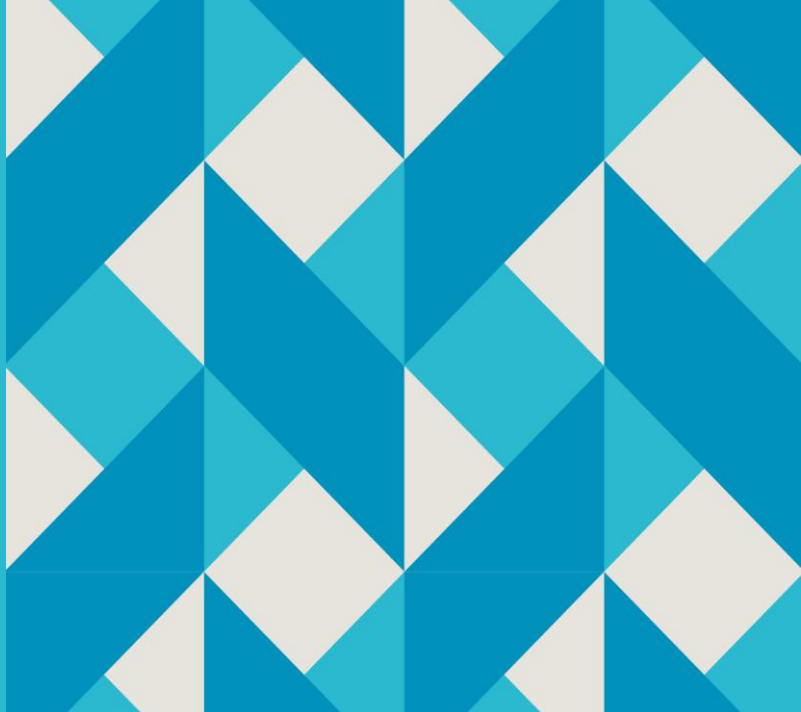




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Genomics

Advanced **genomics** knowledge guide for pharmacists

Co-author:
Hayley Wickens



Co-author:
Dharmisha Chauhan



Endorsed by:



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Background

Genomic technology has developed at pace since the early 2000's and is now in regular clinical use in both diagnostics and treatment in the UK. Pharmacogenomics — the use of genomics to predict how patients will metabolise and respond to medicines, including adverse drug reactions, — is in its relative infancy in clinical practice in the UK. However, pharmacogenetic testing is anticipated to become more widespread within the coming years, and the use of genomics to personalise or individualise medicine choices in cancer and rare disease is already commonplace.¹

The pharmacy workforce has been identified as key to the implementation of genomic medicine,² and genomics is now incorporated into the GPhC standards for initial education and training of pharmacists.³ Whilst an indicative curriculum for undergraduate and trainee pharmacists has been published,⁴ to date there has been no description of genomic knowledge requirements of post-registration pharmacists beyond this.

We have compiled a separate document describing knowledge requirements for all pharmacists (**UKCPA Genomics knowledge guide for pharmacists**); this Advanced genomics knowledge guide describes the 'next steps' for pharmacists wishing to develop their knowledge beyond the basics, and also some of the attributes and knowledge required by specialist genomics pharmacists.

Aim of this guidance

The structured guidance given in this document will help guide individual development and allow education providers to identify gaps in education and training resources.

This guidance has been developed by the authors and the UKCPA Genomics Committee, taking into account the North Thames Genomic Advisors Competency Framework, and with reference to genomics competencies from other professions⁶⁻⁹ and published studies.^{10,11}

How to use this guidance

This document is intended to be an outline to guide practice rather than a prescriptive list, and where used for educational purposes, should be read in conjunction with the appropriate Royal Pharmaceutical Society (RPS) curricula,¹²⁻¹⁴ and, where appropriate, the RPS Prescribing Competency Framework.¹⁵ Further work to produce a syllabus and knowledge guide for pharmacy technicians working in genomics will be developed as roles across both professions develop.

This document supports the delivery of the Pharmacy Genomics Workforce Strategic Framework² Aim 3 (identify pharmacy genomics workforce needs) and Aim 4 (educate and develop the pharmacy workforce).

The sections within the knowledge guide are:

1. Fundamentals of genomics (knowledge)
2. Applications of genomic medicine (knowledge)
3. Genomics skills and behaviours for pharmacists (skills and behaviours)

Advanced genomics knowledge guide for pharmacists

1. Fundamentals of genomics

		Entry level (generalist)	Advanced (generalist, or specialist in field other than genomics)	Genomics specialist
1.1 Basic science: DNA, RNA and protein	a. Fundamental concepts of DNA, RNA and protein, and the basics of transcription and translation	✓	✓	✓
	b. The organisation of human genome into 23 pairs of chromosomes and approximately 20,000 genes	✓	✓	✓
	c. RNA splicing and epigenetics		✓	✓
1.2 Contribution of genetics to disease states	a. Single nucleotide variation leading to impact on sequence (missense, stop gain, frameshift)	✓	✓	✓
	b. Loss of function and gain of function variants and impact on disease	✓	✓	✓
	c. Types of copy number variation and impact on disease (e.g. trisomy, translocations, microdeletions)	✓	✓	✓
	d. Constitutional and somatic variation and the roles of these in development of disease including cancer	✓	✓	✓

e.	Homozygosity and heterozygosity and impact on disease states	✓	✓	✓
f.	Inheritance patterns of single gene disorders (e.g. autosomal dominant/recessive, increased risk of recessive disorders in consanguinity)	✓	✓	✓
g.	Mitochondrial inheritance and relevance for pharmacogenomics	✓	✓	✓
h.	Concepts of penetrance and variable expressivity		✓	✓
i.	Genomic mosaicism and implications for disease			✓

1.3 Normal genomic variation

a.	The extent of normal genomic variation, including that the majority of variation is non-pathogenic.	✓	✓	✓
b.	The existence of the Human Reference Genome and its limitations	✓	✓	✓
c.	The influence of ancestry on normal genomic variation	✓	✓	✓
d.	The role of normal genomic variation in drug response in terms of drug targets, drug metabolism and risk of adverse effects (see also 2.1)	✓	✓	✓
e.	The use of databases of variation (e.g. GnomAD) and their limitations			✓

1.4 Genetic contribution to common complex disease

- a. Genomic factors that influence development of common complex disease, e.g. cardiovascular disease
- b. Interactions of genetics and environment in disease
- c. Awareness of benefits and limitations of polygenic risk scores

✓	✓	✓
✓	✓	✓
✓	✓	✓

1.5 Genomic technologies

- a. Definitions of single gene/SNV testing, panel testing, clinical/whole exome sequencing, whole genome sequencing
- b. Advantages and disadvantages of each testing approach
- c. Point of care vs. laboratory based testing and associated considerations
- d. Comparison of long and short read technologies

✓	✓	✓
✓	✓	✓
✓	✓	✓
	✓	✓

2. Applications of genomic medicine

		Entry level (generalist)	Advanced (generalist, or specialist in field other than genomics)	Genomics specialist
2.1 Pharmaco- genomics: basic principles	a. Concept of pharmacogenomics in drug metabolism (ADME)	✓	✓	✓
	b. Role of pharmacogenomic variation in predicting adverse drug reactions	✓	✓	✓
	c. Role of pharmacogenomic variation in targeted treatment/ precision medicine (e.g. drug-gene matches)	✓	✓	✓
	d. Locating information on pharmacogenomics in manufacturers Summary of Product Characteristics	✓	✓	✓
	e. Awareness of key pharmacogenomics reference sources (e.g. PharmGKB, DPWG guidelines, CPIC)	✓	✓	✓
	f. Concept of pharmacogenomics as part of holistic clinical pharmacy review (e.g. renal function, adherence etc)	✓	✓	✓
	g. Use of key specialist reference sources as above		✓	✓
	h. Concept of phenoconversion		✓	✓

**2.2 Genomics
in medicines
safety**

- a. MHRA Drug Safety Alerts concerning genetic variation leading to adverse outcomes
- b. Regulatory aspects of medicines with linked genomic tests for dosing or indication

✓	✓	✓
		✓

**2.3 Genomics
in the NHS:
systems and
practice**

- a. Awareness of the NHS genomic medicine services and key genomics policies
- b. Awareness of national genomic test directories / national commissioning of genetic tests
- c. Awareness of how a genomic/pharmacogenomic test would be requested in the individual's practice setting

✓	✓	✓
✓	✓	✓
✓	✓	✓

3. Genomics skills and behaviours for pharmacists

		Entry level (generalist)	Advanced (generalist, or specialist in field other than genomics)	Genomics specialist
3.1 Assess and advise on when pharmacogenetic testing is indicated and choose appropriate test	a. Advise prescribers on choice of appropriate NHS-commissioned genomic tests related to medicines	✓	✓	✓
	b. Awareness of strengths and limitations Direct to Consumer pharmacogenomic tests	✓	✓	✓
	c. Awareness of advantages and limitations of different testing methodologies including turnaround time (e.g. POCT vs local vs central laboratory)	✓	✓	✓
3.2 Communicate effectively with patients regarding genomic/pharmacogenomic testing	a. Provide information to patients around options for pharmacogenomic testing, discussing the risks and benefits in a non-directive way, respecting patient autonomy	✓	✓	✓
	b. Describe potential impact of test result on other family members	✓	✓	✓

c.	Awareness of the Code of Testing and Insurance agreed between HM Government and Association of British Insurers on the role of genetic testing in insurance	✓	✓	✓
d.	Understand why consent for data to be used in research may be important and how to discuss this with patients	✓	✓	✓
e.	Understand principles of confidentiality concerning genomic data ¹⁶	✓	✓	✓

3.3
Understand
how to
interpret and
action a
pharmaco-
genetic report

a.	Understand the basic format of a pharmacogenetic report e.g. genes and variants tested, star alleles/diplotype, phenotype	✓	✓	✓
b.	Understand that previous genomic test results may have an updated interpretation due to reclassification of variants	✓	✓	✓
c.	Interpret test report in context of phenotype and holistic clinical picture, including other prescriptions, and make recommendation for prescribing	✓	✓	✓
d.	Apply an evidence-based approach to pharmacogenomic results, utilising appropriate reference sources (see also 2.1d)	✓	✓	✓

- e. Make appropriate records in patient notes with recommendations, considering visibility to other prescribers/professionals within context of e.g. electronic systems, and following local policy
- f. Awareness of national recommendations regarding record keeping for pharmacogenomic results

✓	✓	✓
	✓	✓

3.4 Communicate a genomic result

- a. Communicate a genomic test result to patients/carer in a manner that they understand
- b. Communicate any implications for family members e.g. further testing
- c. Frame context: where there is uncertainty around result or risk, or incidental findings, explain these to the patient

✓	✓	✓
✓	✓	✓
		✓

3.5 Recognising limitations

- a. Recognise personal limitations and refers appropriately to specialist genomics pharmacist
- b. Recognise where a patient may require referral to specialist genomic counselling services

✓	✓	✓
✓	✓	✓

**3.6
Leadership
and
education**

a. Represents the speciality on regional committees/boards to influence genomic strategies and direction		✓	✓
b. Represents the speciality on national committees/boards to influence genomic strategies and direction			✓
c. Provides speciality-specific leadership at a local/regional level		✓	✓
d. Provides speciality-specific leadership at a regional/national level			✓
e. Leads on local initiatives to foster learning so that colleagues can use genomic data safely and effectively for patient benefit		✓	✓
f. Leads on regional/national initiatives to foster learning so that colleagues can use genomic data safely and effectively for patient benefit			✓
g. Establishes links with national specialty specific stakeholders, implements specialty service specification (regarding genomics), and assists regional advisors to navigate the NHS GMS infrastructure.			✓

h. Mentors regional genomic advisors; interacts with Genomic Education Programme (GEP), specialist genomic services, and Academy of Medical Royal Colleges (AoMRC). Represents genomics for specialty, including specialty professional bodies.			✓
i. Represents the specialty on national committees/boards to influence genomic strategies and directions			✓
j. Shapes the NHS National Genomic Test Directory through annual submission of specialty recommendations.			✓

Acknowledgments

UKCPA Genomics committee members

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Lucy Galloway

Aris Saoulidis

Farah Longerstaey

Sadaf Qureshi

Paul Selby

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Nadia Bashir

Raliat Onatade

Kate Tatton-Brown

Endorsing bodies

Yogita Dawda

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The Genomics in Pharmacy page of the National Genomics Education Programme website also provides further reading, case studies, and links to educational resources and key references in genomics:

<https://www.genomicseducation.hee.nhs.uk/genomics-in-healthcare/genomics-in-pharmacy/>