The Evolving Role of the Clinical Geneticist

A summary of the workshop hosted by the Clinical Genetics Society

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The Evolving Role of the Clinical Geneticist

Authors: Jill Clayton-Smith, Ruth Newbury-Ecob, Jackie Cook, Lynn Greenhalgh

Contributors

In attendance:
Jill Clayton-Smith  President, CGS
Ruth Newbury-Ecob  Vice President, CGS
Jackie Cook  Chair, Lead Clinicians Group
Lynn Greenhalgh  Secretary, CGS, Lead Clinician, Liverpool
Diana Baralle  Treasurer, CGS
Diana Eccles  Academic Vice President, CGS
Dhavendra Kumar  Conference Organiser CGS
Meriel McEntagart  CGS council member and Care Group Lead SWTRGS
Alison Male  CGS council member
Derek Lim  CGS council member
Miranda Splitt  CGS council member
Adam Shaw  CGS council member
Mary Porteus  Lead Clinician, Edinburgh
Kai Ren Ong  Lead Clinician, Birmingham
Fiona Lalloo  Lead Clinician, Manchester
Richard Sandford  Lead Clinician, Cambridge
Pradeep Vasudevan  Lead Clinician, Leicester
Jane Hurst  Lead Clinician, NE Thames
Emma Wakeling  Lead Clinician, NW Thames
Annie Proctor  Lead Clinician, Cardiff
Angus Dobbie  Lead Clinician, Leeds
Emma Kivuva  Lead Clinician, Exeter
Ajoy Sharkar  Lead Clinician, Nottingham
Vivienne McConnell  Lead Clinician, Belfast
Debbie Shears  Lead Clinician, Oxford
Peter Turnpenny  Lead Clinician, Exeter
Trevor Cole  Clinical Geneticist
Kate Tatton-Brown  Clinical Geneticist
Laura Boyes  Genetic Counsellor
Gail Mannion  Genetic Counsellor
Hannah Titherage  Clinical Genetics Trainee
Victoria McKay  Clinical Genetics Trainee
Emily Craft  Clinical Genetics Trainee
Verity Hartill  Clinical Genetics Trainee
Anna Znaczko  Clinical Genetics Trainee
Graham Stuart  Consultant Cardiologist
Jude Hayward  General Practitioner
Ellen Copson  Consultant Oncologist

We are grateful for additional comments from Sarah Smithson, Chair of SAC, and Frances Elmslie Chair of CRG
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Executive Summary

Recent advances in technologies for genomic sequencing are already revolutionising the diagnosis and treatment of many disorders with a genomic basis, including cancer. These technologies are currently being introduced into clinical practice and are leading to mainstreaming of genomic medicine with a greater role for other specialties in delivery of genetic services and to changes in workload and patterns of referral to Regional Genetic Services. Currently, many of the referrals which clinical geneticists receive are tertiary referrals. With their broad-based clinical training and ability to deal with all age groups from conception to the end of life, Clinical Geneticists fulfil a unique role, particularly in the diagnosis and management of multisystem disease. In this area, they are likely to remain key players. In the future, however, it is envisaged that for some areas of clinical practice the role of the Clinical Geneticist will move away from making genetic diagnoses and communicating genetic test results to being a multidisciplinary team member providing genetic education and input to complex cases. There will also be a major role in analysis and interpretation of complex genomic investigations for the wider medical community. As such, Clinical Geneticists will need to develop expertise in certain specialist clinical areas to interact more effectively with mainstream clinicians. As clinical interventions and research will be required to evaluate how new genomic discoveries might impact on patient care, new skills such as bioinformatics and clinical trials experience will be required to deliver genomics in clinical practice. This will require additional education and training of the existing workforce as well as incorporation of these new skills into training programmes. Moreover, new methods of service delivery and commissioning of clinical services will be needed to deliver the genetics services of the future safely, ethically and equitably. Recognising the need to evolve and change, this report summarises the outcome of a workshop which focussed on defining the future role of the clinical geneticist by examining what other specialities will require from us, which new roles we should acquire, the training needs to deliver these, new models of working and likely commissioning requirements.
1. Key Messages

1. Clinical Geneticists will continue to play a key role in the diagnosis and management of multisystem genetic disorders. In some areas of clinical practice, the role of the clinical geneticist will need to evolve to encompass new ways of working which will place less emphasis on diagnosis, but more on utilising new genetic technologies, clinical interpretation of sequencing results and bioinformatics.

2. Clinical Genetics will continue to be a distinct medical specialty affiliated to the Royal College of Physicians. The future role, whilst not including provision of acute or general medical services, will support the principles set out in the Future Hospital Commission. Training of clinical geneticists will evolve in accordance with the Shape of Training review and will take into account the specific training needs for our specialty and the future role our trainees will undertake.

3. In the future most clinical geneticists will work with mainstream specialties as members of multidisciplinary teams and will need to acquire appropriate knowledge in these specialised areas. Through these roles they will continue to be involved in diagnosis, interpretation of results, patient management and discussion of ethical issues.

4. Core genetic services will continue to be needed to advise on the appropriate investigation of complex cases and communication of these results. Organisation of extended family testing and predictive genetics testing will also remain within the remit of clinical genetics services.

5. The way in which genetic services are delivered is likely to change from the current primarily face to face model of consultation to a mixed model of service delivery where telephone consultations and e-appointments are also undertaken.

6. Engagement and working with commissioners will be crucial to match the current pace of change.

7. Clinical geneticists will play a key role in genomics education of the NHS workforce.

8. Increased utilisation of web-based interfaces and IT applications will be needed for the development of the specialty.
2. Background

In 2001 the Clinical Genetics Society produced a document outlining the Role of the Clinical Geneticist. In 2011 this was updated to take into account the expansion of DNA diagnostics. Just four years later it was felt that there was a need to revisit this document in the light of rapidly increasing numbers of referrals and increasing demands for Clinical Genetics input into service developments implementing the new genetic and genomic discoveries into healthcare, as well as the need for education of the wider NHS workforce. Clinical Geneticists have already moved from predominantly studying single genes to the application of genomics, the study of the whole genome, its regulation and the complex interplay between genes. Whilst clinical geneticists will remain essential to the diagnosis and management of many multisystem disorders, the diagnosis and investigation of individuals with genomic disorders involving a single system will be less likely to be the core work of Clinical Geneticists now that testing is more widely accessible. Mainstreaming of genetics into clinical practice has already begun, which is beneficial for patients and for the future health of the nation. The increased use of genetic and genomic testing in mainstream clinical care will lead to Clinical Geneticists developing new and different roles and it is anticipated that our highly specialised workforce will have an important part to play in the new genomic era. In addition to ensuring that new technologies are utilised appropriately and developing skills in new areas such as bioinformatics, clinical geneticists will need to be able to provide appropriate input to multidisciplinary teams, working with and educating colleagues in other specialties. The challenges of these new roles are welcomed by clinical geneticists, but there will be a need to change our clinical service delivery model in order to respond to the rapidly increasing demands for clinical geneticists’ time and resources.

The aims of the workshop held in Birmingham on 23rd-24th November 2014 were thus:

- to determine what input healthcare professionals in mainstream specialties and primary care will need from clinical geneticists in the new genomic era.
- to outline the various roles that a Clinical Geneticist of the future will undertake, taking into account the views of our current trainees, and of our genetic counsellor and clinical scientist colleagues.
- to discuss likely training requirements for both established consultants and trainees and how these might be delivered.
- to share areas of best practice and make recommendations for new ways of working.
- to discuss how the evolving role of the clinical geneticist might impact on/be influenced by clinical commissioning.
3. What Do Other Specialities Want From Future Clinical Genetic Services?

Colleagues from cardiology, oncology and general practice provided important views of how those in mainstream medicine envisage the future of clinical genetics. The main message across the specialties is that they all anticipate playing a much greater role in genomic testing for the disorders which they see commonly, but are clear that they will need to work with specialists in clinical genetics to do this. The main roles that clinical geneticists will need to fulfil will be:

- to assist physicians and other colleagues working in acute settings with appropriate advice and support. In particular to act as a point of referral for provision of advice on rare disorders and complex genetic mechanisms and to undertake the communication of complex results
- to continue the role of the clinical geneticist in syndrome diagnosis as this is not something that mainstream practitioners currently feel confident to do. However, introduction of technologies such as face-recognition software systems may reduce the need in this area
- to provide input into the design and standardisation of gene panels for genomic testing
- to provide input to the clinical interpretation of DNA sequencing results at the stage of reporting to mainstream healthcare practitioners, so that the reports they receive are clear, unambiguous and easy for them to communicate to their patients
- to continue to have a role in investigation of and communication with the extended family
- to provide predictive genetic testing
- to provide input into the management of multisystem genomic disorders, especially rare ones. This input may be through guideline development or participation in MDTs
- to participate in the discussion of ethical issues concerning genomics
- to play a lead role in education of clinical genetic trainees and the NHS workforce.

Mechanisms by which these roles could be fulfilled were discussed. The favoured model was that Clinical Geneticists should ideally be part of multidisciplinary teams (MDTs), each geneticist specialising in a particular area and “learning to speak the language” of that specialty. They should function as part of the specialty network. This would enable them to interface better with the specialty, and enhance their skills of clinical interpretation of genomic results. In this role they would also be educators for those in mainstream specialties. This role within an MDT would also facilitate development of management guidelines and clinical interventions which are likely to become more common for genetic disorders as new therapies are developed and we move further into the era of personalised medicine. Becoming a valuable MDT member requires time to develop specialist expertise. One suggestion was to encourage post CCT placements for genetic trainees in a mainstream specialty and for mainstream trainees in clinical genetics. The concept
of post CCT/CST fellowships (rather than formal credentialing) is currently being discussed at JRCPTB as part of the Shape of Training review.

Interfacing with mainstream specialties and primary care would be improved through better use of **IT and web-based resources**. This is particularly important for GPs who see themselves as having a role in the delivery of genetic services to patients, but are aware that this may have to fit into short appointments. Although more disorders are being identified as having a genetic component, these still form only a small part of the GP workload. GPs face the problem of maintaining their generalist and care co-ordinator role whilst acquiring new knowledge. Rather than a GP spending a longer period of time training in genomics they would prefer a case-based approach to genetic education and management of common genetic disorders such as haemochromatosis or familial hypercholesterolaemia. Where possible they would like to interface with clinical geneticists electronically and would like to see **links from their GP computer worktops** directly to resources for information about appropriate management of genetic disorders. These might include risk assessment tools, protocols and management guidelines. Some already exist but these need to be more comprehensive and explicit. Clinical geneticists and clinical scientists need to work with GPs to **develop and update information resources**. The currently provided comprehensive **clinic letters** are helpful for GPs and other specialists but could be **more structured** and contain more information on management with clear **problem lists** and **action plans**. For GPs in particular who are now “paperless” the **move to electronic communication** is preferred.

As more practitioners in primary care and medical specialities begin to deliver genetic services and become familiar with new technologies and the reports produced, the need for every patient to be seen face to face in a genetics clinic will reduce. **Other models of service delivery** by **telephone or e-consulting** will become more appropriate but this will require not only development of **IT capacity** but also a **change in commissioning** because at present the current arrangements encourage face to face consultations only.
4. Which Of Our Current Roles Will Be Less Important In the Future?

- The current Clinical Geneticist’s role as a gatekeeper for genetic testing will be reduced as genomic testing is devolved to specialties.
- Counselling and testing for commoner diseases, especially Mendelian disorders will be devolved.
- Fewer patients will be seen face to face in clinic as more efficient models of care and tariffs for funding of alternative modes of delivery are developed.
- The clinical geneticist’s role in prenatal diagnosis will change, though not necessarily diminish, as Fetal Medicine Teams undertake more testing independently and as newer technologies such as non-invasive prenatal testing are introduced. Maintaining a role in the Fetal Medicine MDT will, however, be important because of the clinical geneticist’s expertise in dealing with rare multiple anomaly syndromes.

5. What Might The Genetics “Clinic” Of The Future Look Like?

Current genetics clinics follow a traditional model of referral for assessment including family history, medical history and examination, followed by investigation tailored specifically to the presentation and differential diagnosis of the index case. Genomic testing means that a wider range of possible diagnoses can be explored through a single test and additional information regarding other unrelated pathologies or risks determined. If genomic testing is the first step in the pathway, then pre-test assessment could potentially be replaced by a post-test assessment aimed at phenotyping to assist interpretation of the genomic results. As an example, a patient with undiagnosed intellectual disability could undergo whole genome sequencing arranged by a paediatrician, and then receive an appointment with a clinical geneticist for discussion and interpretation of the results. We would call this “reverse phenotyping”. The issues around disclosure of additional or incidental findings could be carried out separately. This reflects the model developed for the Genomics England 100,000 genomes project of mainstream clinicians undertaking genomic testing and referring to Clinical Genetics where necessary for further advice/interpretation.
6. How Will Future Clinical Genetics Services Need To Change?

The anticipated changes in the use of genetics/genomics in clinical practice include:

1. Genetic/genomic testing and results will be used much more extensively in primary and secondary care.
2. Results of genetic/genomic testing will increasingly influence management and in some cases determine treatment.
3. The move from single gene testing to genomic testing will lead to greater complexity of result reporting and an increased need for interpretation and advice.
4. There will be an increasing need for skills in the communication of complex information.

We would therefore anticipate:

1. A change in the nature of referrals into the service with a move towards more complex cases.
2. Referrals will need to be triaged more effectively using nationally accepted guidelines. Written/electronic information will need to be provided for the referrer where a referral is deemed inappropriate. Accurate and understandable information will need to be made easily accessible to guide mainstream colleagues regarding appropriate referrals, in a format that fits with their working practices.
3. Alternative ways of working rather than traditional face to face patient contact will need to be explored and developed. These will include telephone and electronic contact with patients and attendance at MDTs. This change of practice will require clinical genetics involvement at planning and commissioning levels and will need to be embedded within the clinical genetics training curriculum.
4. The roles and skills of different members of the Clinical Genetics Team will need to be explored:
   a) Consultants involved in diagnosis, interpretation, advice, communication.
   b) Genetic Counsellors involved in the communication of complex information and working with extended families with known diagnoses.
   c) Administrative roles for collection of individual and family information (pre-clinic preparation), thus freeing up time for consultants and genetic counsellors.

Clinical Genetic services cannot be updated without engaging and working with commissioners. Currently there are multiple commissioning models across the UK, each with their advantages and disadvantages. The main models are block contracts, cost and volume contracts or both. A uniform commissioning model with built-in flexibility to reflect the changes happening now and in the future is needed urgently. Commissioning needs to reflect the different ways of working (face to face and others) and include MDT working. All of these ways of working need to be reflected in the job planning process.
Commissioning of genetic/genomic tests needs to be separated from the commissioning of Clinical Genetic services. Most genetic/genomic testing will be requested by mainstream specialties which is also where cost savings will be made through earlier diagnoses and targeted interventions and therapies. The current gatekeeping role of Clinical Genetics will end but Clinical Geneticists will continue to provide advice to commissioners and colleagues about which tests should be undertaken and how this may result in cost savings and better use of resources in other clinical pathways.
7. Specific Clinical Scenarios

Several common clinical scenarios were discussed in an attempt to arrive at a consensus on which type of referrals should not be accepted given the demands on the workforce in the face of new genetic technologies. For scenarios where the clinical genetics team would not expect to see patients, the development of standard letters which could be used across the various Regional Services was seen as a useful exercise.

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<tr>
<th>Scenario</th>
<th>Should Be Seen?</th>
<th>Comment</th>
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<tr>
<td>Non-dysmorphic patient with autistic spectrum disorder. Array and FraX negative. Mild ID. No family history. Normocephalic. Referred for diagnosis.</td>
<td>No*</td>
<td>Genetics unlikely to contribute to patient management. Basic genetic/genomic investigations can be undertaken by paediatrician.</td>
</tr>
<tr>
<td>Similar patient to above referred for advice regarding recurrence risks.</td>
<td>No consensus</td>
<td>Cannot give an empiric risk figure safely if child has not been seen and the family history fully explored. Some would give empiric risks.</td>
</tr>
<tr>
<td>Request for confirmatory genetic testing after a clinical diagnosis has been made</td>
<td>No (NB Scotland must see)</td>
<td>Testing to be requested by specialty concerned.</td>
</tr>
<tr>
<td>Asked to discuss result of an array or other genomic test carried out in a mainstream setting e.g paediatric nephrologist requests test for nephrotic syndrome.</td>
<td>If clinical geneticist unlikely to add value move to a model where requesting professional is encouraged to give result Offer to see if this is likely to alter management</td>
<td>Need to build capacity and expertise in others This type of case is likely to be discussed in an MDT and decision made there about who will see thereafter</td>
</tr>
<tr>
<td>Request to test parents’ samples to aid interpretation of array or other molecular result</td>
<td>No</td>
<td></td>
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<tr>
<td>Clinicians should take responsibility for tests they order and put mechanisms in place for parental testing.</td>
<td>Laboratory could send out request forms for samples from parents with results of child’s investigations and parents could have samples taken at their GPs.</td>
<td></td>
</tr>
<tr>
<td>Carrier testing where the carrier risk is low e.g. partner of individual affected by or carrier of a rare recessive disorder.</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Need a national consensus as to what is “low”. Some UK departments use 1/70 as cut-off carrier risk. May be at odds with ESHG current guidance.</td>
<td></td>
<td></td>
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<tr>
<td>Carriers detected through GEL e.g. HFE, FH, SMA, CAH, AAT, CF Requests from primary care for cascade carrier testing for AAT, HFE</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Need standard process for managing these across all centres. Consensus statement to be drawn up.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Cascade testing and prenatal diagnosis for neurosusceptibility loci</td>
<td>Uncertain</td>
<td></td>
</tr>
<tr>
<td>Requires debate and national guidance.</td>
<td></td>
<td></td>
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<tr>
<td>Investigation of recurrent miscarriage and male infertility</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Testing by obstetrics/andrology. See if abnormality detected.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Hypermobility variant of Ehlers Danlos Syndrome</td>
<td>No#</td>
<td></td>
</tr>
<tr>
<td>No genetic confirmatory testing. Send standard letter with EDS clinical checklist required for referral and forward EDS service leaflet on Joint Hypermobility Syndrome.</td>
<td></td>
<td></td>
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<tr>
<td>Predictive genetic test</td>
<td>Yes</td>
<td></td>
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* standard letters, which we could all share should be used # as long as clear from referral that there are no features of more serious disorder
**Recommendations:**

- New ways of working to cope with the demands posed by the large number of results from new genomic technologies need to be developed.
- There is a need to engage and work with commissioners to plan and develop new genetic services.
- There is a need for development of common standards for triaging across all of the UK services and a need to share standard letters.
8. What Skills Will The Clinical Geneticist of the Future Need?

The mainstreaming agenda and the delivery of the Genomics England 100,000 genome project in particular will mean that the role of the clinical geneticist will evolve significantly. To take on these new roles in clinical interpretation, education and new models of service delivery the Clinical Geneticist will require new skills. It is likely that some new skills will have to be learned “on the job” where existing consultants are concerned. It is essential that resources for training and education be made available for established consultants as well as being incorporated into the SpR training programme.

Training and education needs identified were:

- Bioinformatics.
- IT skills including E-consulting.
- Clinical interpretation of genomic variants.
- Training in a chosen subspecialty for all future consultants.
- Clinical trials training and experience for some.
- Interfacing with and supporting the role of other physicians/specialists in the care of patients with genetic/genomic disorders.

Future training will also need to take account of the changes to hospital-based patient care in general reflected in our curriculum.
9. Delivering Education To The Future Clinical Genetics Workforce

Trainees

A Postgraduate Certificate (PG Cert) education and training programme hosted and accredited by St George’s, London has been developed and is currently being piloted. This will provide a broad-based training and in particular will provide skills to understand interpret and communicate genomic findings. It involves face to face teaching, a laboratory attachment, written assignments, role play and a two-day workshop as well as keeping a personal and professional development portfolio. Some of the modules are being developed as on-line learning modules. It is possible that following SAC discussion and local arrangements that this programme can be rolled out to all trainees in due course, perhaps through setting up satellite units in other regions.

A number of MScs in Genomic Medicine have been established following a call from Health Education England in 2015. These will begin in September 2015. In addition to funded full and part-time places several of the MSc modules will be available to access as stand alone modules and though not all will be suitable for trainees, some of these could be accessed by trainees to provide training in appropriate areas e.g. in OMICs technologies.

Post CCT sub-specialty exchanges for trainees will be crucial to developing roles as effective members of an MDT

Consultants

Access to modules of the PGCert to be undertaken as CPD modules
Access to modules of the new MSc programme courses in Genomic Medicine. These MScs have been designed with the specific intent that stand-alone modules e.g. in bioinformatics can be used for CPD purposes. Many of these will be developed as e-learning modules.

Recommendations:

- Consultants and trainees need to identify training needs and ways in which these can be addressed to fulfil both specialty-specific requirements and those of the future physician.
- Where consultants and trainees are involved to a significant extent in the delivery of genomic education, this time should be costed and funding provided to back fill clinical duties.
10. The Future Clinical Geneticist as a Researcher

The majority of Clinical Geneticists are actively involved with or would like to be involved in research, as expected in a specialty at the forefront of technological and scientific discovery. 10% of trainees did not envisage participating in significant research activities, but all will be required to identify patients eligible for and be actively involved in recruitment and consenting to NIHR Portfolio studies. It is accepted, however, that opportunities to lead research are likely be limited for those in full-time NHS posts due to the need to combine the responsibilities that being a PI entails with ever-increasing clinical commitments. For those undertaking significant research activities it is crucial that research costs are calculated and are transparently identified in the departmental budget so that clinical duties can be back-filled. Most Trusts have developed criteria for justification of a Research PA and those fulfilling these should ensure that this is in their job plan. Those working in Trusts with Clinical Research Facilities and well managed CRN infrastructure undoubtedly find it easier to participate in research and the resources available in individual Trusts should be explored. Involvement of Genetic Counsellors, students and visiting overseas doctors in research is encouraged, as long as all have the appropriate research training.

It is envisaged that some future clinical geneticists will play a key role in leading trials of clinical interventions to treat genetic diseases; clinical geneticists will often be well placed to identify and recruit patients with genetic diseases where an intervention needs to be tested in a clinical trial.

Recommendations:

- For research undertaken, research costs need to be clarified, invoices for these raised and payment tracked
- Consultants should examine criteria for a Trust Research PA and seek to include in job plan if appropriate
- All consultants and trainees who are undertaking research must have up to date Good Clinical Practice (GCP) training
- For Clinical Trials Of Investigational Medical Products (CTIMPs) appropriate training may include prescribing training depending on local Trust guidelines
- Those seeking to play a wider role in clinical trials should look for experience in developing and delivering clinical trials, working within the regulatory environment, and working within pharmaceutical companies. This could be done through close collaboration or secondment to a Clinical Trials Unit. Some may consider working towards a formal qualification e.g. Dip Pharm Med or other diploma.
11. The Future Clinical Geneticist: Example Job Planning

Job plans will be unique to each consultant, as they are likely to be undertaking diverse roles and working in different ways with a variety of MDTs. The following therefore provide guidance only:

- Consultants should see only complex cases, whether cancer or non-cancer.
- The future consultant role will involve a significant amount of service development, training and education as well as CPD and essential training. For a 10 Programmed Activity (PA) post a minimum of 2 Supporting Programmed Activities (SPAs) should be sought from the employing Trust.
- Where face to face consultations are carried out, each should last a minimum of 30-45 minutes (Pre-clinic contact teams have been used to good effect in some centres to take family histories, and to procure appropriate paperwork and investigation results pre-clinic to maximise clinic time).
- The number of patients that each consultant will be expected to see will vary depending on their contracted hours and their other responsibilities. Additionally, it is anticipated that other forms of consultation will increasingly take the place of some face to face consultations. An estimated number of consultations to be undertaken, tailored to the clinical geneticist concerned, should be calculated during the job planning process to provide guidance. Flexibility should be exercised when fitting these consultations into a job plan.
- As a speciality Clinical Genetics should work towards increasing flexibility in delivery of genetic services with consultants being able to undertake telephone consultations or e-consultations instead of face to face clinics. Arrangements with Commissioners and Trusts should be sought to allow this.
- Where a significant amount of research is undertaken research PA(s) should be sought from Trust/University/Research Network etc.
- Teaching or training duties should be specified in the job plan and additional resources for these should be identified.
- Each job plan should include audit activity and the Clinical Geneticist should be able to demonstrate leading or actively participating in audit activity at appraisal.
- Each consultant should consider developing a sub-specialty interest and participating in one or more MDTs which should be included in the job plan.
- Supporting the laboratory with clinical interpretation and reporting should be included in the job plan as part of Direct Clinical Care (DCC).
12. Guidelines For Correspondence

- A summary clinic letter should be written to the patient and referring doctors unless there are specific reasons not to do so. Reasons for not writing a letter should be documented clearly. Suggested target times for a routine clinic letter are that it should be dictated within 3 working days and approved within 10 working days. For clinic letters pertaining to “urgent” appointments the target is for these to be sent should be 3 days.
- Letters should also be written as above following formal telephone consultations.
- All other correspondence should be dealt with in a timely manner. This correspondence can be cumulative over the working life of a consultant and for senior consultants this may need to be reflected in the job plan or other strategies put in place to deal with this.
- GMC guidelines for record-keeping need to be adhered to and each consultant should have notes and correspondence audited at 2-3 yearly intervals.

13. Clinical Governance

All consultants should have an annual appraisal and participate in 5 yearly revalidation, undertaking the appropriate amount of continuing professional development (CPD). This will ensure that consultants are undertaking the appropriate mandatory training within their Trust. They should also adhere to local Trust policies e.g. for taking notes off site, e-mail policy and chaperoning or have specific agreements with their Trust regarding these. For the most part, this will ensure adequate governance arrangements. There are, however, some circumstances where there may be a role for professional governance group to advise on governance issues specific to our speciality, to offer support and advice when complaints are lodged and to provide a mentoring role for clinical geneticists in difficulty.

Recommendations:

- Clinical geneticists should comply with annual appraisal within their Trust and 5 yearly revalidation
- In view of the nature of our work, particular attention should be paid to ensuring timely and good quality correspondence
- A specific Clinical Governance body should be set up under the auspices of the CGS to provide advice on professional issues and to play a role in support and mentoring for geneticists in difficulty.