic framework follows a similar structure however contains additional categories within each sub-group. To facilitate reading each subgroup was presented on a separate colour coded laminated card (light blue card corresponded to the '*White*' subgroup; yellow card corresponded to '*Mixed*' subgroup; red card corresponded to '*Asian*' subgroup; bright green corresponded to '*Black*' subgroup; and orange corresponded to '*Other Ethnic Group*'). Each service provider was given a set of these cards. Certain categories had also been duplicated within the detailed framework, so that they appear in more than one subgroup section. E.g. the '*Mixed Asian*' category appeared in both the '*Mixed*' and the '*Asian*' subgroups. Certain other categories that were considered to be of clinical relevance were added to the '*Other Ethnic Group*' subgroup. These are shown below labelled with an asterix symbol

	OTHER ETHNIC GROUPS
81	Chinese
84	Vietnamese
85	Japanese
86	Filipino
99	Malaysian
80	*Arab
82	*Iranian
83	*North African
88	*Jewish Ashkenazi
90	*Jewish Sephardic
89	Any Other Group (please specify)

Prior to every appointment a paper copy of the ethnicity recording form was attached to the front of the hospital notes. After completion it was be returned to the centre co-ordinator.

Ethnic data was in most cases recorded at the start of the consultation. Service users were shown the 16(+1) ethnic category framework and asked to identify the group that best describes their ethnic origin. Service providers might have needed to define ethnic origin at this point. They were told to emphasis that ethnic origin describes where a person or person's family originates from: that it is different to nationality and in most cases refers to a geographical origin. Service providers also gave explanations for why such information was being collected.

Within the 16 ethnic category list, certain groups carried the option to select from more detailed categories e.g. 'Any Other White Background'. If service users opted for one of these categories they were presented with further choices derived from the detailed DH framework (**annex 2**).

If service users selected a category containing the option 'please specify' they were offered the opportunity to define their own category. Service providers would enter the relevant code and/or free response as appropriate, into the ethnicity recording form. This pattern was repeated to obtain service user's parental ethnic origin details.

Proxy reporting was only acceptable for young children, those unable to respond through temporary or permanent incapacity, or those who were not familiar with the English language.

Staff members were told that although the pilot aimed for 100% recording of all service users seen during the study period, there was no obligation on the part of user to complete the form. Staff were advised to record "Not stated" when the service user did not wish to respond and 'Not Known' where it was not possible to collect the data.

Whilst service users were selecting an ethnic origin category, the service provider began to fill in other sections of the ethnicity recording form, such as service user's DOB and diagnosis/reason for referral, in order to save time. Service users were also asked to define their preferred language (or languages where families/couples speak different tongues) during the consultation.

The following details were entered on the form;

- Patient unique identifier number
- DOB
- Diagnosis/reason for referral
- Ethnic origin (service user and service user's birth parents)
- If the service user was unable self -assign his/her ethnic origin and appropriate explanation
- Preferred language(s) during the consultation
- Any additional comments as appropriate

All these details apart from the service user's unique identifier number were entered onto a PC database on site at each participating centre.

Advantages of this pilot

- First known pilot of its kind.
- Provide information quickly.
- Confidential.
- Cheap and minimal interference with maximised recording.
- Possible by the co-operation of the three regional centres and the time/staff input by GIG.

The Ethnic Category Recording Form

Patient number	
DOB	
Diagnosis or reason for referral	
Ethnic origin	
Parental ethnic origin (Birth mother)	
Parental ethnic origin (Birth father)	
If the service user was unable to self-assign his/her ethnicity please indicate the reason why by selecting one of the options.	
<ul style="list-style-type: none">▪ Young child▪ Temporary or permanent incapacity▪ Unfamiliar with the English Language▪ Other – please state	
Service user's preferred language(s) during consultation	
Any further comments	

4.3 Results of the pilot study

During the one-month study period 498 appointments were attended across the three clinics. Ethnic data was gathered from a total of 459 patients/clients, giving an overall recording rate of 92%. Individual recording rates for the three centres were as follows: South West Thames Regional Genetics Service -90%; North West Thames Regional Genetics - 97% and Leicestershire Clinical Genetics Service -70%.

Table 1 shows the ethnic breakdown of the patients/clients recorded during this study. Data sets are provided for each centre and for the three centres combined. Each data set contains the observed frequency of individuals in each ethnic origin category and the expected frequency estimated from 2001 local census data. Chi – square analysis was performed to evaluate whether uptake of services across different ethnic groups was proportionate to the background population frequency of each ethnic group. Owing to limited sample sizes analyses could only be performed on the total combined data set using collapsed ethnic group categories (*'White'*, *'Mixed'*, *'Asian'*, *'Black'*, & *'Other'*). Surprisingly, Chi-square analysis revealed that there was no significant difference between the observed and expected ethnic group frequencies, indicating that uptake of services across the 5 main ethnic groups was proportionate to the background population frequency. However, larger sample sizes would need to be assessed in order to substantiate this conclusion and to evaluate uptake across each ethnic sub-group.

TABLE 1

	North West Thames		South West Thames		Leicester		Total	
	Observed Frequency	Expected Frequency	Observed Frequency	Expected Frequency	Observed Frequency	Expected Frequency	Observed Frequency	Expected Frequency
White	150	161.66	180	174.34	48	47.06	378	383.06
British	103	137.91	140	161.77	48	45.71	291	345.39
Irish	2	7.21	2	3.16	0	0.47	4	10.84
Any Other White background	45	16.54	38	9.41	0	0.84	83	26.79
Mixed	7	5.68	1	3.08	1	0.67	9	9.43
White and Black Caribbean	4	1.55	0	0.74	1	0.27	5	2.56
White and Black African	0	0.81	0	0.37	0	0.05	0	1.23
White and Asian	0	1.75	0	1.11	0	0.21	0	3.07
Any Other Mixed Background	3	1.57	1	0.87	0	0.13	4	2.57
Asian or Asian British	30	27.63	4	7	6	6.33	40	40.96
Indian	10	17.09	2	3.18	6	5.39	18	25.66
Pakistani	14	4.69	2	1.62	0	0.31	16	6.62
Bangladeshi	0	1.73	0	0.52	0	0.2	0	2.45
Any Other Asian Background	6	4.12	0	1.67	0	0.43	6	6.22
Black or Black British	12	12.52	3	3.92	0	0.63	15	17.07
Caribbean	3	5.85	2	1.81	0	0.35	5	8.01
African	3	5.7	1	1.76	0	0.25	4	7.71
Any Other Black Background	6	0.97	0	0.35	0	0.04	6	1.36
Other Ethnic Groups	14	5.56	3	2.67	0	0.34	17	8.57
Chinese	3	2.1	2	1.15	0	0.22	5	3.47
Any Other Ethnic Background	11	3.46	1	1.52	0	0.17	12	5.15
Total	213	213	191	191	55	55	459	459

Table 2 gives the ethnic breakdown of the ‘*Any Other*’ sub-group options of the ‘*White*’, ‘*Mixed*’, ‘*Asian*’, ‘*Black*’, and ‘*Other Ethnic*’ groups. Most sub-categories of the ‘*Any Other*’ group options were derived from the detailed DH framework consisting of approximately 60 different ethnic category choices; apart from ‘*Arab*’, ‘*Iranian*’, and ‘*Jewish Ashkenazi/Sephardic*’ sub-categories, which were added for the purpose of this study. The overall percentage of individuals recorded within an ‘*Any Other*’ sub-group category across the three centres was 24%. Interestingly, the percentage levels between the three centres varied considerably with North West Thames Regional Genetics – 33%; South West Thames Regional Genetics Service – 21%; and Leicestershire Clinical Genetics Service – 0%.

There were a number of additional ethnic categories chosen by clients during the study period, which were not found within the pre-designated category form. These included ‘*Belgian*’, ‘*Portuguese*’, and ‘*Maltese*’, classified as ‘*Other white European*’ (code 37) for the purpose of this study, and ‘*Afghan*’, ‘*Iraqi*’, and ‘*Lebanese*’ and ‘*Brazilian*’ classified within the ‘*Any Other, Other*’ sub-group category (code 89).

Table 2

	Total Frequency	Total %
Any Other White Background	83	18.08
English	49	10.68
Scottish	6	1.31
Welsh	1	0.22
Northern Irish	2	0.44
Turkish	2	0.44
Italian	1	0.22
Rep of former USSR	1	0.22
Mixed White	6	1.31
Other White European	8	1.74
Other White	7	1.53
Any Other Mixed Background	4	0.87
Other Mixed	4	0.87
Any Other Asian Background	6	1.31
Punjabi	1	0.22
East African Asian	1	0.22
British Asian	3	0.65
Other Asian	1	0.22
Any Other Black Background	6	1.31
Somali	2	0.44
Black British	2	0.44
Other Black	2	0.44
Other Ethnic Background	12	2.61
Arab	2	0.44
North African	1	0.22
Ashkenazi Jew	3	0.65
Iranian	2	0.44
Any Other Group	4	0.87
Total	111	24.18

Table 3 shows the ethnic breakdown of all the cancer referrals seen during the study period. Chi – square analysis was used to evaluate whether the frequency of cancer referrals across different ethnic groups was proportionate to the background population frequency of each ethnic group. Again, because sample sizes were small Chi – square analyses could only be performed on the total combined data set for the three centres using collapsed ethnic categories. Data from the '*Mixed*', '*Asian*', '*Black*' & '*Other Ethnic Group*' categories was grouped together and compared with the *White* ethnic background data. These analyses showed that there was a significant under-representation of cancer referrals from minority ethnic groups, indeed only 6/110 cancer referrals were from minority ethnic groups.

Table 3

	North West Thames		South West Thames		Leicester		Total	
	Observed cancer referrals	Expected cancer referrals	Observed cancer referrals	Expected cancer referrals	Observed cancer referrals	Expected cancer referrals	Observed cancer referrals	Expected cancer referrals
White	26	22	63	60	15	12.825	104	94.825
British	12		50		15			
Irish	2		1		0			
Any Other White background	12		12		0			
Mixed	1	0.754	0	1.056	0	0.18	1	1.99
White and Black Caribbean	1		0		0			
White and Black African	0		0		0			
White and Asian	0		0		0			
Any Other Mixed Background	0		0		0			
Asian or Asian British	0	3.77	0	2.44	0	1.725	0	7.935
Indian	0		0		0			
Pakistani	0		0		0			
Bangladeshi	0		0		0			
Any Other Asian Background	0		0		0			
Black or Black British	0	1.71	1	1.386	0	0.18	1	3.276
Caribbean	0		1		0			
African	0		0		0			
Any Other Black Background	0		0		0			
Other Ethnic Groups	2	0.754	2	0.924	0	0.09	4	1.768
Chinese	0		1		0			
Any Other Ethnic Background	2		1		0			
Total	29	29	66	66	15	15	110	110

Parental ethnic origin data was obtained from 98% of individuals recorded during this study period. That is, 451/459 individuals provided both their own and their parental ethnic origin details. In the remaining 8 cases the client was not aware of their parental ethnic origin(s). This high level of recording indicates the relative feasibility of obtaining and recording such information in a health care setting.

Recording of parental ethnic origin details was not only useful in identifying more complex or mixed ethnicities, but it also allowed us to check the validity of our data by checking for correspondence between clients reported ethnic origin details and what clients reported as their parental ethnic origin(s).

Across the three centres, 430/451 clients gave parental ethnic origin details that were consistent with their own reported ethnic origins types. Of those remaining, 20 reported as '*White British*' ethnic origin, and 1 reported as '*White English*' ethnic origin, gave parental ethnic origin details that were inconsistent their own ethnic origin types. Assessment of the parental data revealed *Polish, Czech, Italian, White European and also Indian, Russian, Asian, and Mauritius* ancestry within this group. A significant proportion (6/20) also reported Jewish ancestry.

Table 4 shows the breakdown of 'preferred language' data taken across the three centers. 6 % of the total number of patients/clients sampled during the pilot study indicated that their preferred language during the consultation was not English. This statistic translates into 24% when given as a percentage of the total minority ethnic groups (all groups except '*White British*' or '*White English*') seen during this study period. This figure suggests that at least 24% of minority ethnic groups seen in clinic maybe in need of interpreter services. This figure is likely to be higher as those in greatest need are not likely to access the service.

Table 4

Preferred Language	Total Frequency
English	406
Arabic	1
Dari	1
Farsi	2
Punjabi	6
Urdu	5
Gujarati	2
Somali	2
Portugese	2
Spanish	2
Turkish	1
BSL	2
language N/A	3
language not recorded	26
Total	461

4.4 Staff interview finding

After completing the pilot study clinical staff were asked if they would take part in a telephone interview to survey their attitudes and opinions around ethnic data collection. All staff that were interviewed had been involved in the face-to-face collection of ethnic data during the pilot study. A total of 20 Interviews were conducted comprising 6 from Leicester, 6 from South West Thames RGC and 8 from North West Thames RGC. A total of 12 clinical geneticists, 4 genetic counsellors and 4 clinical genetic nurse specialists were interviewed and sessions lasted approximately 10 -15mins. The following provides a summary of the questions and responses.

1) Do you think it is useful to collect ethnicity data in the context of clinical practice? YES/NO

Almost all staff respondents thought it was useful to collect ethnicity data in the context of clinical practice. The majority thought it was useful in terms of service provision, service development, and equity of access. A few remarked upon its role in improving diagnosis, mapping disease prevalence, genetic epidemiology, and pharmacogenetics. However, one individual felt that although such data was useful, staff should not be obliged to collect it. Another expressed the opinion that ethnic data collection was no more than a paper filling exercise, had no function and that the DH categories were totally inappropriate.

Number of respondent that answered YES to this question

Leicester: 6/6,
North West Thames: 8/8
South West Thames: 4/6.

Respondents' comments:

Leicester

- *Useful for assessing peoples needs*
- *Useful in assessing equity of service provision*
- *Useful for looking at disease prevalence in different ethnic groups*
- *Useful for counselling purposes and in providing more culturally competent service*
- *Useful to ascertain difference in uptake amongst different minority groups*
- *Useful in recognising significant under-ascertainment of ethnic minorities families attending cancer clinics*
- *Useful but should include more language information*

South West Thames

- *Yes very important. Not all diseases manifest the same – genetic epidemiology, pharmacogenetics. From clinicians point of view awareness of phenotype is important.*
- *In clinical practice it is useful, but it is collected anyway. We shouldn't be obliged to collect it.*

- *Yes although concerned that categorises might not be representative*
- *Some clinical situations where such information is useful, however don't understand why it should be collected. The categories are inconsistent or meaningless*
- *Paper filling exercise – no function as far as clinic was concerned, hope ethnic data collection will not be introduced, find DH categories totally inappropriate*

North West Thames

- *Useful as long as data is going to be used*
- *Useful for service provision as you need to know what the population consists of.*
- *It Improves diagnosis*
- *Adds to clinical information*
- *Yes in order to provide a better service for the different groups.*

2) Do you think that the clinical consultation period is an appropriate time to gather ethnic origin data? YES/NO

All respondents from North West Thames and the majority from South West Thames thought that the clinical consultation period was an appropriate time to gather ethnic origin data. In contrast, most respondents from Leicester considered this to be an inappropriate time.

Number of respondent that answered YES to this question

Leicester: 2/6
 North West Thames: 8/8
 South West Thames: 4/6.

Respondents' comments:

Leicester

- *Usually okay, one or two situations when it is not*
- *Data should be collected outside the consultation*
- *Should be part of a wider information gathering process*
- *Should be filled in when patients come to clinic*
- *Although it is important, there is a time and a place for it*
- *It should be incorporated into a pre-clinic questionnaire*
- *Ethnic data should not be treated in isolation*

South West Thames

- *Slightly tricky although not sure when else would be appropriate*
- *Ethnic origin needs to be face-to-face; ethnic group form filled in on their own*
- *When dealing with difficult issues found it intrusive*

North West Thames

- *Any other time might be an inconvenience for client and department*

- *General practice to ask ethnic question – specific mutations*
- *Add on – general information gathering – family history*
- *Asking for relevant information – usually ask anyway*
- *It is the right time unless you have a very detailed pre - appointment questionnaire that goes out*

3) Did you find it useful to have more detailed ethnic categories available? YES/NO

The majority of respondents from North West Thames and South West Thames found the detailed ethnic categories useful, whilst in contrast, most of the respondents from Leicester did not.

Number of respondent that answered YES to this question

Leicester: **1/6**
 South West Thames: **4/6**
 North West Thames: **7/8**

Interestingly, the numbers of clients recorded at each centre that belonged to a category derived from the detailed framework varied accordingly; these results are shown below as a proportion of the total number of patients recorded.

Leicester: **0/55**.
 South West Thames: **37/191**
 North West Thames: **64/213**,

Respondents' comments:

Leicester

- *Yes if mixed or not 'White British'*
- *Mind blowing – too much information*

South West Thames

- *Yes although they didn't cover everything*
- *For genetics it's important- need some idea of genetic origin which can be obtained through ethnicity*
- *No they were the wrong ones, weren't detailed enough; need to know which village people are from.*
- *Yes as a back up although not always needed.*
- *Yes because patients did not want to be put in broad categories. Such are not clinically useful.*
- *No because all patients were of straightforward origin.*

North West Thames

- *Showed people that you were trying to be precise*
- *It was confusing sometimes*

- *For certain tests important to know ethnic backgrounds (Cistic Fibrosis and Breast Cancer)*
- *Didn't feel it enhanced the consultation*
- *There were Design issues – bit cumbersome – but useful sometimes*
- *All the categories should be presented on one sheet of A4*
- *It is important for more specific Jewish queries*
- *It is important in determining more complicated backgrounds*

4) Did you ever experience any personal difficulties in gathering ethnic data form patients? YES/NO

Whilst the figures below suggest that the majority of staff did not experience any personal difficulties in gathering ethnic data, some respondents commented that they felt uncomfortable about it when they were breaking bad news or when they were dealing with sensitive matters.

Number of respondent that answered YES to this question;

Leicester: **1/6**

South West Thames: **1/6**

North West Thames: **1/8**

Respondents' comments:

Leicester

- *In every case I was slightly hesitant*
- *No unless I thought it was inappropriate*

South West Thames

- *Forgetfulness was my biggest problem*
- *Yes if you thought it was going to affect the consultation –such as when dealing with sensitive issues*

North West Thames

- *Giving an explanation allowed you to overcome any difficulties*
- *We are used to doing it*
- *I was unclear about who to fill in information about.*

5) Did you ever feel that you could not obtain ethnicity information from a patient? YES/NO

The majority of Leicester staff thought that they could not obtain ethnicity information in certain situations e.g. when dealing with very sensitive cases or when breaking bad news. Although the figures for North West Thames and South West Thames suggest that the majority of staff did not have the same

experience, the comments do suggest that some individuals might have felt unable to in certain circumstances.

Number of respondent that answered YES to this question

Leicester: **5/6**

South West Thames: **2/6**

North West Thames: **2/8**

Respondents' comments

Leicester

- *Felt it was inappropriate because of sensitivity around the counselling*
- *You didn't ask if you felt that the situation was too sensitive*
- *I couldn't obtain it in situations where patients were receiving bad news, or once the counselling had started*
- *I found it difficult in tense emotional situations.*
- *When breaking bad news it seemed very inappropriate to take ethnicity data. It seemed to trivialise things.*

South West Thames

- *If you thought it was going to affect consultation you didn't ask – i.e. when dealing with very sensitive issues.*
- *Yes when dealing with a sick child, or when it felt inappropriate*

North West Thames

- *1-2 times when the consultation was sensitive*
- *Easier to ask at beginning of consultation*
- *Where there was quite a sick new baby- a difficult discussion, I might have just filled the form in myself*

6) Did you ever encounter clients who did not wish to provide ethnicity information?

All staff members stated that during the study period they had not encountered any client, who did not wish to provide ethnicity data. However one of the staff's comments suggests that he or she might have carried out self – selection of clients at certain times.

Respondents' comments:

South West Thames

- *There might be some self-selection*

North West Thames

- *Occasionally people are suspicious but after providing an explanation no problem*
- *Some people wanted further explanations*

7) General comments.

Leicester

- *It wasn't a big burden*
- *Data should be collected outside the consultation*
- *Should be part of a wider information gathering process*
- *Should be filled in when patients come to clinic*
- *Although it is important there is time and a place for it*
- *Should be incorporated into a pre-clinic questionnaire*
- *Ethnic data should not be treated in isolation*
- *Language barrier should also be addressed*
- *Should not try to collect too much data*
- *Language data should be collected*

South West Thames

- *Don't understand why it is needed – final categorises inconsistent or meaningless – would be better to put ethnic origin on the appointment letter on the GP's detail letter.*
- *Need to know why you are collecting it*
- *Ask when it is relevant*
- *Hope it's not introduced*
- *Find DH categories totally inappropriate*
- *Biggest problem is remembering to do it.*

North West Thames

- *Decrease the number of cards*
- *Ask patient first language combined with ethnicity*
- *Easy to use although a little confusing*
- *Easy to do – explanation very comprehensive*
- *Wasn't too cumbersome*

4.5 Discussion

The main issues that emerged from the pilot study were as follows:

- The pilot study clearly demonstrated the feasibility of ethnic data collection within clinical genetics by the high level of recording that was observed in two out of the three genetics centres tested.
- The shortfall, however, observed in the third centre reflects and highlights a significant level of variability across RGC-s in their overall experience in and staff member skills and attitudes around ethnic data collection. This emphasises that staff training will be a necessary component in ensuring uptake of ethnic monitoring across clinical genetics.
- The study showed that it is also possible to gather parental ethnic origin details from service users and that such information can be useful in determining mixed or more complex ancestries. Comparison of parental ethnic origin and service user's own ethnic origin choices also revealed some significant inconsistencies between the two results. For example a significant number of individuals reporting to be of 'White British' ethnic origin were found to have '*Jewish*', '*Eastern European*' or even '*Indian*' ancestry. This highlights how particular groups can be subsumed within the major ethnic group categories, and it emphasises the confusion that can exist around the meaning of ethnic origin and ethnic group.
- The piloted 16 DH ethnic categories might be sufficient for recording crude differences within the population, however, the study showed that significant groups were absent from this framework and that it promoted the labelling of ethnic groups by colour, leading to significant under-ascertainment or mis-representation of particular groups. The classification will need significant refinement to make it clinically relevant. Category choices derived from the wider more detailed ethnic framework (consisting of approximately 60 ethnic categories) were only observed in a small proportion of the total number of recordings. This number might truly reflect the proportion of individuals belonging to those groups, or it may reflect health professionals' reluctance to spend time trying to ascertain more complex ethnic backgrounds. Incidentally, the majority of these recordings were observed in two centres where staff members expressed value in having extra categories for referral. Interestingly, no such recordings were observed at the third centre where the majority of staff found the extra categories to have no added use value.
- The majority of staff that took part in the pilot study thought it was useful to collect ethnicity data during clinical practice. However certain individuals did not, highlighting variability in staff members' attitudes towards the process of ethnic data collection and further emphasising the need for training in this regard. There was also strong variation in whether staff thought it was appropriate to gather such information during the clinical

consultation period. This reflected differences in staff members' perception of whose responsibility it was to collect this information.

The majority of staff indicated that they had experienced personal difficulties in obtaining ethnic origin data at some time during the study, either when breaking bad news or when they felt that they were dealing with sensitive situations.

- **Conversely, during this study not a single member of staff had encountered any client who had refused to provide ethnic origin information. This clearly indicates that barriers to ethnic data collection exist more strongly within the minds of the health professionals than in clients' unwillingness to provide such information.**

- During the study period there was seemingly proportionate uptake of services from the main ethnic groups characterised in this study ('*White*', '*Mixed*', '*Black*', '*Asian*' and '*Other*'). However, analysis of cancer referrals across these groups showed that there was significant under – representation across the minority groups ('*Mixed*', '*Black*', '*Asian*' and '*Other*')

This suggests that although the rate of service uptake may be proportional across the five main ethnic groups, access by diagnosis is not. Further investigation of this result will be required, however it could suggest that there are a large numbers of patients with genetic disorders not being referred.

- 6 % of the total number of patients/clients sampled in this study indicated that their preferred language during the consultation was not English. This statistic translates into 24% when given as a percentage of the total minority ethnic groups (all groups except '*White British*'/ '*White English*') seen during the study period. This figure suggests that at least 24% of minority ethnic groups seen in clinic might be in need of interpreter services, and this figure is likely to be an under- representation, as those individuals in greatest need are not likely to access genetic service and therefore be counted.

5. Proposed ethnic category framework for clinical genetics

5.1 Figure 1 shows the proposed ethnic category framework that this project has developed for use in clinical genetics departments. This framework attempts to identify clinically relevant categories whilst still maintaining consistency with 2001 census/DH ethnic categories. It was developed over the course of this project, reviewed during the stakeholder consultation (ethnicity profiling in clinical genetics workshop) and refined after piloting of DH categories during the multi-centre pilot study.

The structure of this classification follows DH/national census standard framework consisting of 5 main ethnic groups, however the names of these groups have been changed so that they reflect geographical ethnic origins rather than skin colour. 'White' has been replaced with 'Europe', 'Mixed' remains as it is, 'Asian/Asian British' has been replaced with 'South Asian', 'Black/Black British' has been replaced with 'Caribbean/Sub-Saharan Africa', and 'Other Ethnic Group' has been replaced with 'Other Ethnic Backgrounds'. Each group is divided into a set of pre-designated categories with the addition of some open response categories for those who are unable to identify with any of the pre-designated options. Most categories are derived from the DH standard framework or detailed list of DH categories, however certain other additional categories are also presented.

The 'Europe' ('White') subgroup has been expanded in order to identify 'Irish Traveller', 'Gypsy/Romany', 'Jewish', 'Southern European' and 'Mediterranean' groups. The inclusion of 'Irish Traveller' and 'Gypsy/Romany' categories is extremely relevant to clinical genetics due to genetic implications of consanguinity and endogamy that is practised in these groups. The identification of Southern European/Mediterranean groups, and the Jewish group is necessary due to the high frequency of certain ethno-specific diseases within these groups.

The 'Mixed' subgroup contains the option to tick all category boxes that apply, and an open response category for those wishing to use this.

The 'South Asian' group ('Asian or Asian British') is almost identical to that which appears in the national census framework, consisting of 'Indian', 'Pakistani', 'Bangladeshi' categories and an 'Any Other South Asian Background' category for those unable to identify with the pre-designated options, but also contains an 'East African Asian' category. However, because these categories cover vast ethnographic and demographic regions, it might be more relevant to capture particular subgroups, with reference to *region, caste, biraderi, or religion*. This may lead to the identification of specific consanguineous or endogamous communities.

The 'Caribbean/Sub-Saharan Africa' ('Black or Black British') subgroup is also almost identical to that which appears on the national census form, consisting of two main categories, 'Caribbean' and 'African', and an additional open response category, but it also contains a 'Somali' option.

However, the African category is still too broad disguising the level of ethnic and genetic diversity of this group and will therefore have to be reviewed and subdivided accordingly.

The 'Other Ethnic Background' subgroups, consists of 'Chinese' and 'Any Other Ethnic Group' categories as are present within the DH/national census classifications. However there are also some additional categories. These includes 'Japanese' and 'South East Asian' categories to enable these groups to be distinguished from 'Any Other South Asian Backgrounds' that may be identified within the main 'South Asian' subgroup. This group also contains an 'Arab, Iranian, Afghan, North African' category. The identification of some of these groups will be extremely relevant to clinical genetics due to their high frequency of haemoglobinopathies and the practice of customary consanguineous marriage.

Because of its geographical structure this framework will be more useful in terms of clinical management and it will be easy to expand this minimum list if numbers in the local population warrant a separate category to 'Other.'

Figure 2 presents the full set of ethnic origin categories and codes. It is derived from the DH detailed list of ethnic categories (annex 2), but reflects the name changes from skin colour to geographical origin as mentioned above. It also contains additional categories, which are highlighted (*Jewish, Portuguese, Spanish, Arab, Iranian Afghan, N. African*). These categories are absent from the original DH detailed list and therefore have been given new codes.

Please note that this classification is not set in stone, and we would be happy to receive feedback and welcome any improvements towards a universally acceptable form.

Fig 1: The proposed ethnic category framework for clinical genetics

<p>a. Europe (<i>White</i>)</p> <ul style="list-style-type: none"> • British (English, Irish, Scottish, or Welsh) • Gypsy/Romany • Irish Traveller • Jewish • Mediterranean/Southern European e.g. Italian, Greek (<i>please write in box</i>) • Any Other European Background (<i>please write in box</i>) 	<input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/> <input type="text"/> <input type="text"/>
<p>b. Mixed (<i>Mixed</i>)</p> <ul style="list-style-type: none"> • Please tick this box and tick/write in all other boxes that apply to you 	<input type="checkbox"/>
<p>c. South Asia (<i>Asian or Asian British</i>)</p> <ul style="list-style-type: none"> • Indian • Pakistani • Bangladeshi • East African Asian • Any Other South Asian Background (<i>please write in box</i>) 	<input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/> <input type="text"/>
<p>c. Caribbean/Sub-Saharan Africa (<i>Black or Black British</i>)</p> <ul style="list-style-type: none"> • Caribbean • Sub- Saharan Africa • Somali • Any Other Caribbean/Sub-Saharan African Background (<i>please write in box</i>) 	<input type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/> <input type="text"/>
<p>e. Other Ethnic Backgrounds (<i>Other Ethnic Groups</i>)</p> <ul style="list-style-type: none"> • Chinese • Japanese • Any other South East Asian e.g. Malaysian, Filipino (<i>please write</i>) • Arab, Iranian, Afghan, North African (<i>please write in box</i>) • Any Other Ethnic Backgrounds, (<i>please write</i>) 	<input type="checkbox"/> <input type="checkbox"/> <input type="text"/> <input type="text"/> <input type="text"/>

Figure 2 The Detailed Ethnic Origin Codes and Categories

	EUROPE (WHITE GROUP)
01	(British, Mixed British)
02	Irish
03	English
04	Scottish
05	Welsh
38	Northern Irish
06	Cornish
07	Cypriot (part not stated)
08	Greek
09	Greek Cypriot
10	Turkish
11	Turkish Cypriot
12	Italian
30*	Spanish
35*	Portuguese
13	Irish traveller
14	Traveller
15	Gypsy/Romany
38*	Jewish
16	Polish
17	All republics of former USSR
18	Kosovan
19	Albanian
31	Bosnian
32	Croatian
33	Serbian
34	Other republics of former Yugoslavia
36	Mixed European (<i>Mixed White</i>)
37	Other White European, European unspecified, European Mixed

	MIXED GROUP
36	Mixed European (<i>Mixed White</i>)
44	Mixed South Asian
64	Mixed Caribbean/Sub-Saharan Africa (<i>Mixed Black</i>)
21	European and Caribbean (<i>White and Black Caribbean</i>)
22	European and Sub-Saharan African (<i>White and Black African</i>)
23	European and South Asian (<i>White and Asian</i>)
24	Caribbean/Sub-Saharan Africa and Chinese (<i>Black and Chinese</i>)
25	Caribbean/Sub-Saharan Africa and European (<i>Black and White</i>)
26	Chinese and European (<i>Chinese and White</i>)
27	South Asian and Chinese (<i>Asian and Chinese</i>)
28	Other mixed, mixed unspecified

	SOUTH ASIAN (ASIAN or ASIAN BRITISH)
41	Indian or British Indian
42	Pakistani or British Pakistani
43	Bangladeshi or British Bangladeshi
44	Mixed Asian (please specify)
45	Punjabi
46	Kashmiri
47	East African Asian

48	Sri Lankan
49	Tamil
50	Sinhalese
51	British Asian
57	Caribbean Asian
59	Other Asian, Asian unspecified

	CARRIBEAN/SUB-SAHARAN AFRICA (BLACK or BLACK BRITISH GROUP)
61	Caribbean
62	African
63	Somali
64	Mixed Caribbean/Sub-Saharan Africa (<i>Mixed Black</i>)
65	Nigerian
66	Black British
69	Other Caribbean/Sub-Saharan Africa unspecified (<i>Other Black, Black unspecified</i>)

	OTHER ETHNIC BACKGROUNDS
81	Chinese
84	Vietnamese
85	Japanese
86	Filipino
99	Malaysian
80*	Arab
82*	Iranian
83*	North African
88*	Afghan
89	Any Other Background

6. Guidelines on implementation

6.1 Each centre should begin by piloting the new clinical genetics ethnic framework for a period of one month following a method similar to the one outlined in the pilot study.

Below we provide basic guidelines that should be followed by each staff member taking part in the study. (These guidelines are extracted from the information that was provided to each staff member during the pilot study). We advise that each centre identifies a local coordinator who will be able to liaise with GIG over the course of piloting. Through this interaction we also hope to provide support and a point of contact for sharing and learning between different RGC-s.

After piloting this framework different centres may wish to review the ethnic categories and add or remove particular groups as they think necessary.

6.2 Guidelines

The main steps of the pilot are as follows:

- ❑ Ethnic data should be recorded on all service users and (their birth parents) for a period of 1 month. When multiple service users are present, e.g. a couple seeking counselling, ethnic data should if possible be recorded for each service user.
- ❑ The recording should take place during face-to-face consultation between the genetic consultant/counsellor/nurse and service user.
- ❑ The service user's ethnicity will be recorded on a pre-printed form (see attached).
- ❑ Service user's details will be analysed using a PC database, on site.

1) Prior to every appointment a paper copy of the ethnicity recording form will be attached to the front of the hospital notes. After completion it will be returned to the centre co-ordinator.

2) Ethnic data will be recorded at the start of the consultation. Service users will be shown the list of ethnic categories and asked to identify the category that best describes their ethnic origin. Service providers may need to define ethnic origin at this point. They will emphasize that ethnic origin describes where a person or person's family originates from: that it is different to nationality and in most cases refers to a geographical origin. Service providers must also provide explanations for why such information is being collected; e.g. this information is being collected in order to improve genetic services.

3) Certain categories in the list, such as 'Any Other European Background' carry a free text field or the option to select from further categories. If service users opt for one of these categories they should be presented with further

category options. More categories are available for each of the main ethnic groups. Service users may also wish to define their own background. After service users have made their selection, service providers should enter the relevant code and/or free response as appropriate, into the ethnicity recording form. This pattern could be repeated to obtain service user's parental ethnic origin details.

4) Proxy reporting will only be acceptable for young children, those unable to respond through temporary or permanent incapacity, or those who are not familiar with the English language. If staff members have to assign the ethnic origin for any reason, this must be indicated using the options provided.

5) Staff should record "**Not stated**" when the service user does not wish to respond and "**Not known**" when this information is not known to the client or it is not possible to obtain it from the client. There will be no obligation on the part of client to provide ethnic data.

6) Whilst service users are selecting an ethnic origin category, the service provider should begin to fill in other sections of the form, such as service user's DOB and diagnosis/reason for referral, as this might save time.

7) Service users will be asked to define their **preferred language** (or languages where families/couples speak different tongues) during consultation.

The following details will be entered on the form;

- Service user's number
- DOB
- Diagnosis/reason for referral
- Ethnic origin (service user and service user's birth parents-optional)
- If the service user was unable self -assign his/her ethnic origin and appropriate explanation
- Preferred language(s) during the consultation

- Any additional comments as appropriate

The Ethnic Category Recording Form

Patient number	
DOB	
Diagnosis or reason for referral	
Ethnic origin	
Parental ethnic origin (Birth mother)	
Parental ethnic origin (Birth father)	
If the service user was unable to self-assign his/her ethnicity please indicate the reason why by selecting one of the options.	
<ul style="list-style-type: none">▪ Young child▪ Temporary or permanent incapacity▪ Unfamiliar with the English Language▪ Other – please state	
Service user's preferred language(s) during consultation	
Any further comments	

References

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Annex 1 The ethnic categories for use in ALL DH statistical collections (16+1listing)

Ethnic Categories*	Ethnic Classifications
a. White <ul style="list-style-type: none"> • British, • Irish • Any other White background 	01 02 03-19, 31-39
b. Mixed <ul style="list-style-type: none"> • White and Black Caribbean • White and Black African • White and Asian • Any other mixed background 	21 22 23 24-29
c. Asian or Asian British <ul style="list-style-type: none"> • Indian • Pakistani • Bangladeshi • Any other Asian background 	41 42 43 44-51, 57,59
d Black or Black British <ul style="list-style-type: none"> • Caribbean • African • Any other Black Background 	61 62 63-65,66,69
e. Other ethnic Groups <ul style="list-style-type: none"> • Chinese • Any Other Ethnic Group 	81 82-86
f. Not stated	87

Annex 2: the detailed ethnic categories (confirmed by ONS Jan 2001)

Ethnic Code	WHITE GROUP
01	British, Mixed British
02	Irish
03	English
04	Scottish
05	Welsh
38	Northern Irish
06	Cornish
07	Cypriot (part not stated)
08	Greek
09	Greek Cypriot
10	Turkish
11	Turkish Cypriot
12	Italian
13	Irish Traveller
14	Traveller
15	Gypsy/Romany
16	Polish
17	All republics which made up the former USSR
18	Kosovan
19	Albanian
31	Bosnian
32	Croatian
33	Serbian
34	Other republics which made up the former Yugoslavia

36	Mixed white
37	Other white European, European unspecified, European Mixed
39	Other white, white unspecified
	MIXED GROUPS
21	White and Black Caribbean
22	White and Black African
23	White and Asian
24	Black and Asian
25	Black and Chinese
26	Black and White
27	Chinese and White
28	Asian and Chinese
29	Other mixed, mixed unspecified
	ASIAN or ASIAN BRITISH GROUP
41	Indian or British Indian
42	Pakistani or British Pakistani
43	Bangladeshi or British Bangladeshi
44	Mixed Asian
45	Punjabi
46	Kashmiri
47	East African Asian
48	Sri Lankan
49	Tamil
50	Sinhalese
51	British Asian
57	Caribbean Asian
59	Other Asian, Asian unspecified

	BLACK or BLACK BRITISH GROUP
61	Caribbean
62	African
63	Somali
64	Mixed Black
65	Nigerian
66	Black British
69	Other Black, Black unspecified
	OTHER ETHNIC GROUPS
81	Chinese
84	Vietnamese
85	Japanese
86	Filipino
87	Malaysian
89	Any Other Group

