A cross-professional competency framework for communicating germline genomic results

This competency framework outlines six areas of proficiency for healthcare professional 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. 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.

Table 1. Broad levels based on types of genomic tests, for which some of the competencies for communicating genomic results have been further specified

Level	Description	Examples in practice
1	Results for defined genomic tests, including targeted variant analysis or analysis of a select group of single genes.	• Haemochromatosis targeted variant analysis requested in primary care • qfPCR analysis requested by midwives • Familial hypercholesterolaemia testing requested by specialist nurses
2	Results for genomic tests with a broader scope that may involve more complex or uncertain results and carried out by mainstream healthcare professionals.	• Microarray analysis requested in paediatrics • Large gene panels requested by specialists in cardiology or neurology
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 be expected to encompass existing professional competence.	• Large gene panels, exome or genome sequencing requested by clinical geneticists and/or genetic counsellors

Notes:

- ✓ = The competency is expected to apply as described without further clarification.
- Where additional comment is given, each level builds on each other from left to right across the table (so Level 3 would also be competent in areas outlined for Levels 1 and 2).
- **X** = Not expected to possess this competence.

	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 aid patients in understanding or adapting to their result; • Genomic Laboratory Hub colleagues for updating a patient's phenotype, requesting re-analysis of data, or follow-up studies (such as segregation or functional assays); and • other clinical services for 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 information that would have been conveyed to the patient prior to the genomic test (such as information about the condition, test, genetics and inheritance).	✓	✓	✓
	Understands the outcomes of any multidisciplinary discussion(s) that may have taken place to inform the patient's genomic result.	✓	✓	✓
	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 (such as 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 and applies knowledge with regard to capability and limitations of the test. Depending on level of experience, may seek further guidance with regard to complex results, phenotype, family history, or applying further assay/panels.	Assesses whether the test was appropriate given patient phenotype, known family history and whether any results would be considered incidental or additional findings.	Applies knowledge with regard to the capability and limitations of genomic tests (such as 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.	Recognises variant classification terms and that a genetic cause is not ruled out if no variants are found (such as in a diagnostic test context).	Has a general understanding of variant classes and the terminology used (such as benign, variant of uncertain significance, pathogenic).	Applies in-depth knowledge of the principles of variant classification and relevant guidelines (such as 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 (such as 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 (such as 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 different options.	Understands management implications within area of specialty and refers to relevant colleagues for areas beyond scope of practice.	Understands management implications 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 (such as 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 (such as 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.
	Emphasises the uncertainties that exist with regard to genomic results as this applies to current and future management.	✓	✓	✓
	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.	✓
	Understands the implications of genomic testing for insurance, including the UK Code on Genetic Testing and Insurance.	✓	✓	✓

5	**Identifies and explains next steps related to the genomic result.**			
	Outlines any further genomic tests and/or clinical investigations that may be considered based on the genomic result, including 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 (such as by letter or copy of genomic report).	✓	✓	✓
	Acts on national and local clinical guidelines and facilitates onward referrals to other specialists as required. Depending on area of specialty, onward referrals may be regarding: • clinical management and treatment; • discussion about familial implications, including genomic testing in adults and children; and • discussion of reproductive options.	✓	✓	✓
	Communicates a clear plan for follow-up, including the time frame (specific or open-ended), mechanism (such as telephone or face to face), and appropriate contact information for any queries.	✓	✓	✓
	Describes the potential for future changes based on the result (such as 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.	✓	✓	✓
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 a 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 regard 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' 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 specialist colleagues where relevant.	✓