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# Working Group on Genomics in Mainstream Medicine – a progress report for the Royal College of Physicians and Joint Specialty Committees

The Working Group on Genomics in Mainstream Medicine aims to raise awareness and prepare clinicians across a breadth of specialities to integrate genomics into their clinical practice. The group includes clinical champions from varied specialities who are taking the lead through formal and informal structures.

## Clinical champions

Anne Barton (Rheumatology)  
Maria Bitner-Glindzicz (Audio-vestibular)  
Graeme Black (Ophthalmology)  
Krish Chatterjee (Endocrinology)  
Ellen Copson (Medical Oncology)  
Finbarr Cotter (Haematology)  
Neil Gittoes (Endocrinology)  
Pauline Ho (Rheumatology)  
Maria Kinali (Paediatrics)  
Irene Leigh (Dermatology)  
David Lomas (Respiratory Medicine)  
Hugh Markus (Stroke Medicine)  
Huw Morris (Neurology)  
Munir Pirmohamed (Clinical Pharmacology and Therapeutics)  
Imran Rafi (General Practice)  
Jack Satsangi (Gastroenterology)  
John Sayer (Renal Medicine)  
Claire Shovlin (Respiratory Medicine)  
Estée Török (Infectious Diseases)  
Thomas Wagner (Nuclear Medicine)  
James Ware (Cardiology)  
Hugh Watkins (Cardiology)

The field of genomics has grown hugely since the completion of the human genome project in 2003. Technological developments are enabling researchers to identify genomic factors contributing to disease in all medical specialties. In many specialties this massive research endeavour is starting to change practice, enabling more precise diagnoses which can inform prognosis and therapy and using pharmacogenetic testing to avoid serious side effects in individuals.

This has the potential to radically alter the way in which medicine is practised with healthcare delivery moving from a 'one size fits all' approach to providing care that is tailored to the individual.

## The role of education

If these benefits are to be realised across medicine, there is a need to shift genomics from being the sole preserve of geneticists to adoption by all specialties. A transformation in clinical practice will be required. The aim of the Working Group on Genomics in Mainstream Medicine is to raise awareness and prepare clinicians across a breadth of specialties to integrate genomics into their clinical practice. Examples for different specialties are given in Box 1 (overpage).

## The Genomic in Mainstream Medicine Working Group

In 2012 a report *Genomics in Medicine* by the PHG Foundation, Joint Committee on Genomic Medicine (JCMG), the UK Genetic Testing Network and the NHS National Genetics Education and Development Centre highlighted the need to develop capacity in clinical specialties to undertake genomic medicine. Clinical specialties outside genetics would take the lead for their patients with inherited disease developing special relationships with genetics services to provide support. However, it was clear that clinical specialists were largely not ready to take on these extra complexities of care and a major programme of education would be required to develop those in training and already in practice.

The Genomics in Mainstream Medicine Working Group was set up in 2013 as a direct result of a meeting held at the Royal College of Physicians (RCP) to consider these findings. Under the auspices of the RCP the working group includes clinical champions from a range of specialties who are taking the lead through formal and informal structures. The work was led by the PHG Foundation and was a partnership with the Joint Committee on Genomics in Medicine and Health Education England Genomics Education Programme.

### Box 1 Examples of how genomics is impacting on clinical practice

#### Oncology

Molecular testing in diagnosis of cancer and guiding treatment decisions

#### Nephrology

Predicting drug response in patients with atypical Haemolytic Uraemic Syndrome

#### Gastroenterology

Predicting adverse effects when patients with inflammatory bowel disease are to be offered treatment with Azathioprine

#### Cardiology

Determining the genetic cause of a cardiac arrhythmia or hypercholesterolaemia to identify at-risk relatives and offer preventive treatment

#### Dermatology

Identifying subtypes of epidermolysis bullosa to inform prognosis and management

#### Respiratory Medicine

Predicting severe adverse drug reactions to anti-tuberculous therapy or choosing optimal drug treatment in cystic fibrosis according to the specific inherited molecular defect

#### Rheumatology

Use of molecular testing for HLA-B27 antigen to assist diagnosis and prognosis in juvenile idiopathic arthritis

#### Audiovestibular medicine

Using molecular testing to predict individuals and family members who may be extremely sensitive to the ototoxic effects of aminoglycoside antibiotics



## Genomics in mainstream medicine

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### New educational resources

A suite of resources has been developed to raise awareness about genomics across clinical specialities. Each was written by the relevant clinical champion, working with co-authors from the specialty. Organised around a generic template, resources cover genomics in diagnostics, rare disease, cancer, common complex disease and pharmacogenetics, giving speciality specific examples for:

- Audiovestibular medicine
- Dermatology
- Endocrinology
- Gastroenterology
- Nephrology
- Primary care
- Respiratory medicine
- Rheumatology
- Oncology

Ethical, legal and social aspects that will be relevant for all physicians are also included and the resources end with a list of recommended reading and websites / organisations for access to further educational materials.

Further resources in development include those on pathology, cardiology, infectious disease, neurology, paediatrics and ophthalmology.

### Where are the resources available from?

The introductory specialty specific materials can be found on:

The PHG Foundation website  
[www.phgfoundation.org/clinical\\_champions](http://www.phgfoundation.org/clinical_champions)

Health Education England Genomics Education Programme website  
[www.genomicseducation.hee.nhs.uk/resources/genomics-in-mainstream-medicine](http://www.genomicseducation.hee.nhs.uk/resources/genomics-in-mainstream-medicine)

### Other activities

The Working Group has undertaken scoping work on the landscape of genomics education for clinicians in the UK (available as a publication *Genomic Education for Medical Professionals – The current UK landscape*. Slade I, Subramanian D and Burton H. *Clinical Medicine* [in press] and an editorial accepted by *Postgraduate Medical Journal*).

The Working Group has invited presentations on other genomics education initiatives including the Massive Open Online Course (MOOC) in genomics (St George's Hospital), the HEE Masters in Genomic Medicine and the development of genetics within the specialist training curricula (work of Specialist Advisory Committees).



### Steering group

Hilary Burton (Chair and PHG Foundation)

Corinna Alberg (PHG Foundation)

Michelle Bishop (Health Education England)

Trevor Cole (Clinical Genetics)

Ian Cree (Royal College of Pathologists)

Val Davison (Health Education England)

Peter Farndon (NGGEC)

Maxine Foster (Health Education England)

Margaret Johnson (Royal College of Physicians)

Nick Lench (Joint Committee on Genomics in Medicine)

Ruth Newbury-Ecob (Clinical Genetics Society)

Ingrid Slade (Centre for Personalised Medicine)

Sarah Smithson (Specialist Advisory Committee Clinical Genetics)

Kieran Walsh (BMJ learning)

John Wass (Royal College of Physicians)

Ann-Marie Wright (Health Education England)

Mitali Wroczynski (BMJ learning)

### Engaging with other organisations

In addition to the founding members, organisations represented on the Group now include Health Education England, BMJ Learning, Specialist Advisory Committee on Medical Genetics, Royal College of Pathologists, Royal College of Paediatrics and Child Health, Royal College of General Practitioners and Clinical Genetics Society.

### Next steps

In the coming year the Working Group plans to:

- Hold a professional engagement event in conjunction with Genomics England
- Disseminate the resources already complete
- Work with HEE Genomics Education Programme to develop a 'web portal' for the introductory resources
- Continue to work through RCP structures to raise awareness and get genomics into the various curricula
- Encourage clinical champions to raise awareness in different specialties through specialty organisations
- Work with a wider set of clinical champions including those from other Royal Colleges (*e.g.* RCPCH)

### What other educational resources are available?

Information on other resources for genomics education of health professionals can be found on the website of Health Education England's Genomics Education Programme [www.genomicseducation.hee.nhs.uk](http://www.genomicseducation.hee.nhs.uk)

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**Health Education England**



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of Physicians**

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