CGS
CLINICAL GENETICS SOCIETY

Maximising the Patient Benefit of Genomics – the evolving role of the Clinical Genetics Services

AGNC
Association of Genetic Nurses & Counsellors

BSGM
The British Society for Genetic Medicine

UK
CANCER
GENETICS
GROUP

A summary of the workshop hosted by the Clinical Genetics Society and the Genomics Clinical Reference Group chair

June 13th, 2024

A joint document from the Clinical Genetics Society and Genomics Clinical Reference Group supported by the Association of Genetic Nurses and Counsellors, British Society of Genomic Medicine and Cancer Genetics Special Interest Group.

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Executive Summary

Our ability to interrogate the human genome at increasingly high resolution has revolutionised our understanding of the role of genomic variation in health and disease. An individual's genome can now be sequenced and clinically interpreted at a cost which is affordable to many healthcare systems. Genomic sequencing is offered routinely for eligible NHS patients in England, and the promise of genomically-informed healthcare has become reality in many medical specialties beyond Clinical Genetics. Thousands of patients per year now receive accurate genomic diagnoses for rare diseases and tumour predisposition syndromes, which were previously undiagnosable. Often, the diagnosis is made in time to influence disease management and improve patient outcomes. The positive impact of a genomic diagnosis may extend to family members who gain access to predictive genomic testing, to make life-saving decisions about preventative healthcare, and to make informed reproductive choices. Genomic testing is also being applied to blood and tumour samples taken from patients with cancer, revolutionising precision diagnosis, allowing more accurate prognostication, and enabling precision treatment and clinical trial entry.

There are twenty-three clinical genetics services in the United Kingdom, offering expertise in the diagnosis and management of patients with rare and inherited disorders, including tumour predisposition syndromes. The specialist workforce is the most highly trained, patient-facing genomics knowledge resource in the health service and consists of Clinical Geneticists, Genetic Counsellors, Genetic Nurses, Genomic Associates and Genomic Practitioners.

Clinical genetics services (CGSs) have been evolving alongside genomic technologies, incorporating access to state-of-the-art genomic sequencing into new clinical care pathways. Rapid clinical pathways have been developed when there is urgent need to precisely diagnose a genomic disorder, for example in prenatal and intensive care settings, and to inform therapeutic and surgical choices in cancers and rare diseases. Access to testing of cancer genomes has created the opportunity for major service developments within CGS Cancer Genetics teams, in conjunction with colleagues in oncology and pathology.

With the success of greater numbers of patients receiving a genomic diagnosis comes an opportunity and a responsibility to offer pre-test counselling, genomic testing, and preventative care to other family members at risk. If the benefits of a genomic diagnosis are *not* extended to other family members systematically, the healthcare system misses the opportunity to improve public health and implement preventative medicine at national scale.

CGSs are now providing education, training, and multi-disciplinary clinical support to many more of their mainstream colleagues as recognition of the genomic basis of disease increases across all specialties. Genomic maturity is unequal between specialties and between clinical colleagues in primary, secondary and tertiary care, leading to variable and unpredictable levels of demand on CGSs.

Many Clinical Genetics Services have now exceeded their capacity to meet the needs of all patients referred to their service in a timely way using their current resources and models of care. All CGS clinicians aspire to maximise their value to patients requiring genomic expertise. The following document, which is the output of a workshop held in June 2024, is strong evidence that Clinical Leaders within the CGSs have thought deeply about their responsibility to maximise patient benefits of genomics throughout the healthcare system. The participants, representing the clinical leadership from all nations of the United Kingdom, were able to reach a consensus on referral guidelines to clinical genetics services which maximise the value of this limited human resource.

The consensus referral guidelines are a step towards transforming the provision of equitable genomic healthcare in the NHS. Implementing new referral guidelines is within the sphere of control of the CGSs and there is a strong commitment from the Clinical Leadership to do so. The next steps in service transformation will require systems level change, informed by a deeper understanding of the variability in genomic maturity which exists across mainstream care, as well as the current challenges faced by CGSs in all four nations. The CGSs are keen to support the co-development of a roadmap to enable the safe transfer of genomic healthcare between CGS specialists and non-specialist colleagues, whilst recognising the need to maintain highly specialised care provision to patients whose needs are not currently met by other clinicians within the healthcare system.

Key Messages

1. Clinical Genetics Services remain at the forefront of delivering genomic medicine. They are embracing the challenge of evolving their models of clinical care delivery to maximise the patient benefits of evolving genomic technology, which is now clinically available within the NHSE Genomic Medicine Service.
2. The NHS Clinical Genetics workforce is the most highly trained genomics human resource within the healthcare system and will maintain a key role in the diagnosis and management of complex rare disease and inherited tumour predisposition syndromes.
3. Clinical Genetics Services recognise their responsibility to develop and implement national clinical referral guidelines which ensure equitable access to specialised genomic healthcare for those who will benefit.
4. Given the dynamic and fast-paced nature of genomic research and innovation, CGSs recognise that their specialised skill set is also required to educate and train mainstream colleagues, as well as co-design new ways of delivering genomic healthcare within Clinical Genetics Services and in the mainstream setting.
5. The specialist workforce of Clinical Geneticists and Genetic Counsellors, recently joined by Genomic Practitioners and Genomic Associates, are working collaboratively to ensure that each works at the top of their scope of practise whilst respecting their separate professional roles and responsibilities.
6. The high diagnostic yield achieved using a combination of next generation sequencing and excellent bioinformatic pipelines, affords the opportunity of reducing the amount of clinical time spent on diagnostic pathways, to be replaced by increasing demand for expert input to mainstream genomic healthcare, co-ordinating surveillance programmes, provision of lifelong care to those affected by rare disease, and the potential of running therapeutic trials and delivering therapies as examples.
7. The referral eligibility criteria presented in the following document reflects the enormous range of skills within these specialised services and takes the first step towards enabling all services to align their resources for the provision of equitable care across England.
8. Further work will be required to implement these referral guidelines, recognising that regional services understand the diverse populations they serve, and that demand for Clinical Genetics Services also varies according to the genomic maturity of the regional healthcare system within which they are located.
9. Members of the Clinical Genetics Society, Lead Clinicians Group, United Kingdom Cancer Genetics Group and the Association of Genetic Nurses and Counsellors are keen to engage with, and work alongside, their national and regional commissioning colleagues at NHSE, to ensure that the pace of change in genomic technology is translated into population and individual patient-level benefit in an equitable way.

1. Background

There are 17 National Health Service England (NHSE)-commissioned Regional Clinical Genetics Services (CGS), four in Scotland, and one each in Northern Ireland and Wales. Within the current NHSE service specification for Medical Genetics (adopted 01/10/13) there are no formal, national acceptance or exclusion criteria or nationally accepted referral guidelines. The current service specification states that: -

- detailed referral guidelines for clinical referrals and referral criteria for joint/multidisciplinary clinics should be provided on regional CGS websites.
- clinical Genetics services should have robust referral criteria in place to ensure that they only see patients for whom they are likely to be able to offer advice.
- the CGS will have a process in place to determine how referrals will be managed. This will describe the most appropriate person to manage a case (genetic counsellor or clinical geneticist) and will explain which patients need to be seen in a clinic.

After the publication of this service specification in November 2014, the Clinical Genetics Society convened a workshop to discuss the **Evolving Role of the Clinical Geneticist** (Clayton-Smith, Newbury-Ecob, Cook, Greenhalgh) (https://www.clingensoc.org/education/educational-resources/). As part of this workshop, agreement was reached that Clinical Genetics Services would need to review their models of care provision in response to the widespread adoption of genomics into mainstream clinical practice. The following recommendations were made: -

1. Referrals should be triaged effectively using nationally accepted guidelines.
2. Common standards for triaging across all the UK Clinical Genetics services should be developed and adopted.
3. Written/electronic information should be provided for the referrer when genetic care of the patient is more appropriately managed by the referring clinician or another specialty with advice and guidance, or there is a request for advice and guidance.
4. Accurate and understandable information should be made easily accessible to guide mainstream colleagues regarding appropriate referrals, in a format that fits with their working practices.
5. Standard letters for advice and guidance or reasons for rejection should be developed and shared across all Clinical Genetics services.

Since the 2014 workshop, predictions about the evolution of genomic medicine, mainstreaming genomics, and its impact on regional CGSs have become reality. The services are now facing referral pressures which are unparalleled, with largely static staffing levels, leading to longer waiting lists and the necessity to move towards delivering services within a new clinical model. A critical element of this service pressure is the competing need to support mainstream services to use genomic testing for maximum patient benefit, whilst maintaining timely access to the CGSs for service users whose genomic healthcare needs are most appropriately served by specialist CGS clinicians.

The June 2024 CGS-sponsored workshop brought together the lead (or a delegate) clinicians from 13 of the 17 CGS providers in the English NHS, plus representatives from two of the three devolved nations (Wales and Scotland) and from the clinical constituent groups of the British Society of Genomic Medicine (BSGM) (the Clinical Genetics Society and the United Kingdom Cancer Genomics Group). Genetic Counsellors were represented by the Chair of the UK Lead Genetic Counsellor Group, Chair of the Association of Genetic Nurses and Counsellors and the Chair of BSGM.

2. Workshop Objectives

- To delineate where the Clinical Genetics workforce adds value to patient care.
- To develop referral guidelines which determine the acceptance criteria for referrals to Regional Clinical Genetics services.

3. Principles and assumptions underpinning the development of referral guidelines

In order to develop appropriate referral guidelines, workshop participants first discussed the assumptions and principles underpinning the decision-making process.

- The regional Clinical Genetics Services are specialised services with finite and unique resources, and it is the responsibility of the Clinical Genetics workforce to ensure that resource utilisation is efficient, effective, and equitable.
- Referral guidelines should be based upon accepting referrals where the CGSs add the most value to patient care. Due to local resource constraints, differences in patient access to specialist clinics out with regional CGS provision, and the rapidly evolving nature of genomic medicine, this may differ between CGSs until staffing levels are in equilibrium with regional demand across all services.
- Referral guidelines should be dynamic, with regular review to incorporate new genomic evidence and technology advances and evolving inter-disciplinary collaboration.
- Referral guidelines will change as the roles of Clinical Geneticists and Genetic Counsellors and allied professionals evolve, for example with the increase in demand for lifelong complex case management, clinical trials, and provision of therapies.

4. Where does the Clinical Genetics workforce add value to patient care?

Clinical Genetics Services add value to patient care at multiple stages of the diagnostic and treatment pathway when the patient is suspected of, or has been diagnosed with, a genetic condition. The CGSs maximise the benefit to population health and economic value of a diagnostic genetic test result by ensuring that family members at risk are identified and are offered appropriate testing and follow up, and by facilitating pre-implantation or prenatal testing and other reproductive options to minimise transmission to subsequent generations, according to patient choice.

It is beyond the scope of this document to describe the different professional roles within regional clinical genetics services; however, each professional aims to work at the top of their scope of practice to maximise efficiency and protect scarce resource. In summary, the professional workforce consists of Genetic Counsellor and Clinical Geneticist roles, with recent addition of the Genomic Associate and Genomic Practitioner roles. An in-depth description of the scope of professional roles was published by the Association of Genetic Nurses and Counsellors, and the Clinical Genetics Society, in October 2020 https://www.agnc.org.uk/info-education/documents-websites/). This is currently being updated.

4.1 Diagnosing Rare Disease and Tumour Predisposition Syndromes

Clinical Geneticists and Genetic Counsellors receive referrals from colleagues in primary, secondary and tertiary care, requesting their expertise in diagnosis of rare disease and tumour predisposition syndromes. CGS clinicians may provide patient care via an inpatient or outpatient encounter, or in the setting of a multidisciplinary meeting. Referrals are triaged to the appropriate member of the CGS multiprofessional team, depending on the diagnostic requirements such as

- to integrate the patient's medical history and perform a physical examination as part of the diagnostic assessment.
- to integrate results of non-genetic investigations with the clinical and physical assessment e.g. imaging, biochemistry, pathology.
- to take and interpret a family history.
- to contribute expert opinion to inform genomic variant interpretation.

CGS clinicians are adept at rapidly synthesising large amounts of relevant and often complex medical and genetic information, to determine:

- the likelihood that the patient has a genetic disorder
- whether a patient requires a genetic test(s)
- whether a patient requires a non-genetic test(s)
- which test(s) to order
- when to order the test in the sequence of investigations
- the correct interpretation of the test(s) including both positive and negative monogenic genetic tests
- how genetic test results can be used to inform immediate and future patient management
- alternative diagnoses in the absence of a monogenic disorder e.g. teratogenic exposures, polygenic or multifactorial conditions

4.2 Managing Rare Disease and Tumour Predisposition Syndromes

Clinical geneticists and genetic counsellors play a critical role in facilitating the lifelong management of rare diseases and tumour predisposition syndromes, which are often multisystem disorders with a high risk of morbidity and mortality and may require multi-disciplinary team management. Many of these diseases have medical and psychosocial impacts which evolve over a patient's lifetime, as a result of additional organs becoming involved or a single-organ disease progressively worsening. For rare diseases and tumour predisposition syndromes, the appropriate management may change across the patient's lifespan, requiring regular review with increasing complexity of clinical care. A patient may require treatment by a medical or surgical specialist for one aspect of their condition whilst needing to undergo surveillance for other potential manifestations. Individual clinical specialties involved in the treatment of one manifestation of a rare disease typically do not perform this holistic overview of multi-system/organ management.

In addition, many genetic disorders have specific treatments which are only effective for a molecular sub-set of patients, requiring a deep molecular understanding of phenotypes to plan appropriate management.

Clinical Genetics Services create individualised management plans for patients with rare disease or tumour predisposition syndromes, whether the diagnosis has been established by a clinician in the CGS, or the patient is referred once the diagnosis is already established.

- CGS professionals are involved in determining individual treatment plans for patients based on the most recent evidence and the patient's age and current clinical status, which may also include facilitating participation in clinical trials.
- CGS clinicians play a pivotal role in multidisciplinary team (MDT) clinics for genetic diagnoses, for example in tumour predisposition conditions. This can involve ordering investigations according to

national or local surveillance programmes and referring to appropriate specialties when organ-specific manifestations of a condition begin to occur.
- Their strong communication and counselling skills combined with specialised scientific and medical knowledge are crucial for explaining complex genetic details to patients and their families, and for providing necessary support throughout the patient care journey.
- Their ability to synthesise and explain complex genetic information to other healthcare professionals and patients facilitates a deeper understanding and better care outcomes.

4.3 Diagnosing and managing at-risk family members

The CGSs are uniquely commissioned to focus on the medical and psychosocial impact of a genetic diagnosis for the entire family, not just the patient who receives the initial diagnosis. The impact of actively managing at-risk and affected family members increases the value of establishing a specific genetic diagnosis, both to the family and to the wider healthcare system. It enables at-risk or affected family members to be offered services such as: -

- timely information on diagnosis and treatment options, including predictive testing for high-risk tumour predisposition syndromes and other diseases where early diagnosis is proven to have patient benefit.
- pre-conception genetic counselling, to enable informed reproductive decision-making and to support choices including referral for prenatal genetic diagnosis or pre-implantation genetic diagnosis.

4.4 Navigating the complexities of the health, education and social care systems in partnership with patients

Clinical geneticists and genetic counsellors have a comprehensive understanding of the healthcare system and have strong links into diagnostic, management and treatment pathways. They facilitate appropriate referrals with accompanying educational resource for other healthcare providers and the family where required (frequently necessary for rare and ultra-rare diseases).

For some groups of patients, for example pre-symptomatic carriers of tumour predisposition syndromes, CGS are the only care providers currently commissioned to manage their surveillance until disease-specific manifestations require referral for active management. This function is extremely important in order to maximise the benefit of pre-symptomatic genetic testing. The impact of many rare diseases on intellectual development is significant, and CGS professionals advocate for patients to receive appropriate educational support by liaising with Educational Psychologists and educational providers.

The CGS teams recognise and address the social and financial impact of genomic disease, including the need for additional support in attending multiple hospital appointments and accessing social care services.

The CGS is often the source of disease-specific information for the patient and their family across their lifetime, and/or acts as the conduit to relevant and accurate sources of information and patient support, thereby empowering families to manage their disease in partnership with the professionals in their circle of care.

4.5 Education and Training, Research, Service Development and Equality, Diversity and Inclusion

CGSs act as subject matter experts and pathfinders in Service Development, enabling disruptive genomic testing technologies and research discoveries to be translated into routine clinical care in the NHS. Members of the Clinical Genetics workforce develop or co-create new care pathways to enable newly eligible patients to access tests.

- support mainstream colleagues by providing education, training and additional clinical input and management for complex cases
- enable regional standardisation of care pathways due to their regional remit

- develop new pathways in response to national guidelines e.g. NICE NG241 familial ovarian cancer
- expand the remit of CGSs beyond monogenic disorders when clinical validity and utility has been established e.g. risk estimation in breast cancer using multi-input risk modelling

Clinical geneticists and genetic counsellors play a critical role in systematically capturing genotype, phenotype, and longitudinal clinical data, by leading and contributing to disease-specific databases or research studies. This is exemplified by the requirement of clinical genetics services to submit details of all patients with genomically-proven tumour predisposition, to the National Disease Registry Service national cancer predisposition register, to ensure that their diagnosis is appropriately linked to evidence-based and clinically available screening. Understanding and documenting the natural history of any condition is a prerequisite to begin clinical trials with appropriate endpoints. It also enables the development of disease-specific clinical guidelines/recommendations and patient care pathways to inform evidence-based care for the benefit of patients, healthcare professionals, and health service commissioners.

CGSs provide an interface between clinical trials and rare disease patients by signposting their patients to appropriate trials. Increasing numbers of the Clinical Genetics workforce may develop expertise in medical trials as more genomically-driven treatments are developed.

Educational and clinical support from CGS teams to their mainstream colleagues supports the widespread adoption of genetic testing, interpretation of complex somatic and constitutional genetic test results, and incorporation of genetic data to inform personalised management. CGS clinicians also provide leadership in medical education and educational scholarship to enable agile, sustainable, and timely training to the specialist genomic workforce to keep pace in a rapidly evolving field.

CGS clinicians are aware of barriers to accessing healthcare, specifically in populations affected by rare disease, and are involved in the identification of unmet healthcare needs. CGSs adopt new ways of working to prevent discrimination, and advocate for additional resource to overcome such barriers.

5. Utilising the expertise within NHSE Clinical Genetics Services for maximum patient benefit

The expertise contained within the regional CGSs, both as individual professionals and their collective knowledge and functional networks, is both a specialised and finite resource. Many clinical geneticists and genetic counsellors contribute to supra-regional, national and international clinical patient care. The NHSE Regional Clinical Genetics Services therefore have a responsibility to direct their resources for maximum patient benefit.

Regional CGSs collectively acknowledge their responsibility to produce referral guidelines which support their mainstream colleagues in primary, secondary and tertiary care. If such guidelines are to be effective for resource allocation, they need to be matched by efficient and consistent triage and case management within the CGS. The following guidelines are not exhaustive and will require adaptations to allow for differences between demands placed upon regional services. It is also acknowledged that regional CGSs already have their own referral guidelines in place, and that adoption of the nationally developed guidelines will require time and resource. Furthermore, mainstream clinicians and clinical pathways will require a period of enhanced support and scaffolding to absorb additional demands on their own services where referrals cannot be accepted by the regional CGS.

5.1 Eligibility criteria for acceptance of referrals

1. Predictive genetic testing for a known genetic condition within an individual's family
2. Diagnosis of a suspected genetic disorder (rare/complex/multisystem)
3. Genotype-phenotype correlation where there is uncertainty regarding the diagnostic validity of an identified genetic variant
4. Patient with a likely pathogenic or pathogenic genetic variant requiring
    a. reproductive counselling
    b. expert phenotyping
    c. individualised management plan
5. Family members of an existing CGS patient in whom appropriate examination and investigations would assist in diagnosis for the patient
6. Fetal abnormalities and high-risk pregnancies
7. Pre-conception advice for parents at risk of having a baby with a severe childhood-onset disorder
8. When CGS input is a commissioned as a formal requirement in specific pathways (e.g. Fetal Medicine, NICU for rapid WGS pathway)
9. Patients in whom there is a strong clinical suspicion of a genetic disorder but where standard of care testing has failed to enable a diagnosis and where diagnosis may be made on a clinical basis or by orthogonal method(s)
10. Patients with a genetic disorder when they transition between child and adult services
11. Patients for whom genetic counselling and psychological support is required to aid management, or support complex personal or health-related choices

5.2 Criteria for declining referrals

1. Referring clinician can perform the investigation within their own scope of practise (e.g. a neurologist requesting a genetic test relating to neurological disorder)
2. Pharmacogenomic testing or interpretation of a pharmacogenomic test result in relation to prescribing specific drugs
3. Interpretation of a genetic test result generated by an unaccredited laboratory
4. Individuals with conditions with very low likelihood of being due to a specific genetic disorder
5. Individuals at moderate risk of breast cancer in the absence of a confirmed familial heritable genetic risk factor
6. Individuals with a somatic variant in a gene not known to be associated with non-syndromic cancer predisposition, not otherwise meeting criteria for germline genetic testing
7. Individuals with disorders for which genetic testing is not recommended based on the NGTD
8. Carrier testing for autosomal recessive disease in healthy individuals at population risk*
9. When a commissioned pathway exists in another service
10. Referrals for the following conditions
    a. Hemochromatosis
    b. Hypermobile-type Ehlers-Danlos syndrome
    c. Alpha-1 antitrypsin deficiency
    d. Familial Hypercholesterolaemia
    e. Haemoglobinopathies
    f. MTHFR
    g. Thrombophilia

*Referral for autosomal recessive carrier testing would be accepted as part of prenatal genetic counselling or reproductive planning under certain circumstances.

6. Next steps to support the effective implementation of national referral guidelines

6.1 Accessibility of referral guidelines and process

Referral guidelines, processes and forms must be easily accessible and ideally in a recognised or standardised format for all referring clinicians. This may require individual CGSs to review the following elements of their service: -

- The current platform or system in place for accessing referral guidelines and making referrals to Clinical Genetics services by primary, secondary and tertiary care colleagues.
- The clarity of the processes requested (e.g. family history form completion)
- The integration of CGS referral guidelines and forms into existing referral systems (e.g. South and West Devon Formulary and Referral system)

6.2 Mainstreaming Genomics in collaboration with the Genomic Medicine Service Alliances

CGSs wish to work alongside the GMSAs to determine their required contribution to mainstreaming. Clinical Geneticists and Genetic Counsellors suggested the need to develop a nationally agreed data set of conditions that have been, or will be, mainstreamed, in order to enable national agreement on resource requirement and allocation.

Preconditions to equitable access to mainstream genomic testing may include the development of new competencies in mainstream colleagues, national education and training resources, patient resources such as FAQs and the availability of appropriate time and human resource within appointments, recognising the additional work required to support patients in making informed choices.

6.3 Consent

National consensus is required with respect to standardisation of consent as it applies to genomic tests, across CGSs and mainstream services. Further work is required to standardise the consent process to ensure ethical and legal compliance, and clarity for clinicians and patients.

6.4 Standardisation of the triage process

National referral guidelines provide the first and critical step in defining which patients will benefit from gaining access to CGSs, however their implementation requires a consistent and transparent approach to triaging referrals. Equitable access and efficient resource management are foundational goal of an effective triage process.

The following requirements were discussed, and will require further focus: -

- The need for local triage Standard Operating Procedures (SOPs), including defining the competencies required to participate in the triage process
- Training and education to allow CGS clinicians to perform triage consistently

6.5 Regional Considerations

Region-level variation in factors such as financial flow-through, demand and capacity of each CGS will impact the implementation of national referral guidelines. Understanding and addressing regional nuances are key to successful referral guideline implementation. The NHSE Genomics Unit (the GU, responsible for delivering the NHSE Genomics Programme) is currently developing a minimum dataset for commissioning purposes, in order

to provide evidence for the service developments required to match the demands of an increasingly genomically- informed healthcare system. The GU is also carrying out a workforce survey and developing a workforce model, which will allow comparison of capacity in the regional CGSs.

The group suggested that additional work may be required to inform the variance between services' abilities to implement the national guidelines. This would be a comprehensive analysis of regional and national variations which impact upon a service's ability to follow national referral guidelines, feeding back to the Genomics Unit to inform future region-specific commissioning requirements.

6.6 Proposals for inclusion in a future costing model

The group discussed which distinct episodes of care delivered by the Clinical Genetics Service should be considered during the development of a new tariff, which would be applied consistently across all CGSs. The following could be captured as discrete patient care episodes, requiring clear definitions:

- Face-to-Face
- Virtual
- Inpatient (ward) referrals
- Advice & Guidance (chargeable letters)
- MDT coordination
- MDT/GTAB attendance
- Surveillance management
- Carrier management
- Virtual case review
- Provision of advice to laboratories reporting on somatic genomic variation

APPENDIX

CLINICAL GENETICS SERVICES

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W: https://www.ouh.nhs.uk/services/referrals/genetics/clinical-genetics.aspx

Sheffield
Sheffield Clinical Genetics Services, Sheffield Children's NHSFT, Northern General NHS Trust, Herries Road, Sheffield S5 7AU
T: 0114 271 7025
E: Sheffield.clinicalgenetics@nhs.net
W: https://www.sheffieldchildrens.nhs.uk/services/clinical-genetics/

Southampton
Wessex Clinical Genetics Service, Mailpoint 627, Princess Anne Hospital, Coxford Road, Southampton SO16 5YA
T: 023 8120 6170
E: GeneticsTeam@uhs.nhs.uk
W: www.uhs.nhs.uk/genetics

SCOTLAND

Aberdeen
North of Scotland Genetic Service, Clinical Genetics Centre, Ashgrove House, Foresterhill, Aberdeen AB25 2ZA
T: 01224 552120 01463 705823
E: nhsh.clinicalgenetics@nhs.scot
E: gram.clinicalgenetics@nhs.scot
W: https://www.nhsgrampian.org/service-hub/north-of-scotland-medical-genetics/

Dundee
East of Scotland Genetic Service (Dundee), Ninewells Hospital, Dundee DD1 9SY
T: 01382 632035
W: https://www.nhstayside.scot.nhs.uk/OurServicesA-Z/Genetics/index.htm

Edinburgh
Lothian NHS Board, Waverleygate, 2-4 Waterloo Place, Edinburgh EH1 3EG
T: 0131 536 9000 /T: 0131 537 1116
E: wgh.clinicalgenetics@nhslothian.scot.nhs.uk
W: https://services.nhslothian.scot/geneticservice/clinical-genetic-service/

Glasgow
West of Scotland Genetic Services, Level 2A, Laboratory Medicine, the Queen Elizabeth University Hospital, 1345 Govan Road G51 4TF
T: 0141 354 9201/T: 0141 354 9207 /T: 0141 354 9300
E: GeneticsReferrals@ggc.scot.nhs.uk
W: https://www.nhsggc.scot/staff-recruitment/staff-resources/laboratory-medicine/clinical-genetics/

IRELAND

Belfast
Department of Medical Genetics, A Floor, Belfast City Hospital, Lisburn Road, Belfast BT9 7AB
T: 028 9504 8022; T: 028 9032 9241
E: genetic.medicine@belfasttrust.hscni.net
W: https://belfasttrust.hscni.net/service/laboratory-services/clinical-genetics/

Dublin
Department of Clinical Genetics, Children's Health Ireland (CHI) at Crumlin, Crumlin, Dublin, D12 N512, Ireland
T: + 353 1 409 6739
W: https://www.childrenshealthireland.ie/list-of-services/clinical-genetics/

WALES

Cardiff
All Wales Medical Genetics Service (AWMGS): Wales Genomic Health Centre, Cardiff Edge Business Park, Longwood Drive, Whitchurch CF14 7YU
T: 029 2183 4000
E: se.genetics@wales.nhs.uk
E: SW.Genetics@wales.nhs.uk
E: North.Genetics@wales.nhs.uk
W: https://medicalgenomicswales.co.uk

This list can also be found on the BSGM website by scanning this QR code.