

**Clinical Genetics and Antenatal / Fetal Medicine:  
Liaison and Training**

**A REPORT OF  
THE CLINICAL GENETICS SOCIETY  
PRENATAL GENETICS GROUP**

**December 2008**

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## SUMMARY OF RECOMMENDATIONS

### To improve patient care:

1. a) All Regional Genetic Centres should appoint a Lead Clinical Geneticist for Prenatal Genetics;  
b) Genetics Centres and Antenatal/Fetal Medicine Units should have dedicated contact routes during routine working hours that are widely known. Antenatal Screening Coordinators are well placed to facilitate these contact routes.
2. Genetic Centres should appoint, and/or train, Genetic Counsellors with a specialist interest in Prenatal Genetics.
3. Regional Genetic Centres should produce guidelines for referral to Clinical Genetics, and Antenatal/Fetal Medicine Units must know how to refer to Genetic Services.
4. a) Genetic Centres and Antenatal/Fetal Medicine Units should establish regular joint meetings, no less frequent than three-monthly;  
b) Geneticists should have regular formal contact with all Maternity Units within their region, at least annually, to discuss policy and cases.
5. National guidelines for the storage of fetal tissues and samples should be produced and implemented locally.

### To improve training:

1. a) All Clinical Genetic trainees should be attached to an Antenatal or Fetal Medicine Unit for a minimum agreed period as part of their training;  
b) Core training in genetics exists for Obstetric trainees, and those undergoing subspecialty training in Fetal Medicine already have dedicated time in regional genetics centres. This group recommends that training should be subject to an assessment of both knowledge and skills;  
c) All Midwives should receive training in basic genetics.
2. Local arrangements should be made for trainee Clinical Geneticists to see fetuses and perinatal deaths, and attend post-mortem examinations.

### Audit and Evaluation:

The impact of these recommendations should be audited and evaluated after a three-year period.

## BACKGROUND

Prenatal Genetics is an acute activity of any Clinical Genetics department. It is often the area of genetics practice where much has to be achieved in a very short space of time while less urgent work is put on hold. From the parent(s) point of view, acute anxiety and grief are frequently experienced. Clinical and laboratory accuracy, and prompt access to the latest information, are crucial.

The decision to initiate a Prenatal Genetics sub-group of the Clinical Genetics Society (CGS) was made at the CGS Council Meeting in March 2005. There had been a growing perception of the need to enhance working practices and liaison between Clinical Geneticists and Genetic Counsellors, on the one hand, with Fetal Medicine Specialists, Obstetricians, Midwives and Antenatal Screening Coordinators on the other. The issues of concern do not relate to the management of routine screening programmes (e.g. Down syndrome), but rather to the management of genetic problems and fetal abnormality that fall outside the remit of these programmes. Although individually rare, collectively genetic conditions are common. Whilst examples of good working practice and liaison exist in a number of centres, there are many areas that do not benefit from close integrated working relationships, resulting in sub-optimal care for the mother and fetus because of late or untimely availability of genetics expertise. The speed of change in medical genetic knowledge and technology, Fetal Medicine expertise and imaging technology, NHS reforms, and the introduction of commissioning for specialised services nationally, all combine to prompt the need for:

- i. a review of existing working relationships and practices between Clinical Genetics and Maternity Services;
- ii. exploration of ways in which Clinical Geneticists and Antenatal Services/Fetal Medicine Units might improve their integrated working practice for the benefit of the pregnant mother and fetus;
- iii. consideration of resource implications and the implications for education and training.

The Clinical Genetics members of the Group first met in June 2005 to establish terms of reference (Appendix 1). Correspondence took place between the Group and the President of the Royal College of Obstetricians and Gynaecologists (RCOG), as well as the Chair of the British Maternal and Fetal Medicine Society (BMFMS). The RCOG and BMFMS both expressed support for the Group and the BMFMS identified two individuals from their own membership, who became full members of the Group. In recognition of the key liaison, educational and front line roles of Nurses, Midwives and Antenatal Screening Coordinators, the group expanded during 2006 with members from these disciplines.

In addition, the existence of the Group was brought to the awareness of the National Screening Committee (NSC), and several individuals of the Group have dual membership through NSC sub-committees.

## AIM

To improve the service offered to families at risk of genetic disorders in pregnancy by promoting improved liaison, communication, and integrated working practice between Clinical Genetics Centre and Antenatal/Fetal Medicine Units.

## PREVIOUS COLLEGE STATEMENTS AND OTHER DOCUMENTS

1. *Royal College of Physicians document "Genetic Screening and Prenatal Diagnosis" (1989)*

This document gives a thorough account of the field at the time, including substantial content on basic genetics, with the focus firmly on the provision of *screening* services in antenatal medicine. Nevertheless, a number of clear recommendations regarding the multi-disciplinary nature of the prenatal diagnosis team, delivery of services, and implications for education and training, were made. For example:

***"Each region needs to develop an organisation for ensuring delivery of genetic screening and prenatal diagnosis. This organisation should include clinical geneticists and fetal medicine centres, neonatologists and paediatric pathologists, obstetric and paediatric consultants, primary care physicians, community physicians, health workers involved in family planning, health visitors, midwives, nurses, and experts in health education and community medicine."*** (p52)

***"Because of the large numbers involved, and the relative simplicity of some issues in large-scale screening programmes, genetic information and counselling must be provided at the community level. The ideal professionals to provide information and counselling would be specially trained health visitors and midwives, ..."*** (p52)

***"Professional training in medical genetics and the principles of genetic counselling should be provided for all maternal and child health workers ..."*** (p51)

Despite this recommendation, the group is aware of only one new post that was created directly as a result of the document (Oxford). However, it directly inspired the genetics career of at least one member of this Group.

2. *Joint Royal College of Obstetricians and Gynaecologists/Royal College of Paediatrics and Child Health document "Fetal Abnormalities: Guidelines for Screening, Diagnosis and Management" (1997)*

This document provided a comprehensive review of the screening for, and management of, fetal abnormalities. The 'Guidelines for use in Practice' section included recommendations for multidisciplinary involvement and recognised the role of Clinical Geneticists, as follows:

**"Once the diagnosis of the [fetal] abnormality has been established as accurately as possible, and depending on its nature, it is often important to offer additional discussion with other members of the team:-**

**(list includes)**

**neonatal paediatrician**

**clinical geneticist**

**genetic health visitor or field worker" (para 5.4, p19)**

**"Arrangements must be made for support, counselling and follow-up [after the birth]. A referral to a clinical geneticist for the discussion of the implications for future pregnancies should be offered." (para 5.7, p21)**

There is also a 'Guidelines for Purchasers (Key Questions)' section that posed a range of questions concerning quality and the importance of multidisciplinary working.

3. *Royal College of Obstetricians and Gynaecologists*

The RCOG produced two documents, in 1997 and 2000, dealing mainly with prenatal ultrasound. Only passing reference was made in these documents to liaison with Genetics Services.

4. *ABC of Antenatal Care (BMJ Publications, 2002)*

This popular and influential publication is the fourth edition of a work that first appeared in 1992, based on a series of articles that appeared in the BMJ. Neither the chapter on 'Organisation of antenatal care', nor 'Detection and management of congenital abnormalities', make any reference to the possibility of a contribution from clinical genetic services. The section on Family History within 'Normal Antenatal Management' mentions only diabetes and twinning.

5. *Human Genetics Commission publication, "Making Babies: reproductive decisions and genetic technologies" (2006)*

This document primarily addresses the increasing burden of choice faced by parents, or potential parents, in the face of rapidly developing genetic screening and testing

technologies. In relation to ***“those who receive diagnoses of serious fetal conditions”***, the following is stated in relation to counselling support:

***“... we found that this group do not always receive the support that they need.”***

And: ***“we recommend that a review of information, counselling and support services for those whose fetuses are diagnosed with serious conditions should be commissioned by the Department of Health.” (p12)***

Clear recommendations about the need for integrated services and close working liaison have been made in the past ([1] and [2]), though not consistently ([3] and [4]) or prominently. Recommendations, apart from some limited examples, have not been generally implemented and their impact is now all but lost. Furthermore, early documents were not produced electronically and are no longer easily available. To an extent, however, the first and most significant of these reports (1989) was ahead of its time because the identification of disease-causing genes, and genetic testing technologies, began to accelerate from the mid-1990s and is now highly sophisticated compared to 1990.

#### THE NATIONAL SCREENING COMMITTEE

Current policy development in this area centres largely on the relevant activities of the National Screening Committee (NSC). The Fetal Maternal and Child Health (FMCH) subgroup has been set up to address all aspects of antenatal (plus newborn and child health) screening programmes. The relevant activity of the NSC is summarised in Appendix IV.

#### FETAL PATHOLOGY SERVICES

An issue of recurring concern to Clinical Geneticists is the availability of high quality post-mortem information on fetal loss due to abnormality, and the availability of suitable samples for diagnosis by genetic analysis. Whilst the national organisation of Fetal Pathology services is currently a separate issue, the acquisition of suitable samples is an area requiring close liaison between Fetal Medicine and both clinical and laboratory genetic services. Further discussion is provided in Appendix V and an example guideline provided in Appendix VI.

## SURVEYS OF EXISTING PRACTICE

In order to try and assess current levels of liaison and the strength of working relationships between Clinical Genetics Centres and Antenatal/Fetal Medicine Units, separate but complementary questionnaires were drawn up, one for UK Regional Genetics Centres and the other for Fetal Medicine (FM) Units and Maternity Units (in District General Hospitals—DGHs). The questionnaires and their detailed findings are attached in Appendices IV and V. As well as serving to evaluate current levels of liaison and integrated working practice, these surveys have also served as an audit of the impact of the recommendations made in documents (1) and (2) above. The key issues to emerge from the surveys are described in this section.

### SURVEY OF UK REGIONAL GENETIC CENTRES: KEY ISSUES

All 22 UK Regional Genetic Centres were surveyed. The survey (Appendix II) was conducted by face-to-face or direct telephone contact, with a representative from each centre directly questioned.

#### Key Issues

1. **Departmental Organisation.** More than half of the UK Regional Genetics Centres had no lead Geneticist for prenatal diagnosis and designated Genetic Counsellor posts for prenatal diagnosis existed in only 60% of units. Some units had efficient referral pathway systems, often through Antenatal Screening Co-ordinators and Genetic Counsellors, whilst others identified sub-optimal clinic organisation with inappropriate referrals.
2. **Service Delivery.** Most centres believed they provided a satisfactory or good service for prenatal diagnosis for genetic disorders, even those centres with no designated Prenatal Genetics service. However, there were marked differences between units in terms of numbers of patients seen and the caseload mix, suggesting that there is scope for many centres to provide an enhanced level of service. Some centres aspired to enhancing the level of service, though this was not universally the case.
3. **Liaison and Joint Working.** Very good liaison and joint working arrangements exist in some centres and 70% of centres had regular joint meetings with FM colleagues, though these were almost all confined to tertiary FM units. Liaison

with peripheral antenatal units was generally poor. Some difficulties stem directly from the geographical constraints but in some centres concern was expressed regarding a perceived reluctance for FM specialists to engage fully with the Regional Genetics Centre in the prenatal process. Geneticists were most likely to be involved where there had been a post-mortem report following a TOP / IUD which supported the need for genetics input.

4. **Geographical Issues.** Geographical issues were seen as the most challenging problem for delivering ideal liaison and joint working practices in Prenatal Genetics. Most Regional Genetics Centres are located within a tertiary referral centre and aim to provide their service over a large region. However, this can lead to variable levels of liaison and working arrangements, inequitable service delivery, poor follow-up and continuity of care. In some centres the clinical service and genetics laboratories are located on completely different sites but in general solutions are found to ensure good communication.
5. **Training and Education.** Approximately 70% of Clinical Genetics SpRs receive specialist training in prenatal diagnosis but only a minority rotate through a structured programme. In many centres it is proving difficult for SpRs to achieve exposure to fetal post-mortem examinations, but some exposure is considered necessary for all trainees, and essential for those who wish to make Prenatal Genetics and/or dysmorphology their special interest. There is a perceived need for more Prenatal Genetics teaching in peripheral units, for example to Midwives working in antenatal clinics, as well as the provision of specific events for trainees in Obstetrics & Gynaecology.
6. **Fetal Pathology.** Fetal Pathology services are one of the cornerstones of Prenatal Genetics services. The current lack of satisfactory Fetal Pathology services is a national issue and was highlighted as an area of major concern. Almost all units iterated problems with liaison, delayed reports, and difficult interpretation. These issues are compromising the quality of SpR training in both Clinical Genetics and Obstetrics/Fetal Medicine.

## SURVEY OF BMFMS MEMBERS: KEY ISSUES

There are a total of approximately 200 maternity units nationally. It was felt that this was too large a number to conduct a postal questionnaire and that the response would probably be poor. Instead, all attendees of the BMFMS Conference in Cardiff, April 2006, were asked to complete a questionnaire (Appendix III) individually. In total 126 forms were returned, some completed by individuals, others by groups. Nineteen (15%) of the completed questionnaires failed to identify the unit of origin by name, which meant that the responses could not be correlated with their level of service (i.e. tertiary or non-tertiary). These were excluded from the analysis. 63% of the remaining 107 forms were completed by Consultant Obstetricians or Consultants with a special, or subspecialty, interest in Fetal Medicine, with 27% completed by SpR trainees (See Appendix Vb). This final sample may not be fully representative of maternity units nationally but is hopefully representative of BMFMS membership.

### Key Issues

1. **Knowledge of Local Status.** A high proportion of respondents did not accurately identify the status of their own unit, i.e. whether tertiary (Fetal Medicine Unit—FM Unit) or non-tertiary (District General Hospital—DHG). FM Unit clinicians comprised 47% of the total but 35% of these did not appreciate that they were part of a tertiary service. Of the responders from DGHs (53% overall), 52% erroneously recorded that they were part of a FM Unit. These misconceptions extended across all grades of clinician. This may reflect misunderstandings about the meaning of the term ‘FM Unit’ and/or there may be a lack of clarity in the definition of a FM Unit.
2. **Awareness of Genetics Services.** The apparent lack understanding of the status of their own Antenatal/Fetal Medicine service may imply that there is also a lack of understanding of the level of service available from Regional Genetics Centres. This, in turn, suggests poor, or limited, communication between the two services and under-utilisation of genetics expertise.
3. **Joint Clinics and Access to Genetics.** More than half of FM Unit clinicians, and approximately two-thirds of DGH clinicians, stated that joint clinics with Clinical Geneticists did not take place. However, most responders stated that they had easy and uncomplicated access to advice from a Clinical Geneticist when sought. Easy access to a Genetic Counsellor was denied in 14% of DGH responders and 8% of FM Unit responders. The mutual proximity of the two services was

considered a significant factor in determining close links through clinics and meetings.

4. Respondents reported good access to genetics clinics and meetings, both in DGHs and tertiary centres. As most Regional Genetics Centres are located in large, tertiary centres, with close links to a university, one would not expect that the access of DGH Obstetricians to genetics clinics would be as good as those working in tertiary centres. Most were satisfied with their training.
5. **Guidelines for Referral.** Concerning 'guidelines for referral to genetics', only a quarter of FM Unit respondents were sure that such guidelines existed.
6. **Using Genetics Expertise.** Concerning the degree to which Antenatal/Fetal Medicine Units routinely seek the expertise of Clinical Geneticists:
  - I. When a family history of genetic disease emerged, a quarter indicated that they would do so only occasionally whilst the remainder indicated that they would usually consult. The pattern was similar following the detection of a fetal abnormality by scan (though the questionnaire did not define specific abnormalities).
  - II. Only 17% of respondents said that they would routinely refer to Clinical Genetics following the detection of a sex chromosome abnormality. (The working group consider it likely that this low figure reflects the way the question was interpreted; i.e. the question may have been perceived as referring to lethal Turner syndrome fetuses rather than the full range of sex chromosome abnormalities.)
  - III. Concerning the termination of an abnormal fetus, most respondents indicated that they would only occasionally, or never, notify Clinical Genetics (which may reflect the perceived lack of availability of a geneticist and/or poor understanding of the potential contribution from Clinical Genetics).

## FULL RECOMMENDATIONS

### To improve patient care:

1. a) All Clinical Genetic Centres should appoint a Lead Clinical Geneticist for Prenatal Genetics. This is likely to be the best model for facilitating service development, improved liaison with Fetal Medicine and Obstetrics, and closer working practices. The role of the Lead is outlined below;
  - b) The Regional Genetic Centre should have a dedicated contact route and telephone number during routine working hours that is well known to all staff in antenatal clinics in the catchment area of the genetic service. It may be necessary to identify (an) individual(s), medically trained and/or specialist Genetic Counsellor(s), available on a rota basis to respond to urgent prenatal genetic enquiries;
  - c) The antenatal clinics and services should have a dedicated contact route and telephone number that is well known to the local genetics service. It may be necessary to identify individuals (probably Screening Midwives) who are available on a rota basis to respond to and undertake liaison with the genetic service;
  - d) Antenatal Screening Coordinators are well placed to facilitate these contacts and already play a key role in liaison between Antenatal/Fetal Medicine and Genetic Centres.
2. Genetic centres should appoint and/or train, Genetic Counsellors with a specialist interest in Prenatal Genetics (in the 40% of centres that have no designated Counsellor at present). These individuals would become the service's main point of contact with Antenatal/Fetal Medicine. Based on the Wessex experience of providing a Prenatal Genetics service, one wte Genetic Counsellor per 25,000 live births appears to be appropriate. Trained Genetic Counsellors have the knowledge and competencies to provide genetic counselling in relation to routine issues and diagnoses as well as organise genetic investigations. Clinical Geneticists provide diagnostic insights and strategies.
3. Genetic Centres should produce basic guidelines for referral to Genetics, and find ways to disseminate these as widely as possible within their catchment area (see Appendix VIb). Local modification of such guidelines may be required.
4. a) Genetic Centres and Antenatal/Fetal Medicine services should encourage regular joint meetings, occurring not less frequently than three-monthly;

- b) Geneticists should have regular formal contact with all maternity units within their region, at least annually, to discuss policy and cases. As a minimum such contact can take place at joint perinatal meetings, for example, but visits to maternity units, with presentations, should be encouraged.
5. National guidelines for the storage of fetal tissues and samples should be produced and implemented locally (see Appendix VIa). The possibility of making an accurate diagnosis in an abnormal fetus is severely compromised by lack of samples that might easily have been collected.

To improve training:

1. a) All Clinical Genetic trainees should be attached to an Antenatal or Fetal Medicine Unit for a minimum agreed period as part of their training, at least on a part-time basis. Local service arrangements will determine the range of training opportunities available but the Group recommend that a minimum of three months experience is gained (at least part of which, e.g. one month, should be an attachment to Fetal Medicine). Training should include attendance at Fetal Medicine lists that incorporate invasive procedures and anomaly scans. Trainees should be supervised in the genetic management of cases observed during their training period. It is desirable that training is supplemented by attendance at an organised Prenatal Genetics training course. The current curriculum requirements for Clinical Genetic trainees in 'Prenatal Diagnosis and Fetal Dysmorphology' ([http://www.pmetb.org.uk/fileadmin/user/QA/Curricula/Approved\\_curricula/CLINICAL\\_GENETICS/Clinical\\_genetics\\_3\\_Jul\\_07\\_v.Curr\\_0008.pdf](http://www.pmetb.org.uk/fileadmin/user/QA/Curricula/Approved_curricula/CLINICAL_GENETICS/Clinical_genetics_3_Jul_07_v.Curr_0008.pdf)) do not specify a period of time attached to an Antenatal or Fetal Medicine Unit. We therefore recommend that the curriculum is amended accordingly;
- b) Core training in genetics exists for Obstetric trainees ([http://www.rcog.org.uk/resources/public/pdf/TP\\_8\\_Antenatal\\_Care.pdf](http://www.rcog.org.uk/resources/public/pdf/TP_8_Antenatal_Care.pdf)) as well as those undergoing subspecialty training in Fetal Medicine ([http://www.rcog.org.uk/resources/public/pdf/mfmmodule2\\_competences.pdf](http://www.rcog.org.uk/resources/public/pdf/mfmmodule2_competences.pdf); [http://www.rcog.org.uk/resources/public/pdf/mfmmodule2\\_experience.pdf](http://www.rcog.org.uk/resources/public/pdf/mfmmodule2_experience.pdf)). For both Obstetric and Fetal Medicine trainees the acquisition of basic genetics *knowledge* must be matched by the acquisition of appropriate basic genetic *skills*. Therefore, both knowledge and skills should be subject to assessment. The Group did not define the assessment methodology, as this went beyond the original remit. The Group recommends that further work in this area is undertaken, through the National Genetics Education and Development Centre.

- c) All Midwives should receive training in basic genetics. Midwives offering antenatal screening should receive additional training in the enquiry, documentation and interpretation of family history information and pedigrees. Again, further work is recommended in collaboration with the Royal College of Midwives and Nursing and Midwifery Council in order to set achievable standards and goals.
2. Local arrangements should be made for trainee Clinical Geneticists to see fetuses (normal and abnormal) and perinatal deaths, attend fetal post-mortem examinations, and receive instruction in fetal dysmorphology.

Audit and Evaluation:

The impact of these recommendations should be audited and evaluated after a three-year period, for which the lead responsibility should lie with the Clinical Genetics Society.

## ROLE OF THE LEAD CLINICAL GENETICIST FOR PRENATAL GENETICS IN GENETIC CENTRES

The Lead Clinical Geneticist should:

1. Be involved in the development of referral guidelines to assist Antenatal and Fetal Medicine Units in making appropriate clinical contact with Regional Genetic Centres.
2. Help to establish and disseminate the direct contact route(s) for all the antenatal clinics and fetal medicine units to facilitate direct contact with a Clinical Geneticist or Genetic Counsellor when required.
3. Help to establish and organise regular (not less than three-monthly) multidisciplinary team and audit meetings between Clinical Geneticists and their local Fetal Medicine Unit(s).
4. Help to establish formal contact, on at least an annual basis, with all Maternity Units within the catchment of the Regional Genetics Centre.
5. Facilitate local training and education, for trainees in Clinical Genetics and Obstetrics, and Fetal Medicine specialists in training, including direct clinical exposure. This should include midwifery staff where appropriate.
6. Facilitate where necessary the assessment of knowledge and skills acquired by Obstetricians undergoing core training in genetics, and Fetal Medicine trainees undergoing specialist training in genetics, in order to ensure that the required standard is achieved.
7. Contribute to the development of local implementation strategies for national guidelines (with clinical and laboratory genetics input) for the storage of fetal tissues and samples (once these guidelines are agreed).
8. Be responsible for ensuring that timely genetic counselling provision is available for appropriate cases.

## APPENDIX I

### Terms of Reference, CGS Prenatal Genetics Group

1. To review the existing guidelines and statements from national bodies that address working relationships and clinical liaison between Fetal Medicine and Clinical Genetics.
2. To review current models of care between Fetal Medicine and Clinical Genetics.
3. In the light of the findings from these reviews to appropriately work with the Royal College of Obstetricians and Gynaecologists to produce a joint document whose aim would be to:
  - a) describe best practice;
  - b) propose good models of care, with resource implications;
  - c) propose models of education and training appropriate for trainees in Obstetrics and Fetal Medicine, trainees in Clinical Genetics, and Midwives.

The group reports to the Clinical Genetics Society Council and the RCOG and BMFMS are sent copies of minutes.

## APPENDIX II

### a. The National Screening Committee

The implementation of antenatal screening policies inevitably has an impact on cross-disciplinary liaison and working practice involving clinical genetic services. The Fetal Maternal and Child Health (FMCH) subgroup of the UK National Screening Committee (NSC, established 1996), has been set up to address all aspects of antenatal (plus newborn and child health) screening programmes and the NSC advises central and devolved government on screening policy. The remit of the FMCH in relation to antenatal screening includes:

- 1) Quality Information and Education. For example, ensuring antenatal and newborn screening requirements are fed into the national programme for information technology. A sub-group, Public Information and Professional Development (PIPD), oversees development, implementation and evaluation of information and education and training specific to screening;
- 2) Programme specific issues. Individual screening programmes and their implementation are managed by specific subgroups, which include Fetal Anomaly screening, NHS Sickle Cell and Thalassaemia screening, and Newborn Blood spot (including Cystic Fibrosis) screening (<http://www.screening.nhs.uk/an/index.htm>).

Local implementation is devolved to nine regional screening teams, which comprise a Public Health lead, an Antenatal and Child Health Screening Coordinator, and an Education and Training Facilitator. (<http://www.screening.nhs.uk/cpd/contacts.htm>). Regional teams aim to have robust networks with local health professionals, primary care, public health, laboratories and universities, and to maintain liaison with these groups, including pre- and post-registration midwifery establishments. Activities include co-ordination of 'sharing best practice' forums and communication of information to and from the NSC. Provision of education and training is a priority, including assessment of training needs, commissioning of specific training events, and planning longer term programmes. Regional teams also develop specific resources for local needs, often using resources developed by the NSC that are widely shared with relevant professional bodies:

(<http://www.screening.nhs.uk/cpd/home.htm>).

NSC resources relevant to Prenatal Genetics include:

- **Screening Choices: A resource for health professionals offering antenatal and newborn care**

With an increasing range of screening and diagnostic tests available during pregnancy it is recognised that choices and decisions may be stressful and raise anxieties and dilemmas. A non-directive approach is advocated and the need for specialist help acknowledged.

- **Higher Education - Antenatal Screening Course Template**

Regional teams work with local universities to deliver a course locally, using the template as a guide. The module is offered at; Diploma, Degree or Masters level depending on individual university criteria. Teaching on the course is supported by the regional teams

- **Study Day Templates**

A training needs analysis identified the following priority areas, dominated by genetics and birth defects: *Fetal Anomaly*, *Genetics (1 day)*, *Genetics (intensive 2 day)*, *Infectious Diseases in Pregnancy*, *Haemoglobinopathies*

- **Resource Cards – Free to all Midwives in England from 2006**

A set of 16 double sided, pocket-sized resource cards, covering all NSC Antenatal Screening programmes.

- **Fetal Anomaly Screening Programme**

This comprises a comprehensive multidisciplinary Training Pack that supports all aspects of the screening programme and has rolled out across England. It consists of a handbook and two CD ROMs covering 7 study sessions. A number of study days for Sonographers have been funded in collaboration with Antenatal Results and Choices (ARC).

- **Sickle Cell and Thalassaemia screening programme**

The NHS Sickle Cell & Thalassaemia screening programme has developed a comprehensive range of education resources and programmes which include; professional information leaflets, 'interim awareness' workshops, a multidisciplinary training CD-Rom and the commissioning of a national programme of education '**Professional Education for Genetic Assessment and Screening**' (PEGASUS, <http://www.pegasus.nhs.uk/about.htm>). The **PEGASUS** programme facilitates training in basic genetics for health professionals involved in antenatal and newborn screening in England. PEGASUS is an online set of flexible learning materials that can be supported by study sessions; there is also a comprehensive week-long course for specialist counsellors. The training implementation strategy addresses the education needs of three distinct groups of professionals: 'front-line professionals' (those routinely seeing patients, such as midwives), 'specialist practitioners' (those seeing 'at risk couples') and those professionals with public health and commissioning functions. Regional teams, in collaboration with the PEGASUS team, are responsible for ensuring effective

implementation of this programme. Among the contributing centres to PEGASUS is the Birmingham National Genetics Education and Development Centre.

- **Newborn Blood Spot Screening Programme**

The Training Resources cover information about conditions screened for as part of national programmes in the UK. Training is also available through the London Ideas Genetic Knowledge Park.

b. Congenital Anomaly Registers

In order to monitor the rapidly changing nature of prenatal screening and diagnosis, information on detection and false positive rates are essential. The NSC liaises with members of BINOCAR (British Isles Network Of Congenital Anomaly Registers – [www.binocar.org.uk](http://www.binocar.org.uk)) to provide this information. The aim of BINOCAR is to provide continuous epidemiological monitoring of the frequency, nature, cause and outcomes of congenital anomalies for the population of England, Scotland and Wales, by means of national, regional and disease specific registers of congenital anomalies. Around 53% of England, all of Wales and 18% of Scotland are ‘covered’ by regional congenital anomaly registers with membership of BINOCAR. The registers are population based and collect data on cases with prenatal suspicion of congenital anomaly and on babies born with a congenital anomaly. The Chief Medical Officer in his report of 2004 stated: “*Congenital anomaly registers are a precious national resource: without them, babies’ lives may be lost and the causes of many anomalies present at birth will not be found.*” BINOCAR are working closely with the CMO and DH to establish national coverage for England using Regional Congenital Anomaly Registers.

## APPENDIX III

### Fetal Pathology and Storage of Fetal Samples

A common type of referral to clinical genetics is the request for genetic counselling after a couple (or another member of the wider family) have suffered fetal loss due to an abnormality that may, or may not, have a known diagnosis. Three types of data are important, namely, accurate family history, a post-mortem report on the affected fetus(es), and cytogenetic or molecular tests on the fetus. Progress is so rapid in the identification of disease-causing genes that tests might be possible for the next pregnancy that could not be performed at the time of the pregnancy loss. It is therefore increasingly important for suitable tissue samples and/or DNA to be kept from the abnormal fetus, if at all possible. Genetics laboratories performing prenatal diagnosis will be familiar with the difficulties surrounding exactly which fetuses qualify for storage of tissue or DNA, given that storage capacity is limited. Each laboratory should have a policy or guideline in place for the selection of cases from which samples are to be stored. An example guideline (Genetics Laboratory, Southmead Hospital, Bristol) is given in Appendix VIa.

The situation of planned feticide for cases without a diagnosis also requires special consideration in terms of sample collection and storage. In order to maximise the opportunity for making a diagnosis that informs genetic risk and the possibility of being able to offer prenatal diagnosis in future pregnancies, samples should ideally be taken prior to feticide. This includes amniotic fluid, an aliquot of which should be frozen (e.g. to test for Smith-Lemli-Opitz syndrome) whilst a culture of amniocytes should be performed and the resulting fibroblasts stored. In addition, fetal whole blood should be taken for extraction and storage of DNA and a culture of lymphocytes undertaken.

The storage of tissues and samples is now subject to legislation laid out in the Human Tissue Act (HTA), 2004, which came into effect on 1<sup>st</sup> September 2006. The HTA regulates the removal, storage and use of material from the dead (as well as the living) for 'Scheduled Purposes' with appropriate consent. Previously there was no statutory framework governing this activity but professional guidelines applied. The Scheduled Purposes for which consent is required under the Act includes:

*“Obtaining scientific or medical information about a living or deceased person which may be relevant to any other person (including a future person) – e.g. genetic information.”*

The term 'material' for the purposes of the Act refers to any *cellular* tissue. This does not include DNA, which, after extraction from cellular material, is not regulated by the Act. However, *cellular* material stored for a scheduled purpose, which includes extraction and analysis of DNA from that stored material, is subject to the Act. Within Clinical Genetics practice there is a general tendency to practice consistency with respect to obtaining consent, which, in relation to DNA, goes beyond the requirements of the HTA. It would be seen as good practice if such consistency were practised by all disciplines involved in Prenatal Genetics.

'Appropriate consent', under the terms of the Act and in the context of Prenatal Genetics, will be an adult with parental responsibility or, if none, a 'qualifying relative'. In many clinical scenarios it will be the parent(s) of an affected child/fetus that will be seeking genetic information for the sake of a later pregnancy, and consent will therefore be uncomplicated. However, there are many real and potential scenarios where members of the wider family may seek genetic information following the birth or termination of an affected child/fetus. Such scenarios should be anticipated at the time that consent is obtained for the storage of material.

The purpose of highlighting the issue of sample storage is included here because of its increasing relevance in establishing a diagnosis, providing accurate genetic risk counselling, and informing the possibility of future prenatal diagnosis. The need for this information in the wider family, as well as the immediate family, should be anticipated and incorporated into counselling at the time that consent is discussed. Consideration of the dimensions of the wider family is standard in clinical genetic practice, as well as familiarity with legal and professional guidelines governing consent. Good liaison and working practice between genetics services and Fetal Medicine/Obstetric services helps to ensure that appropriate storage of fetal samples is a routine feature of multidisciplinary care.

APPENDIX IV

a. Questionnaire to UK Regional Genetics Centres

**Questionnaire Re Prenatal Genetics Input in UK**

	Y	N	N/A
1. Do you have a Lead Geneticist for Prenatal Diagnosis in your department?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
If not describe your arrangements?			
2. Does the designated clinic Consultant look after local referrals?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
3. Do you have a designated Genetic Counsellor for Prenatal Diagnosis?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
4. Do you have a designated Genetic Counsellor/ Midwife liaison at the local hospitals	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
5. Do you have joint meetings with your Fetal Medicine Colleagues?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
6. If you answered yes to (5), how frequently?			
A. Weekly			
B. Monthly			
C. Other			
7. Do you have designated Prenatal Clinics?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
8. Do you have designated weekly slots for patients in other clinics?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
9. Within what time frame can a patient be seen?			
A. Within 48 hours			
B. Within 5 working days			
C. Within 10 working days			
10. What is your tertiary referral centre for Fetal Medicine in your region?	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
11. Who do you consider are your other Fetal Medicine Units in your region?			
12. Approx number of patients referred/ week			
13. Do your SpRs receive specialist training in prenatal diagnosis:			
By attending joint clinics with FM Unit?	<input type="checkbox"/>	<input type="checkbox"/>	

By attending Prenatal Genetics Clinic?  
For how long?

By examining fetal post-mortems?

14. Are you involved with genetic diagnosis following an abnormal scan?
15. What arrangements are made for follow up by the Genetics team?
- A. After TOP/IUD with PM
  - B. After TOP/IUD without PM
  - C. After a live birth
16. What do you feel are the deficiencies (if any) in your centre's provision?
17. If there are any deficiencies (Q.16), how do you think they could be rectified?

## b. Data: Survey of UK Regional Genetic Centres

### ***Demographics***

Responses from all 22 UK regional genetic centres were obtained. The survey (Appendix IVa) was conducted by face-to-face or direct telephone contact, with a representative from each centre directly questioned.

### ***Departmental Organisation***

Just over half (55%) of the UK's Regional Genetics Centres had no Lead Geneticist for prenatal diagnosis and in 70% referrals were handled by the designated Consultant for a particular local clinic. Designated Genetic Counsellor posts for prenatal diagnosis existed in only 60% of units and 55% had a designated Genetics Counsellor or Liaison Midwife at the local hospitals (mostly the latter). Some units had an efficient system of channelling referrals from the local hospitals to Clinical Genetics, frequently through the Antenatal Screening Co-ordinators and Genetic Counsellors.

Some units identified sub-optimal clinic organisation with inappropriate referrals sometimes added to the caseload. Most centres made the point that the follow-up of cases often proves difficult due to the regional nature of most genetic services, in some cases covering a very large geographical catchment area from offices that are usually located in tertiary centres.

### ***Service Delivery***

Most geneticists felt that their centre provided a satisfactory or good service for prenatal diagnosis for genetic disorders, even those centres with no designated Prenatal Genetics service. Centres without a designated service expressed an aspiration for development of new clinics to provide an enhanced specialised service, including the appointment of designated Genetic Counsellors.

There were marked differences between units regarding the numbers of patients seen and the caseload mix seen at the time of initial diagnosis (Fig. 1), which suggests that there is scope for many centres to provide an enhanced level of service. Patients were seen within 48 hours of referral in 60% of units, and the remainder seen within five days. Fifteen (63%) centres had designated prenatal clinics but only three of the seven other centres indicated having weekly slots for such patients in other clinics. Otherwise, patients were seen urgently in genetics centres' clinic rooms, or at the patient's local hospital where possible. Such arrangements preclude the Clinical Geneticist consulting the patient in close proximity to an Ultrasound Department.

There was also a substantial difference in the extent to which units proactively involve themselves in antenatal clinics. However, the less proactive units did not necessarily believe that the quality of their service was compromised.

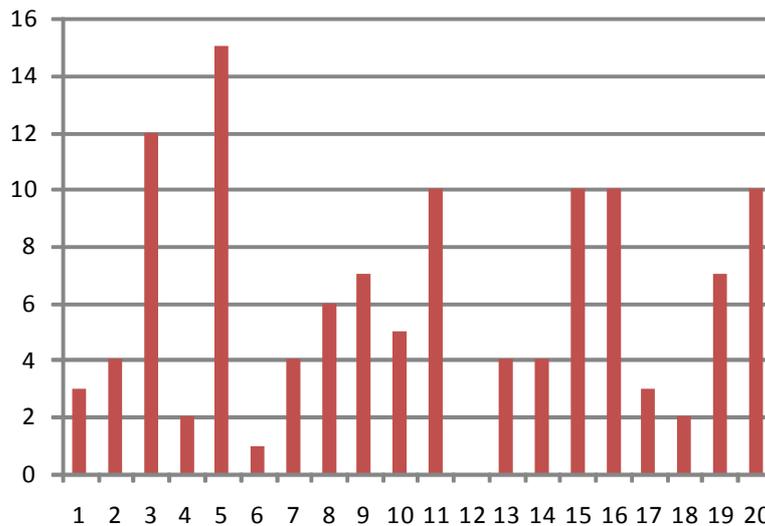


Figure 1. Numbers of patient referrals per week across the 20 centres responding to the survey.

### Liaison and Joint Working

Some centres described very good liaison and joint working arrangements, including regular joint Fetal Medicine management meetings. Figure 2 shows the frequency of these meetings. 70% of centres had regular joint meetings with Fetal Medicine colleagues, though these were almost all confined to tertiary FM Units. Liaison with small, peripheral, Maternity Units was generally poor, though these units would have small numbers of cases suitable for genetics input.

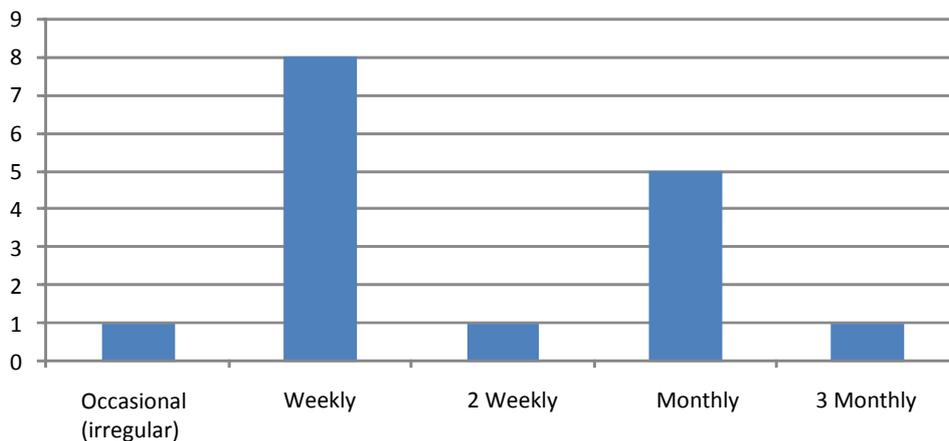


Figure 2. Frequency of joint fetal medicine / Genetics meetings (n = 14)

Other centres highlighted difficulties in joint working arrangements, including the selection of inappropriate referrals cases for Clinical Geneticists. Some difficulties stem directly from the geographical constraints of relatively small departments providing a service over a large area with several FM Units within their region. Close liaison with all the FM Units was not possible, and as a

consequence service delivery inequitable. Some centres expressed concern regarding a perceived reluctance for FM specialists to engage with them in the prenatal process, despite proactively seeking closer liaison, joint Fetal Medicine management meetings, and an enhanced role for Clinical Geneticists. In other centres Clinical Geneticists were frustrated that they were generally over-committed and unable to attend Fetal Medicine meetings.

Figure 3 shows that Clinical Genetics are more likely to be involved if there has been a PM report following a TOP / IUD which supports the need for a genetics consultation. Rates of referral after an abnormal scan, or after an IUD / TOP without a PM, were lower (80%), whilst only 60% routinely receive a genetics follow-up in the period shortly after the live birth of a baby with abnormalities.

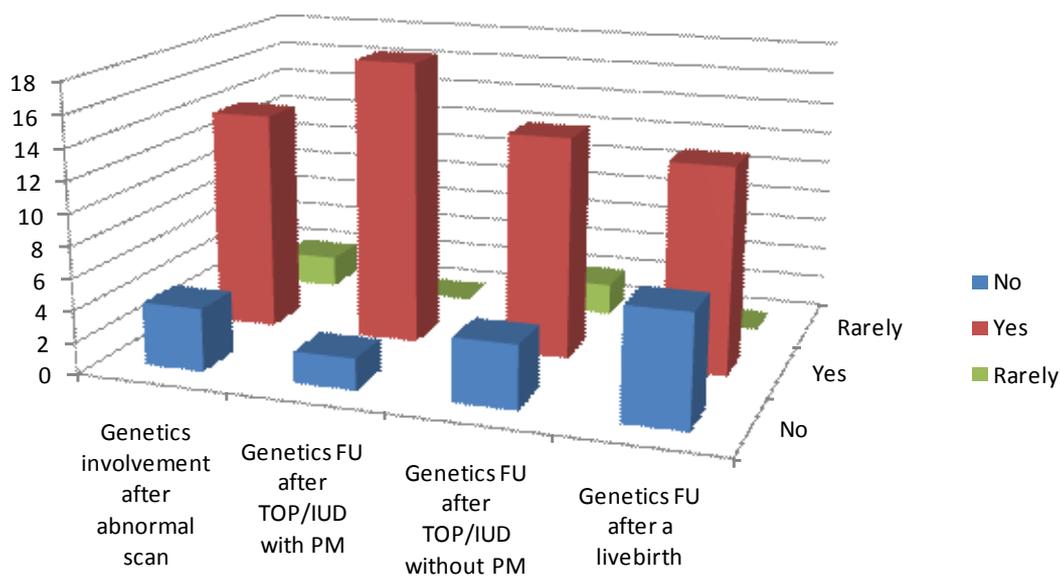


Figure 3. Routine Genetics involvement following the diagnosis of a suspected abnormal fetus / neonate.

### Geographical Issues

As stated, most Regional Genetics Centres are located within a tertiary referral centre and aim to provide their service over a large geographical region. However, this can lead to inequitable service delivery as well as poor follow-up and continuity of care.

For some regional centres the clinical service and genetics laboratories are located on completely different sites, sometimes in different towns/cities. In general, however, solutions are found to ensure good communication. More significantly, in a number of centres the location of three key components of the overall service – Fetal Medicine, Clinical Genetics and Fetal Pathology – are on separate sites, making close liaison difficult and joint meetings infrequent.

In general, geographical issues were seen as the most challenging problem for delivering ideal liaison and joint working practices in Prenatal Genetics.

### ***Training and Education Issues***

Currently, SpRs in Clinical Genetics have no specific requirement to train in Prenatal Genetics. 70% of SpRs receive specialist training in prenatal diagnosis but those rotating through a structured programme are a minority. In many centres it is proving difficult for SpRs to achieve exposure to fetal post-mortem examinations, but some exposure is considered necessary for all trainees, and essential for those who wish to make Prenatal Genetics and/or dysmorphology their special interest.

There is a perceived need for more Prenatal Genetics teaching in peripheral units, for example to midwives—especially those in training (which matches the aspirations of the FMCH subgroup of the NSC).

There is a perceived need also for training days in Prenatal Genetics to be made available for trainees in Obstetrics & Gynaecology.

### ***Fetal Pathology Services***

Good Fetal Pathology is a cornerstone of Prenatal Genetics. The current lack of satisfactory Fetal Pathology services is a national issue and was highlighted as an area of major concern. Almost all units stated a need for more liaison but fetal post-mortem examinations are often being performed well outside the region where the mother resides, reports may take a long time to come through, and interpretation of the information in reports can be difficult. Subsequent communication between Clinical Geneticist and Fetal Pathologist may be slow and unsatisfactory, and these arrangements limit training opportunities for Clinical Genetics SpRs.

### ***Additional Comments***

Prenatal Genetics is an acute work of a Regional Genetics Centre's activity. However, in some centres there appeared to be a reluctance of trainees and some consultants to fully embrace the challenge to enhance the service.

Close liaison between the two disciplines is essential and more flexibility within some units may be required. It is desirable that all units have joint meetings with Fetal Medicine, but where there are local geographical difficulties there should at least be provision for discussion of immediate management, as well as teaching and audit. Educational initiatives, for which there is an increased need generally, have enormous potential to improve the overall service. Fetal imaging technology is developing rapidly but adds a new dimension for multidisciplinary working, including challenges to the dysmorphology skills of Clinical Geneticists.

Genetic Counsellors based in peripheral hospitals have a potentially key role in maintaining good liaison and working arrangements, provided they maintain a profile with the FM or Obstetric Unit. Counsellors can be the key workers ensuring good follow-up, especially in the peripheral hospital that may be geographically distant from the hub of the Regional Genetics Centre.



9. Do you consult a Clinical Geneticist following the detection of fetal abnormalities?  
 Regularly / Occasionally / Never
10. When an abnormal fetus is terminated, does your unit notify Clinical Genetics in order that geneticists have an opportunity to examine the fetus prior to being sent for post-mortem?  
 Regularly / Occasionally / Never
11. Do you refer to a Clinical Geneticist/Genetic Counsellor following detection of a sex chromosome abnormality?  
 Always / Sometimes / Only if requested by the patient / No
12. Do you request molecular genetic tests without consulting Clinical Genetics?  
 Regularly / Occasionally / Never
13. Do you get feedback on your patients through the Clinical Genetics Department? Yes / No
14. Would you find a closer liaison helpful? Yes / No
15. How do you think Clinical Genetics could more usefully contribute to Fetal Medicine Services?

16. Other Comments:

.....

.....

Many thanks for your time and cooperation. There will be representatives of the group at each exit to collect your form at the end of this session.

Rob Sawdy  
 BMFMS  
 DGH Representative to the CGS Prenatal Genetics Working Group

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## b. Data: Survey of BMFMS Members

### ***Demographics of the Respondents***

All attendees of the BMFMS Conference in Cardiff, April 2006, were asked to complete a questionnaire (Appendix III) individually. In total 126 forms were returned, some completed by individuals, others by groups. Nineteen (15%) completed questionnaires failed to identify the unit of origin by name, which meant that the responses could not be correlated with their level of service (i.e. tertiary or non-tertiary). These were excluded from the analysis. The majority of the remaining 107 forms were completed by Consultant Obstetricians or Consultants with a special, or subspecialty, interest in Fetal Medicine, with a significant number completed by SpR trainees (Fig. 1).



*Figure 1. Clinical grades of survey respondents*

A high proportion of respondents did not accurately identify the status of their own unit, i.e. whether tertiary (Fetal Medicine Unit—FM Unit) or non-tertiary (District General Hospital—DGH). FM Unit clinicians comprised 47% of the total but 35% of these did not accurately state that they were part of a tertiary service. Of the respondents from DGHs (53% overall), 52% erroneously recorded that they were part of a FM Unit. These apparent misconceptions extended across all grades of clinician. This may reflect a misunderstanding about the meaning of the term ‘FM Unit’. There may also be a lack of clarity about the definition of a FM Unit.

### ***Liaison and Joint Working***

Most FM Unit respondents (63%) and half of DGH respondents noted that there were subspecialty or special interest training module trainees in their unit. The actual proportion of these in FM Units is likely to be much higher. The frequency with which these trainees attend genetics clinics or meetings is shown in Figure 2. It appears that training needs are largely satisfied.

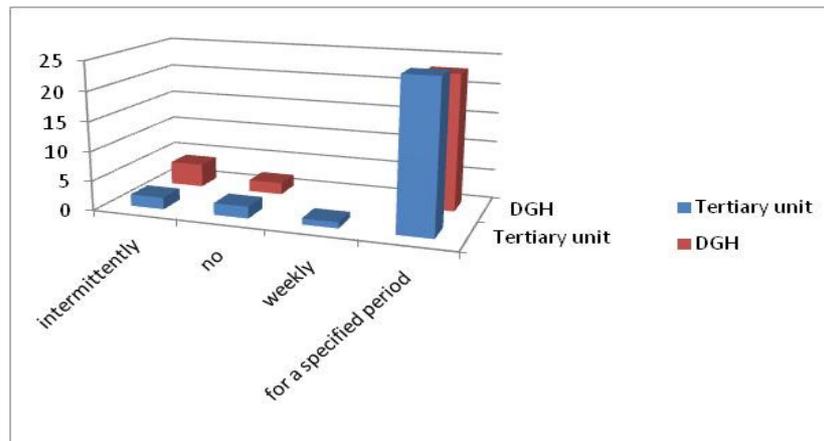


Figure 2. Frequency FM trainees attendance at Genetics clinics or meetings. (Counts of responses).

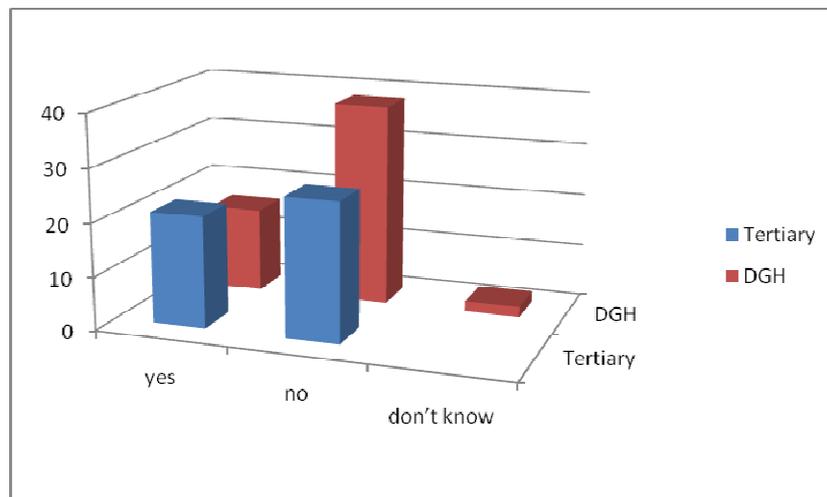


Figure 3. Prevalence of joint FM / CG clinics: Response counts in DGH and FM Units.

Clearly, from Figure 3 joint liaison is less prevalent in DGHs, though more than half of FM Unit responses stated that joint clinics were not in place. The geographical proximity of the two departments was considered a significant factor in determining the number and frequency of joint clinics and meetings.

Access to genetic services in both types of hospital setting was regarded as good and deemed uncomplicated. Most respondents stated that they had easy access to advice from a Clinical Geneticist or a consultation. However, easy access to a Genetics Counsellor was reported to be absent in 14% of DGH, and 8% of FM Unit, respondents.

18% of respondents did not know whether their units had guidelines for referral to CG services. This response was twice as common for FM Unit respondents, where barely more than a quarter of respondents were sure that such guidelines existed (Fig. 4).

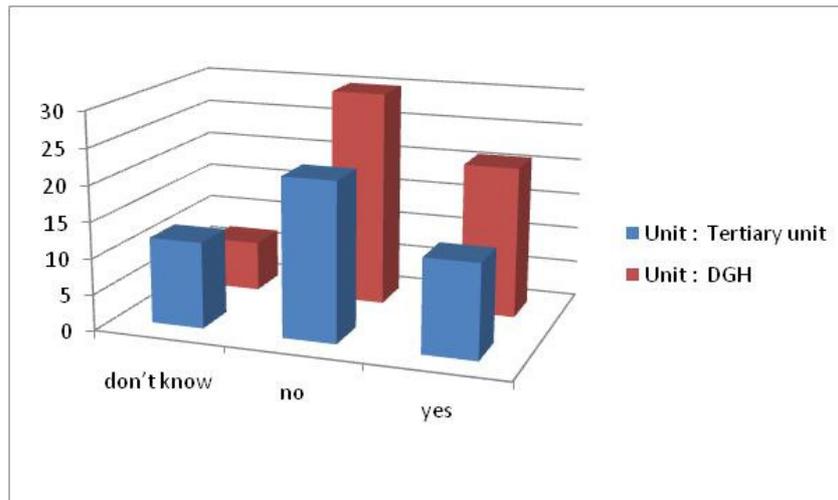


Figure 4. Presence of referral guidelines to CG services within a department: Response counts in DGH and FM Units.

### Reasons for Seeking Help from Geneticists

Most respondents stated that they would consult Clinical Genetics when a family history of a genetic disorder was documented, though a quarter of these admitted they would do so only occasionally. A very similar pattern of responses was evident concerning referral to Clinical Genetics following the detection of a fetal abnormality. But more respondents highlighted that they would never, or only occasionally, notify Clinical Genetics when an abnormal fetus was terminated (in order for Clinical Geneticists to have an opportunity to examine the fetus prior to post-mortem) (Fig. 5).

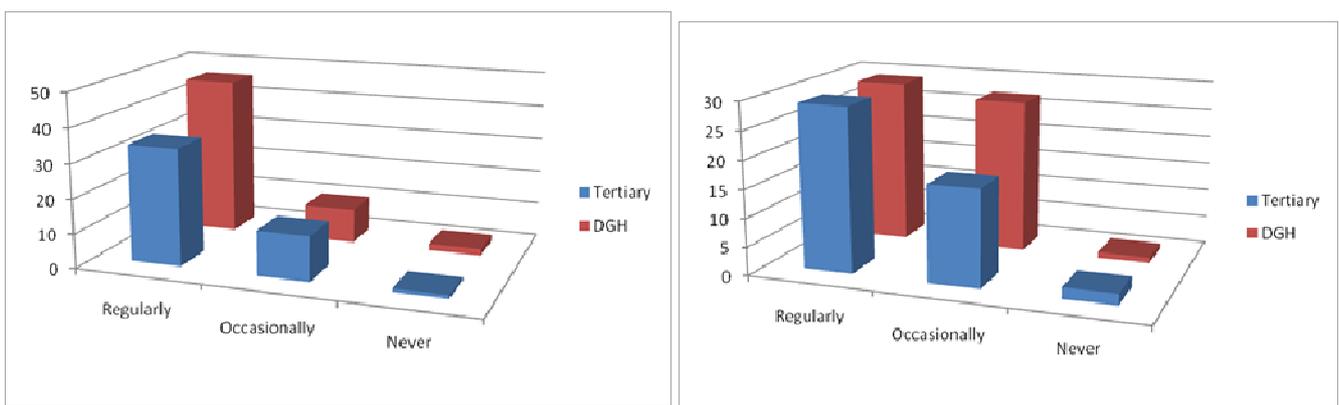
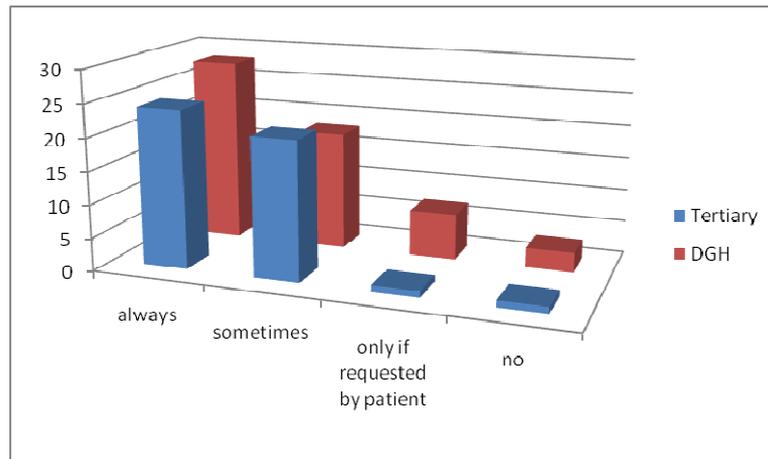


Figure 5. Proportion of respondents who would contact the Clinical Genetics service following the documentation of a positive family history for genetic disease at booking (left panel); or following detection of a fetal abnormality by ultrasound (right panel): Counts of responses between DGH and FM Units.

Only 17% of responders said that they would refer to a Clinical Geneticist or Genetic Counsellor following the detection of a sex chromosome abnormality. A further 36% said that they would only do so if requested by the patient, and 47% indicated they would never do so (Figure 6). This low level of referral may be the result of respondents interpreting the question as a reference to lethal forms of Turner syndrome, rather than a broader interpretation that would include Klinefelter syndrome (47,XXY), 47,XXX syndrome, and 47,XYY syndrome. It was intended that the questionnaire covered all sex chromosome abnormalities but it may have been interpreted narrowly.



*Figure 6. Proportion of respondents who would make a referral to a Clinical Geneticist / Genetics counsellor following the diagnosis of a sex chromosomal abnormality: Counts of responses between DGH and FM Units.*

Feedback (e.g. by letter) on the patients referred to Clinical Genetics was reported to be satisfactory by 87%. However, 63% of respondents agreed that they would find closer liaison helpful.

## APPENDIX VI: EXAMPLE GUIDELINES

### a. Storage of Fetal Samples

[This guideline was researched and drawn up by the Bristol Genetics Laboratory, Southmead Hospital, Bristol, during 2005-06]

### **Guidelines for Storage of Fetal Samples for Diagnostic purposes**

**Storage of Samples:** Samples taken at TOP or post-mortem can be stored for diagnostic purposes in the form of frozen cell culture. This is advisable for:

Termination of pregnancy or miscarriage at, or after, 16 weeks for fetuses with a normal karyotype

With anomalies—single or multiple

Suspected genetic syndromes

Unexplained stillbirths or IUDs

At the request of the Clinical Geneticist, Pathologist, Fetal Medicine Unit

**Duration of Storage:** For as long as possible or indefinitely.

**Consent:** Consent for storage of cell lines is not needed according to recent legislation. On the issue of consent, the recommendations are

**In the case of fetuses less than 24 weeks,** specific consent is not legally required. However, it is recommended that for ethical reasons, consent should be taken for testing and storage of tissue for diagnostic purposes.

**In the case of fetuses 24 weeks or more,** consent taken for post-mortem also includes consent for tissue sampling, appropriate testing on the samples and storage of cell lines for diagnostic purposes. This should be highlighted by the person taking consent. There is evidence that post-mortem reveals additional information in 20 – 40 % of cases. This should also be highlighted during the consent taking process.

**Consent for testing using the cells stored in liquid nitrogen** must be sought separately. This can be done at a later stage when the couple are seen in clinic by the Clinical Geneticist for information about recurrence risks. Although, the couple should be informed about this when consent for storage or post-mortem is taken.

**Consent is usually taken by:** The referring obstetrician.

**Consent can be given by:** Any qualifying relative.

On the cytogenetic request forms, please indicate that storage of cells is required and that consent has been taken.

## b. Antenatal Referrals to the Genetic Service

[This guideline was drawn up by the Prenatal Genetics Group for the purpose of this report]

# **Guidelines for Antenatal Referrals to the Genetic Service**

## **1. Family History**

Family, or personal, history of a genetic, or suspected genetic, disorder including non-specific learning disability.

## **2 Chromosome Abnormalities**

Chromosome abnormality detected by invasive prenatal diagnosis, except trisomy 13, 18 and 21.

Includes: All sex chromosome abnormalities except lethal 45,X (Turner syndrome)

- i. Chromosome mosaicism
- ii. Chromosome translocations (except known balanced familial translocations)
- iii. Chromosome inversions, deletions, duplications and markers

## **3. Ultrasound Abnormalities**

The finding of ultrasound abnormalities may lead to referral to a tertiary fetal medicine centre for a second opinion and a Clinical Genetics referral may be appropriate at the same time. If no tertiary ultrasound appointment is requested, unexplained multiple congenital abnormalities should be referred to the Regional Genetics Centre for a diagnostic opinion.

## **4. Prenatal Diagnosis**

In those patients requiring DNA or enzyme diagnosis, or prenatal diagnosis for a known chromosome rearrangement, the Clinical Genetics Department should be informed ahead of invasive testing so that the laboratory performing the test will be appropriately informed and prepared with the correct probes when the sample arrives.

## **5. Free Fetal DNA Tests**

Fetal Sexing by free fetal DNA at 8-9 weeks gestation for women at risk of X-linked or sex-limited disorders who are requesting diagnostic CVS.

## **6. New Genetic Technologies and Tests**

Genetic technologies are evolving rapidly and new tests may have become available since a patient last had contact with a genetic department. Therefore, even in cases that appear to have a clear management pathway, contact and/or referral may result in the use of new technologies or tests. The results may modify diagnosis, genetic risk counselling, management, and possibly therapeutic intervention for the benefit of the mother and/or child.

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## BIBLIOGRAPHY

Genetic Screening and Prenatal Diagnosis  
Royal College of Physicians, 1989

Fetal Abnormalities. Guidelines for Screening, Diagnosis and Management  
Report of a Joint Working Party of the RCOG and RCPCH. Royal College of Paediatrics and  
Child Health, 1997

ABC of Antenatal Care  
Blackwell BMJ Books, 4<sup>th</sup> Edition, 2002

Human Tissue Act 2004

The Human Tissue Act 2004: an assessment of the Act and its implications for the specialties of  
clinical and laboratory genetics.

Alison Hall, Anneke Lucassen, Gail Norbury, Heather Skirton, Alistair Kent, John Crolla, on  
behalf of the Joint Committee on Medical Genetics, 2006

Making Babies: reproductive decisions and genetic technologies  
Human Genetics Commission, 2006