RCGP Curriculum

Supercondensed Curriculum Guide

GENOMIC MEDICINE

Role of the GP

- Take and consider family histories in order to identify families with, or at risk of, genetic conditions and familial clusters of common conditions such as cancer, cardiovascular disease and diabetes
- Identify patients and families who would benefit from being referred to appropriate specialist services
- Manage the day-to-day care of patients with genetic conditions, even if the patient is under specialist care
- Coordinate care across services, including transitions from paediatric to adult services
- Communicate information about genetics and genomics
- Understand how genomic information is used within the context of routine clinical practice
- Be aware of developments in genomic medicine (e.g. direct-to-consumer genomic testing) and their implications for general practice.

Knowledge and Skills Self-Assessment Guide

Symptoms, Signs and Modes of Presentation

- Clinical suggestion of inherited disease (e.g. multiple family members affected at a younger age)
- Genetic ‘red flags’ (e.g. recurrent miscarriage)
- Predisposition to common diseases (e.g. coronary artery disease or cancer)
- Symptoms and signs of specific conditions (see ‘Common and Important Conditions’)
- Symptom complexes and multisystem involvement
- Variability of symptoms and signs between family members for some genetic conditions due to variation in penetrance and expression (e.g. neurofibromatosis type 1).

Knowledge and Skills Self-Assessment Guide

Common and Important Conditions

- Autosomal dominant conditions (e.g., familial hypercholesterolaemia, polycystic kidney disease, Huntington’s Disease, thrombophilias, Lynch syndrome, BRCA1 pathogenic variant in breast cancer)
- Chromosomal disorders (e.g., Down syndrome, trisomy 18, Turner syndrome, Klinefelter syndrome)
- Common conditions with a multi-factorial inheritance pattern (e.g., ischaemic heart disease, hypertension, diabetes, cancer, obesity)
- Autosomal recessive conditions (e.g., cystic fibrosis, hereditary haemochromatosis, haemoglobinopathies)
- X-linked disorders (e.g., Fragile X Syndrome, Duchenne and Becker Muscular Dystrophy, haemophilia).

GENOMIC MEDICINE

Knowledge and Skills Self-Assessment Guide

Assessing Genetic Risk

- How to take a family history (relevant questions, interpretation, how to draw a pedigree)
- Basic inheritance patterns (autosomal dominant and recessive, X-linked, mitochondrial, multifactorial)
- Principles of assessing genetic risk including polygenic risk scoring
- Other factors contributing to genetic risk (e.g. ethnicity, consanguinity).

Knowledge and Skills Self-Assessment Guide

Investigations

- Genetic and genomic tests (diagnostic, predictive, carrier testing) and their limitations
- Diagnostic tests in primary care (e.g. cholesterol, ultrasound for polycystic kidney disease, testing for hereditary haemochromatosis)
- Carrier testing for families with autosomal recessive conditions such as sickle cell, thalassaemia or cystic fibrosis
- Antenatal and new-born screening programmes
- Genomic sequencing of pathogens.

Knowledge and Skills Self-Assessment Guide

Other

- Spectrum of risk-reducing measures, from lifestyle modification to targeted treatments for certain conditions (eg mastectomy and/or oophorectomy for BRCA1/2 mutation carriers, colectomy for adenomatous polyposis coli (APC) mutation carriers, aspirin for Lynch syndrome, statins for familial hypercholesterolaemia, venesection for haemochromatosis, losartan for patients with Marfan syndrome) and targeted gene therapy.

GENOMIC MEDICINE

How this might be tested in MRCGP

AKT

- Knowledge of antenatal and newborn screening programmes
- Consent, capacity, and confidentiality of genetic testing
- Interpretation of a pedigree analysis chart.

SCA

- Woman with one affected sibling requests genetic screening for breast cancer
- Woman attends for pre-conceptual advice because her nephew has Duchenne muscular dystrophy
- Neurology letter (provided): 'symptoms suggest cerebellar ataxia, with autosomal recessive inheritance'. Patient attends to discuss the implications of her own probable diagnosis for her children.

WPBA

- Audio Consultation Observation Tool (Audio COT) with a parent discussing the chances of passing his thalassemia-associated variant (trait) to his children
- Log entry about communicating with an adult patient who has Down syndrome
- Log entry about a mother who is finding it hard to cope with her child having cystic fibrosis.

LEARNING OPPORTUNITIES (Examples)

Core Content

- **Communication and Consultation**
  - Understanding basic genomic science & nomenclature
  - Taking a family history
  - Risk-benefit conversations
  - Risk-reducing measures
  - Online risk assessment tools
  - Non-directive & non-judgemental consultation skills
- **Prescribing**
  - Pharmacogenetics
- **Teamworking**
  - Surveillance, follow-up, coordination of care, referral pathways
- **Co-morbidity**
  - Multifactorial inheritance in common complex diseases
- **Medico-legal/ Ethics**
  - Confidentiality & use of genetic information
  - Right 'not to know'
  - Genetic testing and screening
  - Implications for family members
  - Reproductive genetics.

Primary Care

- Daily practice and out-of-hours
- Specialist clinics (e.g., midwife, GP with extended role clinics such as cardiology, epilepsy).

Community

- Community paediatrics
- Health visitors
- Learning disability teams
- Regional specialist genetics services.

Acute Care

- Following the patient journey (e.g., via ward rounds, MDT meetings, discharge planning)
- Seeing emergency presentations and referrals from primary care
- Managing acute complications related to genetic conditions (e.g., sickle cell crisis).

Multidisciplinary Team

- Pharmacist (e.g., identifying familial hypercholesterolaemia as part of lipid optimisation, pharmacogenomics in hospital medicine)
- Specialist nurse (e.g., family history breast care nurse).

Other Specialties

- Clinical genetics – consultant geneticists, genetic counsellors, genomic associates and family history administrators
- Obstetrics
- Oncology
- Paediatrics
- Surgery
- Specialties related to specific clinical conditions with genetic associations (e.g., cystic fibrosis, breast cancer).