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Genomics

Genomics knowledge guide for pharmacists

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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. As genomic medicine services develop, we anticipate a need to describe knowledge requirements for all pharmacists, where their primary speciality is not genomics.

Aim of this guidance

The structured guidance given here will help inform individual development and allow education providers to identify gaps in education and training resources. This document has been produced for postgraduate pharmacists in general clinical practice. Specialist genomics pharmacists and pharmacists who are specialists in genomics-related fields, e.g. cancer, are encouraged to refer to the **Advanced genomics knowledge guide for pharmacists**.

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 professions[6-9] 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 Framework[2] 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)

Genomics knowledge guide for pharmacists

1. Fundamentals of genomics

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
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 consanguinuity) g. Mitochondrial inheritance and relevance for pharmacogenomics
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)

1.4 Genetic contribution to common complex disease	a. Genomic factors that influence the 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

2. Applications of genomic medicine

2.1 Pharmacogenomics: 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)
2.2 Genomics in medicines safety	a. MHRA Drug Safety Alerts concerning genetic variation leading to adverse outcomes

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

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 of 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 the 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 pharmacogenetic 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 the context of phenotype and holistic clinical picture, including other prescriptions, and make recommendations for prescribing d. Apply an evidence-based approach to pharmacogenomic results, using 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, for example, electronic systems, and following local policy
3.4 Communicate a genomic result	a. Communicate a genomic test result to a patient/carer in a manner that they understand b. Communicate any implications for family members, e.g. further testing
3.5 Recognising limitations	a. Recognise personal limitations and refer appropriately to specialist genomics pharmacist b. Recognise where a patient may require referral to specialist genomic counselling services.

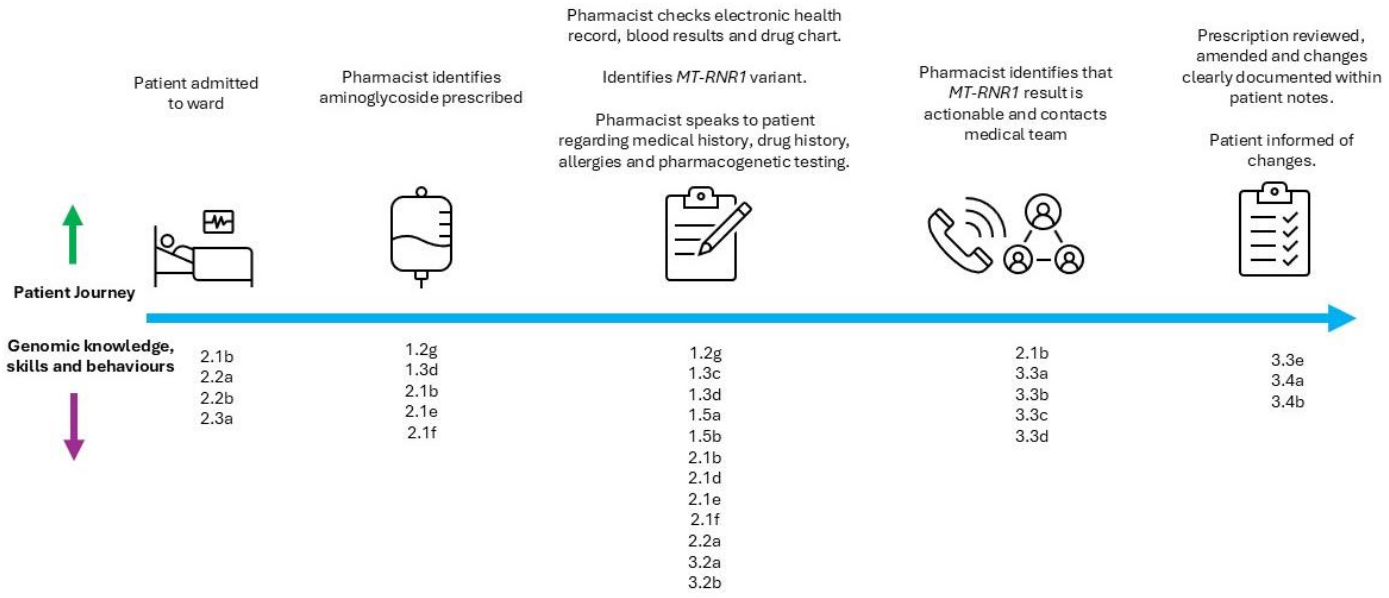
Examples of applied genomics in medicines optimisation that may be useful for educators

- Role of genomics in diagnosis and treatment of diabetes
- Role of genomics in familial hypercholesterolaemia, including diagnostic criteria and case identification
- Role of genomics in diagnosis and treatment of cystic fibrosis
- Role of genomics in diagnosis and treatment of mental health disorders
- Role of genomics in antimicrobial stewardship, including disease outbreaks, rapid diagnosis and antimicrobial susceptibility testing.

Case studies

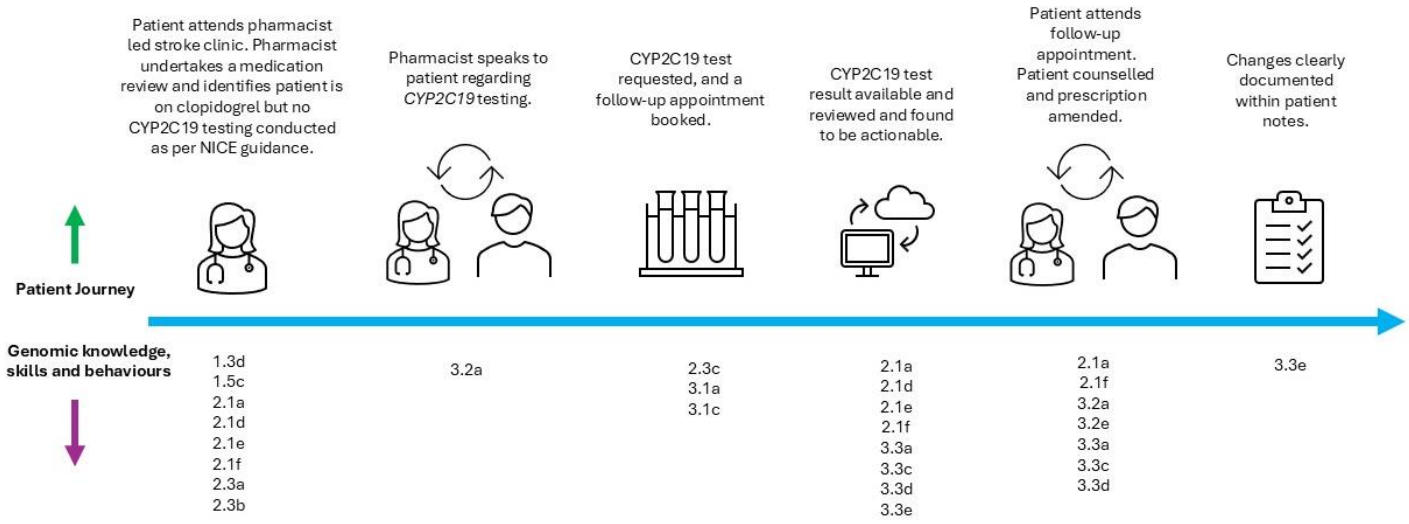
Case study 1: Inpatient ward *MT-RNR1* pharmacogenetic testing and aminoglycoside prescription

A 27-year-old female with Caucasian and Chinese ancestry is admitted to a respiratory ward for intravenous treatment for bronchiectasis. Based on hospital antimicrobial guidelines, an aminoglycoside in combination with another antibiotic has been prescribed after a ward round. As the ward pharmacist you are reviewing the patient's drug chart to start your clinical review.



Case study 2: Primary care review within a pharmacy-led stroke clinic within a GP surgery

A 65-year-old male with South Asian ancestry has been referred to your stroke clinic for a clinical review two months after discharge from a stroke ward. You are reviewing his medications and notice that a *CYP2C19* pharmacogenetic test has not been requested as per the NICE guidelines. You know that the test is commissioned and can be requested from your regional genomic laboratory hub.



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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/