A competency-based approach to defining training needs in clinical bioinformatics

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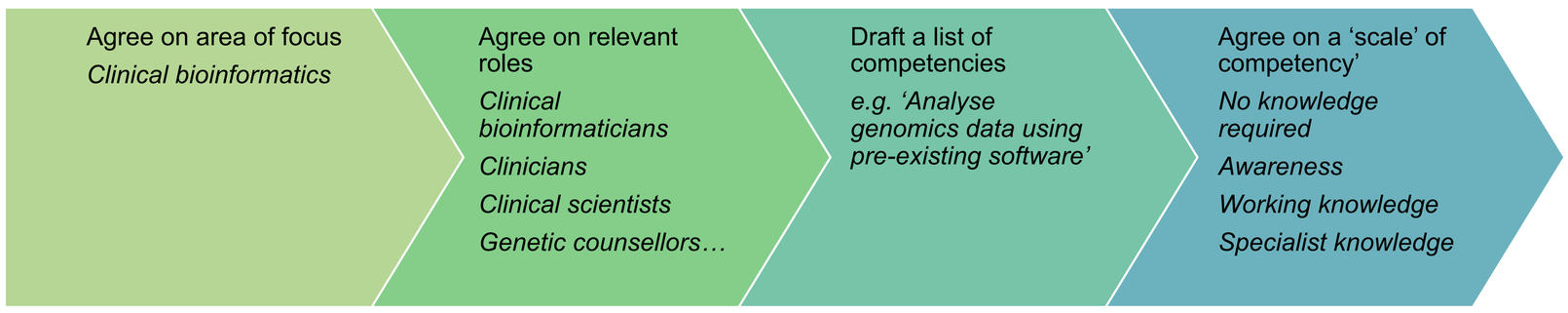
To fully realise the potential of genomic medicine and of other high-throughput molecular approaches to the prediction, diagnosis and management of disease, it will be necessary for both the current and the future generation of healthcare professionals to work more closely with bioinformatics data and systems – and with bioinformaticians. Depending on the role of the individual, training needs vary from awareness of the role of bioinformatics in the clinic to the development and implementation of new methods to analyse and interpret clinical bioinformatics data. To analyse training needs and map these to currently available training, we have used LifeTrain's principles to draft a competency framework for bioinformatics as it relates to genomic medicine. We refined the framework by seeking input from five or more practitioners or experts in each specified role. Our goal is to develop this draft competency matrix into a flexible tool – both to enable training professionals to identify gaps in existing training programmes so that they can be rapidly filled, and to enable individuals to plan their professional development by finding training that plugs competency gaps. As a proof of principle we have performed a preliminary mapping of the matrix to three training programmes developed by Health Education England.

Defining the scope of the profile

This work was performed by the Clinical Bioinformatics Task and Finish Group commissioned by Health Education England to provide recommendations on bioinformatics training requirements for the UK's National Health Service. Our remit was to identify the bioinformatics training requirements to support the 100,000 Genomes Project in the short term and the adoption of genomic medicine in the long term.



Creating and refining the matrix

We based our initial competency profile on existing lists of competencies required for bioinformatics (Welch, L. et al.) and clinical bioinformatics (Raza, S.) This was refined through discussion with Task and Finish group members. We then created a blank matrix with roles as columns and competencies as rows. Group members and their colleagues used this to define the competencies that they considered necessary for healthcare professionals to use data emerging from the 100,000 genomes project to inform clinical decision-making. We also asked all participants to add any competencies that they considered to be missing from the profile. We collated input from all participants (at least five for each role) into the matrix depicted on the right.

	Role	Clinical bioinformatician	Other bioinformatician	Specialist clinician with genetics/genomics expertise	Other specialist clinician	Other clinician	Clinical genetic Scientist	Other healthcare scientist	Specialist nurse/counsellor	Nurses and other allied health professionals	IT specialist	Data specialist
	No. responses	11	6	5	5	6	8	6	6	5	7	7
	Example	NHS diagnostic bioinformatician [1]	Academic bioinformatician, industry bioinformatician	Clinical geneticist or pathologist, haematologist, microbiologist with leadership responsibility in clinical lab	Cardiologist, neurologist, oncologist, paediatrician	General Practitioner	NHS diagnostic clinical scientist, microbiologist, statistical/analytical epidemiologistt	Genetic technologist, Immunologist, epidemiologist	Genetic counsellor; Preimplantation genetic diagnosis nurse; clinical nurse specialist in surgery or oncology; Genetic Diabetes Nurse	Non-specialist nurse; physiotherapist	Systems administrator	Curator, data scientist
Competency	Write computer programmes and algorithms that can analyse data	Specialist knowledge	Specialist knowledge	No knowledge required	No knowledge required	No knowledge required	Awareness	No knowledge required	No knowledge required	No knowledge required	No knowledge required	Specialist knowledge
	Analyse genomics data using pre-existing software, including linking genotype to phenotype/microbial strain comparisons	Specialist knowledge	Specialist knowledge	Specialist knowledge[2]	Awareness	No knowledge required	Specialist knowledge	Awareness	No knowledge required[3]	No knowledge required	Awareness	Working knowledge
	Employ good software development practice	Working knowledge	Specialist knowledge	No knowledge required	No knowledge required	No knowledge required	No knowledge required	No knowledge required	No knowledge required	No knowledge required	Working knowledge	Specialist knowledge
	Apply computer science theory to computer system design	Working knowledge	Working knowledge	No knowledge required	No knowledge required	No knowledge required	No knowledge required	No knowledge required	No knowledge required	No knowledge required	Specialist knowledge	Working knowledge
	Manage and organise genomics data and results	Specialist knowledge	Specialist knowledge	Awareness[2]	No knowledge required	No knowledge required	Working knowledge	Awareness	Awareness[3]	No knowledge required	Awareness	Specialist knowledge
	Apply statistical research methods to genomics, medical, and population genetics	Working knowledge	Specialist knowledge	Working knowledge	No knowledge required	No knowledge required	Awareness[4]	Awareness	No knowledge required	No knowledge required	Awareness	Awareness
	Use health informatics systems and understand their relevance to clinical genomics	Working knowledge	Awareness	Specialist knowledge	Awareness	Awareness	Awareness	Awareness	Working knowledge	No knowledge required	Working knowledge	Specialist knowledge
	Principles of genetics, genomics and genome-sequencing technology	Specialist knowledge	Specialist knowledge	Specialist knowledge	Awareness	Awareness	Specialist knowledge	Working knowledge	Specialist knowledge	Awareness	No knowledge required	Awareness
	Principles of genetic disease	Working knowledge	Working knowledge	Specialist knowledge	Working knowledge	Working knowledge	Specialist knowledge	Working knowledge	Specialist knowledge	Awareness	No knowledge required	No knowledge required
	Principles of systems biology	Working knowledge	Working knowledge	Awareness	No knowledge required	Awareness	Awareness	Awareness	No knowledge required	Awareness	No knowledge required	No knowledge required
	Principles of next-generation sequencing	Specialist knowledge	Specialist knowledge	Awareness	Awareness	Awareness	Specialist knowledge	Awareness[6]	Awareness	No knowledge required	No knowledge required	Awareness
	Ethical, legal and social implications of clinical use of genomic data (including issues surrounding identification of patients, clinical benefits and risks, patient consent, incidental findings and ethical implications of unexpected clinically actionable findings	Working knowledge	Working knowledge	Specialist knowledge	Working knowledge	Working knowledge	Specialist knowledge	Awareness	Specialist knowledge	Awareness	Awareness	Awareness
	Interpret genetic variation in a clinical context, including understanding limitations of analysis, assessing quality and evidence for clinical interpretation	Specialist knowledge	Working knowledge	Specialist knowledge	Working knowledge	Awareness	Specialist knowledge	Awareness[5]	Specialist knowledge	No knowledge required	No knowledge required	No knowledge required
	The role of various types of healthcare professional in genomic medicine	Working knowledge	Awareness	Specialist knowledge	Working knowledge	Working knowledge	Working knowledge	Awareness	Specialist knowledge	Awareness	Working knowledge	Awareness
	The scientific discovery process and of the role of bioinformatics in it	Specialist knowledge	Specialist knowledge	Working knowledge	Awareness	No knowledge required	Working knowledge	Awareness	Awareness	No knowledge required	No knowledge required	Awareness
	The risks (and benefits) to patients and their families arising from the prediction of causal variants	Specialist knowledge	Awareness	Specialist knowledge	Working knowledge	Working knowledge	Specialist knowledge	Awareness	Specialist knowledge	Awareness	No knowledge required	Awareness
	Integrate and jointly analyse genomic and other data						[7]	[7]				

References

Welch L. et al. (2014) Bioinformatics Curriculum Guidelines: Toward a Definition of Core Competencies. PLoS Comput Biol 10(3): e1003496. doi: 10.1371/journal.pcbi.1003496

Raza, S. (2014). Defining the role of a bioinformatician. Big data and healthcare delivery (http://www.phgfoundation.org/briefing_notes/314/)

Mapping to existing training

Key:

No training available but no requirement identified	No training provided by the courses that we mapped, but training available outside the NHS	No training course identified	Training available through other HEE routes

Notes
1. STP genetics curriculum review is in progress
2. Could be included in Clinical Bioinformatics HSST

	Level		
Competency	Awareness	Working knowledge	Specialist knowledge
Write computer programmes and algorithms that can analyse data	Required by clinical genetic scientists[1]	MSc Clinical Bioinformatics	Required by clinical bioinformaticians[2], other bioinformaticians; data specialists
Analyse genomics data using pre-existing software, including linking genotype to phenotype/microbial strain comparisons	MSc Genomic Medicine	MSc Genomic Medicine, MSc Clinical Bioinformatics	MSc Clinical Bioinformatics
Employ good software development practice (software carpentry)	HEE Intro to Bioinformatics	Required by clinical bioinformaticians, IT specialists but available as part of specialist training or as standalone software carpentry workshops	MSc Clinical Bioinformatics
Apply computer science theory to computer system design	No training available but no requirement identified	MSc Clinical Bioinformatics	Required by IT specialists but may be provided through higher education in computer science
Manage and organise genomics data and results	HEE Intro to Bioinformatics, HEE intro to consent and ethics	MSc Clinical Bioinformatics	MSc Clinical Bioinformatics
Apply statistical research methods to genomics, medical, and population genetics	MSc Genomic Medicine	MSc Clinical Bioinformatics	Required by statistical/analytical epidemiologists
Use health informatics systems and understand their relevance to clinical genomics	HEE Intro to Bioinformatics, MSc Genomic Medicine	MSc Genomic Medicine	Required by specialist clinicians with genetics/genomics expertise; data specialists
Principles of genetics, genomics and genome-sequencing technology	HEE Intro to Bioinformatics, HEE intro to genomics, HEE intro to consent and ethics, MSc Genomic Medicine	MSc Clinical Bioinformatics, MSc Genomic Medicine	MSc Clinical Bioinformatics
Principles of genetic disease	HEE Intro to Bioinformatics, HEE intro to genomics, MSc Genomic Medicine	MSc Clinical Bioinformatics, MSc Genomic Medicine	STP Genetics, CCST Clinical Genetics, MSc Genetic Counselling
Principles of systems biology	HEE Intro to Bioinformatics	MSc Clinical Bioinformatics	No training available but no requirement identified
Principles of next-generation sequencing	HEE Intro to Bioinformatics, MSc Genomic Medicine	No training available but no requirement identified	MSc Clinical Bioinformatics
Ethical, legal and social implications of clinical use of genomic data (including issues surrounding identification of patients, clinical benefits and risks, patient consent, incidental findings and ethical implications of unexpected clinically actionable findings	HEE Intro to Bioinformatics, MSc Genomic Medicine	HEE intro to consent and ethics, MSc Clinical Bioinformatics, MSc Genomic Medicine	STP Genetics, CCST Clinical Genetics, MSc Genetic Counselling
Interpret genetic variation in a clinical context, including understanding limitations of analysis, assessing quality and evidence for clinical interpretation	HEE Intro to Bioinformatics; HEE Intro to genomics, HEE intro to consent and ethics, MSc Genomic Medicine	MSc Clinical Bioinformatics, MSc Genomic Medicine	MSc Clinical Bioinformatics
The role of various types of healthcare professional in genomic medicine	HEE Intro to Bioinformatics, MSc Genomic Medicine	Required by clinical bioinformaticians; specialist clinicians; other clinicians; clinical genetic scientists; IT specialists	Required by specialist clinicians with genetics/genomics expertise; specialist nurses/counsellors
The scientific discovery process and of the role of bioinformatics in it	HEE Intro to Bioinformatics, MSc Genomic Medicine, MSc Clinical Bioinformatics	MSc Clinical Bioinformatics, MSc Genomic Medicine	Required by clinical bioinformaticians; other bioinformaticians
The risks (and benefits) to patients and their families arising from the prediction of causal variants	HEE Intro to genomics, MSc Genomic Medicine	HEE intro to consent and ethics, MSc Genomic Medicine, MSc Clinical Bioiformatics	MSc Clinical Bioinformatics
Integrate and jointly analyse genomic and other data	MSc Genomic Medicine	Required by epidemiologists	Required by statistical/analytical epidemiologists

As a proof of principle we selected three curricula that have specifically been developed with the aim of realising the potential of genomic medicine in the NHS and mapped them to the profile to identify potential gaps in training provision:

- HEE's Genomics Education Programme introductory courses and a new course on ethics and consent, currently in development
- Modernising Scientific Careers (MSC) Scientist Training Programme MSc in Clinical Science – Clinical Bioinformatics
- The new HEE Genomics Education Programme MSc in Genomic Medicine

Conclusions

Many of the specialist training requirements to support the 100,000 genomes project will be met through the HEE commissioned MSc in Genomic Medicine (and CPD accredited modules therein) or through the MSC clinical scientist training scheme in bioinformatics. There are also a number of specialist training courses available outside HEE. These can contribute to the development of bioinformatics skills but have not been specifically designed to meet the requirements for clinical staff and scientists delivering the 100,000 genomes project or for genomic medicine more generally. The mapping could be used to identify other appropriate course providers and/or develop new training.

The HEE Clinical Bioinformatics Task and Finish Group

Clinical Bioinformatics Task & Finish Group
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