The RCGP Curriculum

The Curriculum Topic Guides

Super-Condensed Topic Guides 2021

Genomic Medicine

The Role of the GP and emerging issues in primary care

- Take and consider family histories in order to identify families with, or at risk of, genetic disorders or 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, developmental delay in conjunction with other morbidities)
- 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).

Common and Important Conditions

- Autosomal dominant conditions (e.g. familial hypercholesterolaemia, polycystic kidney disease, Huntington's Disease, thrombophilias)
- Chromosomal disorders (e.g. Down syndrome, trisomy 18, Turner syndrome, Klinefelter syndrome)
- Common complex conditions 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).

Assessing Genetic Risk

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

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 (e.g. Down syndrome, cystic fibrosis, sickle cell and thalassaemia)
- Genomic sequencing of pathogens.

How this might be tested in MRCGP

AKT

- Interpretation of a genogram
- Recognising the presentation of common genetic conditions in primary care
- Consent, capacity and confidentiality of genetic testing.

RCA

- 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.

How to learn this topic

This section describes *examples* of opportunities for learning.
We recognise that Covid-19 restrictions have significantly affected their accessibility

Acute

- 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

Other relevant specialties:

- Clinical genetics – consultant geneticists, genetic counsellors
- Obstetrics
- Paediatrics
- Specialties related to specific clinical conditions with genetic associations e.g. cystic fibrosis, breast cancer

Primary Care

- Day to day practice
- OOH
- Community
- Specialist clinics e.g. midwife

Core Themes

- **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 –** pharmacogenomics
- **Co-morbidity –** multifactorial inheritance in common complex diseases
- **Teamworking –** surveillance, follow up, coordination of care, referral pathways
- **Ethical and medico-legal** – confidentiality & use of genetic information, right 'not to know', genetic testing & screening, implications for family members, reproductive genetics.

Community/MDT

- Community paediatrics
- Health visitors
- Learning disability teams
- Private sector (e.g. genetic testing)
- Regional specialist genetics services

Tips

- Audit
- Significant Event Analysis
- Clinical governance
- Risk Assessment
- Dr as teacher
- Leadership
- BNF
- NICE guidelines