Genomics Education Programme

NHS
Health Education England

A cross-professional competency framework for communicating germline genomic results

Why is this framework needed?

The NHS in England is implementing a Genomic Medicine Service, leading to the increased utilisation and mainstreaming of genomic testing across a growing number of specialties. This has resulted in a requirement for workforce development to ensure all healthcare professionals who access genomic testing are competent to do so and are able to address the unique considerations of genomics, including the uncertainty of genomic information, complexity of results and impact on the wider family.

In 2019, the GEP published a competency framework for facilitating genomic testing, including the knowledge, skills and behaviours needed to request and consent patients to a genomic test. There is also a need to establish competencies for communicating the results of a genomic test, as this can be a complex and challenging time for patients and family members.

What is the framework?

This framework outlines six areas of proficiency for healthcare professionals who are responsible for communicating germline genomic results. It is aimed at healthcare professionals currently working in the NHS, and is intended as a cross-professional guide for best practice. In order to demonstrate the potential use of this framework across a range of tests and areas of practice, the competencies have been mapped against a matrix, outlining three broad levels based on the type of genomic tests and associated competencies required. Of note, these levels do not pertain to specific role titles or professions, and should be considered based on the context of the test and scope of practice.

How can this framework be used?

The framework can be used by individual healthcare professionals as a guide to help them identify their learning needs. For educators and trainers, the framework provides a mechanism to recognise the training needs of health professional groups, and to structure training so that conversions about genomic test results can be delivered consistently across different specialties. This framework will also guide the development of future tools made by the GEP to support the NHS workforce.

What input do we need?

Throughout 2019 and 2020, meetings took place with a range of stakeholders including healthcare professionals in various disciplines, as well as patient and public contributors. The framework was developed via a consensus approach based on the nominal group technique, including reviewing clinical scenarios in iterative rounds, mapping themes and voting on areas of inconsistency. Before this framework is finalised, we are inviting individual healthcare professionals and professional bodies to review the current framework. The final version will be published along with case studies of how the framework can be used in practice.

Stages in the return of results pathway

As part of the development process for a competency framework about returning genomic results, stakeholders identified six stages in the return-of-results pathway where healthcare professionals (HCPs) are involved. Of note, these do not encompass the existing roles, professional standards and competencies for clinical scientist and other laboratory staff.

1. HCPs who have a formal role that involves genomic data interpretation in the lab (including patients for which they have no direct clinical responsibility).
2. HCPs interacting with genomic data directly and independently to solve a clinical question about patients for which they have clinical responsibility.
3. HCPs contributing to the genotype-phenotype correlation and reporting decisions using their clinical expertise (as part of multidisciplinary discussion).
4. HCPs responsible for communicating genomic results to the patient and/or family members.
5. HCPs involved in ongoing clinical management of the patient (part of more general/group-specific genomic knowledge competencies).
6. HCPs directly involved in managing genetic aspects of the patient's condition.

The scope of this current framework is focussed on stage 4, for HCPs responsible for communicating genomic results to patients and family members.

Key points to consider prior to reviewing the framework

- The framework is aimed at healthcare professionals across specialities and roles that are currently practicing in the NHS.
- In order to demonstrate the potential use of this framework across a range of tests and areas of practice, the competencies have been mapped against a matrix, outlining three broad levels based on the type of genomic tests and associated competencies required. These levels do not pertain to specific role titles or professions, and should be considered based on the context of the test and scope of practice.
- It is important for any healthcare professional to seek guidance from colleagues with a higher level or different area of expertise (including genomic specialist clinicians, laboratory healthcare scientists, clinicians specialising in the management of a condition) regarding complex results or their implications where needed.
- Standard clinical competencies (e.g. regarding patient communication) have not been included within this framework as they are already expected to be part of clinical practice. As well, this framework includes but is not limited to existing competencies for genomic specialists (e.g. consultant clinical geneticists, genetic counsellors).

Competencies for communicating genomic results

Level 1: Results for defined genomic tests, including targeted variant analysis or analysis of a select group of single genes

(E.g. haemochromatosis targeted variant analysis requested in primary care; qfPCR analysis requested by midwives; familial hypercholesterolaemia testing requested by specialist nurses).

Level 2: Results for genomic tests with a broader scope that may involve more complex or uncertain results and carried out by mainstream healthcare professionals

(E.g. microarray requested in paediatrics, large panels requested in cardiology or neurology).

Level 3: Results for genomic tests with a broader scope that may involve more complex or uncertain results and carried out by genomic specialist healthcare professionals, where these competencies would encompass existing professional competence

(E.g. large panels, exome or genome sequencing requested by clinical geneticists and/or genetic counsellors).

X = not expected to possess this competence. Each level subsequently builds on each other from left to right across the table (e.g. Level 3 would be competent in areas outlined for Level 1 and 2). Where there are no additional comments, the competency is expected to apply to all levels.

	Competency	Level 1	Level 2	Level 3
1	**Recognises scope of practice and seeks further assistance where relevant.**			
	Knows how to contact their local Genomics Laboratory Hub, Clinical Genetics service, and additional clinical services where relevant.			
	Recognises one's professional responsibilities and boundaries, and when to refer to relevant specialists for further support, including: • cases that would benefit from specialist genetics involvement, such as genetic counselling to understand or adapt to their result; • Genomic Laboratory Hub colleagues for updating a patient's phenotype, requesting re-analysis of data, or follow-up studies (e.g. segregation or functional assays); and • other clinical services for clinical patient management.			
	Collaborates with other specialists by requesting and/or attending multidisciplinary review meetings as appropriate.			
	Knows how to access educational resources to support continuing professional development.			

2	**Plans for the provision of the genomic result in the context of the patient's current clinical pathway.**			
	Assesses what took place at the time of consent, with respect to information that would have been conveyed to the patient prior to the genomic test (e.g. information about the condition and the genetics/inheritance).			
	Understands the outcomes of any multi-disciplinary discussion(s) that may have taken place to inform the patient's genomic result.	X (Unlikely to be applicable or a requirement to synthesize this information)		
	Obtains any new information learned about the patient's phenotype and/or family history that may inform the results conversation.	X (May not have wider access to patient records or required expertise to synthesize new information about patient or family history in context of the result)		
	Plans for an appropriate mechanism, environment and amount of time to convey the genomic result (e.g. letter, telephone, face to face, joint consultation with other relevant specialists).			
3	**Applies knowledge to understand the genomic result and its implications.**			
	Assesses the suitability of the genomic result in relation to the patient's clinical situation.	Checks that the test is in line with what was requested, but seeks further guidance with regards to complex results, phenotype, family history, or applying further assay/panels.	Assesses whether the test was appropriate given the patient's phenotype, correlates with known patient and family information and whether any results would be considered incidental or additional findings.	Applies knowledge with regards to the capability and limitations of genomic tests (e.g. the assay used, gene coverage, panels applied, expected diagnostic yield), and the implications this could have for clinical

				management or re-analysis of data.
	Understands the outcome of variant classification as outlined on the laboratory report, including that the interpretation of a result can change over time.	Recognises variant classification terms and that a genetic cause is not ruled out if no variants are found (e.g. in a diagnostic test context).	Has a general understanding of variant classes and the terminology used (e.g. benign, variant of uncertain significance, pathogenic).	Applies in-depth knowledge of the principles of variant classification and relevant guidelines (e.g. Association for Clinical Genomic Science).
	Understands the implications of the result for clinical care, including referring to relevant management guidelines.	Understands guidelines within area of specialty but seeks further advice where relevant (e.g. aspects outside of specialty, rare syndromes, or where specific guidelines do not exist)		
	Initiates next steps that may be needed to clarify a genomic result where relevant, including those outlined in the laboratory report.	X (Seeks advice from or refers to colleagues regarding a plan to action any next steps.)		
4	**Communicates the genomic result and its implications in relation to the patient's clinical symptoms and/or family history.**			
	Understands general genomic concepts in order to facilitate the results conversation.			
	Explains the result in context of the reason for testing (e.g. whether for diagnosis, carrier or predictive status).			
	Describes the confirmed or suspected inheritance pattern (with numerical figures where relevant), including risks to other family members.			
	Outlines any implications for current or future clinical management (or lack thereof), including potential advantages and/or disadvantages of	Understands management implications within area of	Understands management implications	

	different options.	specialty and refers to relevant colleagues for areas beyond scope of practice.	within area of specialty and refers to relevant colleagues for areas beyond scope of practice.	
	Conveys the scope and limitations of the genomic result, including potential additional or alternative explanations.	Understands the impact of these factors (e.g. variable expressivity or incomplete penetrance) within scope of practice and based on the type of test, but may discuss with or defer to other colleagues for further discussion.	Understands the impact of these factors (e.g. variable expressivity or incomplete penetrance) within scope of practice and based on the type of test, but may discuss with or defer to other colleagues for further discussion.	Applies in-depth knowledge of these factors in order to convey these concepts and plan next steps as appropriate.
	Emphasizes the uncertainties that exist with regards genomic results as this applies to current and future management.			
	Understands the implications of genomic testing for insurance, including the UK Code on Genetic Testing and Insurance			
	Describes the wider familial impact of a result, including clinical management and genetic testing implications	Conveys that a result has family implications.	Conveys that a result has family implications and may facilitate further discussion depending on scope of practice.	
	Outlines any reproductive options that may or may not be available based on the result.	Describes that there may be reproductive implications.	Describes that there may be reproductive implications.	
5	**Identifies and explains next steps related to the genomic result.**			
	Outlines any further genomic testing and/or clinical investigations that may be considered based on the genomic result, including conveying when no further tests are anticipated at the present time.	X (Refers to colleagues to carry out further tests).	Seeks advice from or refers to colleagues to carry out further tests outside of area of practice.	
	Recognises the potential for research initiatives that may be relevant to			

	the patient based on the genomic result.			
	Identifies relevant patient resources and support groups.			
	Provides a copy of the result to the patient and/or family in writing (e.g. by letter or copy of genomic report).			
	Acts on clinical guidelines (nationally and locally) and facilitates onward referrals to other specialists as required, regarding: • clinical management and treatment; • discussion about familial implications including genomic testing in adults and children; and • discussion of reproductive options	Understands and acts on guidelines within area of specialty but otherwise refers to relevant colleagues to discuss available options.	Understands and acts on guidelines within area of specialty but otherwise refers to relevant colleagues to discuss available options.	
	Communicates a clear plan for follow-up, including the time frame (e.g. specific or open-ended), mechanism (e.g. telephone or face to face), and appropriate contact information for any queries.			
	Describes the potential for future changes based on the result (e.g. to treatment options, variant classification or new information identified through research) and where the patient may be re-contacted, while appropriately managing expectations.	Conveys that knowledge about the result may change, but not expected to discuss these concepts in depth.		
	Conveys the importance of the patient and family notifying of any changes in their medical or family history, as this may have an impact on their genomic result or future care.			
	Documents clinical contact about the genomic result appropriately, including: • recording the patient's diagnosis and result appropriately in relevant patient record systems; • recording patient diagnosis in registries with appropriate consent and where available; and • communicating to relevant professionals involved in the wider care of the patient.			

	Understands one's ongoing responsibilities to the patient, where duty of care may extend beyond the initial feedback of genomic findings.			
6	**Applies core clinical skills to support the patient and family in adapting to the genomic result. *** ** Note that this competency does not include core clinical skills that healthcare professionals are already expected to have and employ with patients in any area of practice, including:* • *Tailoring discussions based on the age, language and cognitive abilities of the patient and any other individuals present* • *Assessing the patient's understanding, reaction, concerns and expectations in light of the result*			
	Addresses the different types of uncertainty raised by the genomic result.			
	Considers the factors that may influence an individual's response and adaptation to a genomic result, including: • additional physical and mental health history; • cultural, religious, familial and personal values; and • timing of the conversation with respect to the patient's care and/or other life events	Provides support across these areas depending on the context and complexity of any issues and time available; however, refers to specialists such as genetic counsellors who can further address these areas.	Provides support across these areas depending on the context and complexity of any issues and time available; however, refers to specialists such as genetic counsellors who can further address these areas.	
	Recognises where further support is needed with regards to the patient and family members' reaction, expectations, concerns and coping in light of the genomic result.			
	Explores communication of the result within the family, including supporting the patient to share information and addressing any barriers to communication.			
	Employs effective communication skills to support families with the immediate and longer-term psychosocial impact of a genomic result for the individual and family, including a 'normal' or 'negative' result.	Recognises when further support may be required for onward referral to specialist colleagues.	May provide some support depending on the context and complexity of any issues and time available; however, refers to specialists colleagues where relevant.	