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Evolving Roles Within the Genomic Medicine Workforce: A Revised Framework for UK Practice

Abstract

Background: The delivery of genomic medicine in the UK continues to evolve in response to technological advancements, increasing demand, and national strategic priorities. This paper presents an updated framework describing the professional roles within specialist genomic services, refining responsibilities across key domains of clinical care, education, research, and mainstreaming.

Methods: Building upon previous role descriptors, this updated framework integrates national consultation, emerging service models, and policy alignment. A new colour-coded matrix delineates role-specific scope of practice for Clinical Geneticists, Genetic Counsellors, Genomic Associates, and Genomic Practitioners.

Results: A key development is the formal inclusion of the Genomic Practitioner role, reflecting workforce diversification. Expanded emphasis is placed on mainstreaming genomic services and involvement in clinical and psychosocial research. The framework remains aspirational and adaptable, acknowledging regional variability and anticipating future service developments.

Conclusion: This updated framework provides a structured and transparent tool for workforce planning, interprofessional collaboration, and service development across genomic medicine. It aligns with national genomic strategies and supports the equitable and efficient delivery of genomic care.

Introduction

The field of genomic medicine is undergoing rapid transformation, requiring continual re-evaluation of workforce structures and professional roles. This article presents an updated framework for defining and differentiating the roles of professionals within UK specialist genomic medicine services. The framework reflects the growing complexity of clinical genetics practice, increased integration with mainstream healthcare, and the formal recognition of emerging roles such as the Genomic Practitioner.

This iteration builds on earlier frameworks that focused on individual professional role descriptions. In doing so, it refines the scope of practice and responsibilities for Clinical Geneticists, Genetic Counsellors, Genomic Associates, and Genomic Practitioners. It aims to promote clarity, efficiency, and adaptability within multidisciplinary teams providing genomic services across the NHS.

To facilitate a clear interpretation of role responsibilities, a **color-coded system** has been implemented:

- **Green** – Activities routinely within the professional group's scope of practice.
- **Amber** – Activities that may be undertaken by some professionals but are not routinely performed across the group.

- **Red** – Activities outside the routine scope of practice for the professional group.

We understand that across the United Kingdom, and in fact across different regions within England, commissioning structures and constraints on workforce means that the roles below are exceeded in some capacity, or there remains some overlap.

Crucially, this framework is **aspirational**—it anticipates the continued evolution of genomic services and provides a strategic vision for role adaptation, streamlining responsibilities, and optimizing patient care. As genomic medicine progresses, this document serves as a foundational guide to aligning professional practice with emerging healthcare needs and best practices across the NHS and beyond.

Key Updates: Mainstreaming and Research

A notable advancement in this update is the expanded focus on **Mainstreaming**, which emphasizes the integration of genomic expertise within broader healthcare settings. This document provides enhanced clarity regarding the role of genomic professionals in supporting mainstream clinicians, including guidance on referral pathways, genomic result interpretation, and multidisciplinary collaboration. Furthermore, the responsibilities of managing and leading specialist nurse-led mainstreaming teams (e.g., *Familial Hypercholesterolemia clinics, family breast screening services, and pre-implantation genetic diagnosis in IVF clinics*) have been more explicitly defined. As anticipated, 50% of healthcare interactions in 10 years will have a genomic component which emphasises the need to extent our practice into the mainstream. [link to genomic strategy]

The **Research** section has also been expanded, reflecting the increasing involvement of genomic professionals in *leading, supporting, and referring patients to research studies*. This update portrays specific responsibilities concerning *gene therapy trials, psychosocial research, and variant interpretation studies*. Additionally, it underscores the growing emphasis on evidence-based genetic counselling, ensuring that clinical practice is informed by rigorous research methodologies and empirical findings.

This framework aligns with the perspectives outlined in the Clinical Genetics Society's 2024 report, "Maximising the Patient Benefit of Genomics – the evolving role of the Clinical Genetics Services," which emphasizes the need for evolving models of clinical care delivery to maximize patient benefits from advancing genomic technologies. [Link]

Area	Specific activity	Clinical Geneticist	Genetic Counsellor	Genomic Associate	Genomic Practitioner
Triaging referrals	Referrals are assessed and triaged				
Advice and guidance letters for refused referrals	Letters written in response to referrals that require clinical advice, but do not meet GMS referral guidelines				

Area	Specific activity	Clinical Geneticist	Genetic Counsellor	Genomic Associate	Genomic Practitioner
	Standard letters based on clinical triage decisions e.g. referral to moderate risk breast screening				
	Responsibility for responding to referrals that do not require any clinical advice nor clinical contact				
Access to the appointment	Facilitating patient access, including establishing if patient wants to be seen, supporting minority populations to access services, supporting patients with disabilities/audio/visual impairment to access services, contacting patients to explain what clinical genetics can offer, arranging interpreters				
	Acting as a chaperone in clinic				
	Arranging or obtaining measurements or obtaining relevant samples prior to clinic, e.g. taking patient's weight and height or accessing tumour samples for future testing				
Preparation for appointment	Transcribing a written pedigree into electronic software				
	Gathering relevant medical records, pathology reports, death certificates, tumour blocks and associated information needed for appropriate clinical assessment and counselling				
	Obtaining relevant samples to help confirm diagnosis in proband				
	Collating appropriate patient leaflets, consent forms for the clinic as determined by senior staff				
Patient/family type seen	General genetics (adult or paediatric)				
	Cancer genetics (adult or paediatric)				
	Prenatal				

Area	Specific activity	Clinical Geneticist	Genetic Counsellor	Genomic Associate	Genomic Practitioner
Physical medical examination	Examination of a patient to make a clinical diagnosis and/or to support or stratify genetic testing				
	Specific physical examination that might be considered routine with respect to particular conditions (e.g. head measurement for a Cowden's clinic)				
Family history	Taking a family history				
	Reviewing and evaluating a family history to determine genetic risk				
Psychosocial history	Taking a detailed psychosocial history to determine effect of genetic diagnosis on individual and wider family members				
Investigations	Medical investigations: Employ a range of tailored investigations including genetic, biochemistry, radiology, haematology etc for clinically undiagnosed patients				
	Routine medical investigations for specific, defined conditions, e.g. ophthalmology or audiological investigations as part of conditions involving visual and/or hearing impairment				
	Genetic investigations: Choose appropriate genetic testing for patients with specific family history indicative of genetic risk (e.g. family history of cancer)				
	Arrange tumour tissue testing or somatic testing according to genomic testing directory or clinical indications. [Consent for this described below]				
	Genetic investigations: Choose appropriate genetic testing determined by a pre-existing definitive clinical diagnosis/clinical presentation (e.g. breast cancer)				

Area	Specific activity	Clinical Geneticist	Genetic Counsellor	Genomic Associate	Genomic Practitioner
	Arrange for parental/relative samples for segregating variants of uncertain significance following MDT or clinical discussion				
	Take samples (e.g. blood saliva) for genetic testing				
	Taking skin samples for genetic testing				
Consent	Holds informed consent conversation with a patient for genetic testing				
	Recording of consent for testing after patient has discussed with clinician				
	Arrange and consent for predictive/presymptomatic testing				
	Arrange and consent for cascade genetic testing amongst extended family (e.g. BRCA, Fra-X testing)				
Counselling and support	Generic genetic counselling skills e.g. disclosure of diagnosis, breaking bad news etc.				
	Supporting patients and families adjusting to a genetic diagnosis or coping without one				
	Making appropriate onward referrals for further psychological support				
	Identifying complex grief reactions and interpreting complex family dynamics				
	Specific application of genetic counselling theory to person-centred care, e.g. application of reciprocal-engagement models and/or reflective practice models				
Genomic variant interpretation	Interpreting gene variants to determine clinical decisions, as part of a multidisciplinary team				
	Integrating the results of clinical presentation and investigation to determine whether a rare phenotype supports variant pathogenicity				

Area	Specific activity	Clinical Geneticist	Genetic Counsellor	Genomic Associate	Genomic Practitioner
	Interpreting whether an established clinical presentation supports variant pathogenicity				
	Reviewing 100KGP data in Clinical Interpretation Portal or Genomics England Research Environment for diagnosis				
	Administration to track down relatives to provide evidence in support of variant interpretation				
Management and Treatment	Reviewing and recommending peer-reviewed management guidelines. Writing, e.g. NICE guidance				
	Organising appropriate disease screening and acting as patient advocate to arrange access to services				
	Devising individual management guidelines for a rare disorder based on research evidence				
	Prescribing pharmaceuticals or molecularly targeted therapies				
	MDT coordination, collating agenda items, taking meeting minutes				
	All administration required for clinic and follow up				
	Ordering of clinic supplies, test kits, appropriate proformas, consent forms				
Follow Up	Follow up care of the nuclear family (e.g. parents and children)				
	Follow up care of the extended family (e.g. 2nd and 3rd degree relatives)				
	Monitoring/chasing outstanding records/samples/screening and any administration work needed to support the clinical geneticists and genetic counsellors				

Area	Specific activity	Clinical Geneticist	Genetic Counsellor	Genomic Associate	Genomic Practitioner
Research	Leading or referring to research studies relating to patient's genetic diagnosis				
	Leading on gene therapy trials as PI (ATMPs)				
	Leading or being a site investigator for Clinical Trials of Investigational Medical Products				
	Providing genetic counselling to patients taking part in clinical trials				
	Finding and referring to surveillance trials (e.g. for cancer screening)				
	Referring to psychosocial research (e.g. genetic counselling or communication research)				
	Leading genetic counselling research specifically on the evidence base behind genetic counselling practice				
	Administration for research studies				
	Undertake research and quality improvement schemes to determine the evidence for changes to clinical practice to ensure optimal patient care.				
Mainstreaming	Providing advice and support to other healthcare workers e.g. when it is appropriate to refer back to CGS for at risk family members				
	Support mainstream services with clinical interpretation of genomic testing				
	Participation in multi-disciplinary team meetings				
	Managing and leading a specialist nurse mainstreaming team (e.g. Familial hypercholesterolemia clinic, family breast				

Area	Specific activity	Clinical Geneticist	Genetic Counsellor	Genomic Associate	Genomic Practitioner
	screening clinics, pre-implantation genetic diagnosis within an IVF clinic)				
Education	Respond to requests for education for patients and public				
	Organise and deliver education for current and/or prospective health professionals				
	Developing educational material such as leaflets, interactive infographics and decision aids				
	Liaising with patient support groups to participate in patient led events and sharing of verified information				
Management	Running a genetic register				
	Training and mentoring colleagues from genetics services				
	Training, mentoring and supporting non-genetics healthcare colleagues				
	Acting as Clinical Lead for a clinical genetics service				
	Acting as management lead for clinical genetics service				
Leadership	Sitting on regulatory bodies for own profession				
	Contribute to national networks relevant to own profession				
	Designing professional competency to practice frameworks				

Conclusion:

This updated framework represents a critical step in aligning genomic workforce practice with the growing demands and opportunities of genomic medicine. The clear delineation of roles supports interprofessional collaboration, role optimisation, and equitable task distribution. It also supports workforce planning, particularly in light of the introduction of roles such as Genomic Practitioners and increasing engagement with mainstream services.

This revised framework provides a structured foundation for understanding and evolving roles within genomic services. It has been developed by experts working within and outside the clinical genomic services. We hope it supports the delivery of high-quality, patient-centred care and promotes the integration of genomics across the NHS and broader health system.