Guidance on implementation of the 2016 amendments to 2010 Clinical Genetics curriculum

Background

The laboratory section of the Clinical Genetics curriculum has been rewritten as a necessity in a very rapidly changing discipline. The technologies that are used by genetics laboratories and therefore accessed and interpreted by clinical geneticists have changed and we need to ensure that the curriculum reflects this.

Clinical Genetics trainees are already learning these new technologies and working with the labs and research institutes that are undertaking the analyses.

The change to the curriculum will ensure trainees can record progress in the updated competencies in the ePortfolio.

In addition to the change to the laboratory section, the following additional changes have been made:

- The assessment section has been updated with current information on the specialty examination
- The Multiple Consultant Report (MCR) has been added to the assessment methods and blueprinted to the syllabus as agreed with GMC for all physicianly specialties
- Generic sections on Good Medical Practice and Equality and Diversity have been updated in line with policy and legislation.

Transition of current trainees

Year 1-3 trainees (ST3-ST5)

- Trainees will be expected to use the amended curriculum with immediate effect and will be expected to demonstrate that they have met the revised laboratory competencies.

Year 4 trainees (ST6)

- Trainees in their final year of training will not be required to record evidence or progress for the new section.

All trainees should already be meeting the MCR requirement.

Eportfolio guidance

- The 2016 amendments have been made to curriculum on the ePortfolio. Trainees should use the 'Laboratory Genetics – revised 2016' as per the transition guidance above. The previous version of this syllabus topic will be retained so any linked evidence can still be viewed.

Mapping of 2016 changes to 2010 (2013 amendments) Clinical Genetics curriculum

Section 11: Laboratory Genetics

2010 Clinical Genetics (2013 amendments)	2010 Clinical Genetics (2016 amendments)
Knowledge	**Knowledge**
Techniques for conventional chromosome analysis in different tissues	Understand techniques for conventional cytogenetic analysis in different tissues
Laboratory techniques and application of new cytogenetic tests, eg FISH/CGH	Understand the principles of FISH analysis and its applications
	Interpret clinical consequences of chromosome rearrangements
	Apply array-CGH in different clinical settings and interpretation of CNV's (including use of databases such as DECIPHER and ECARUCA)
Use of ISCN nomenclature	Use ISCN nomenclature correctly
Molecular genetic techniques in common usage- (DNA extraction, Southern Blotting, PCR, DNA sequencing	Know the molecular genetic techniques in common usage: DNA extraction, Southern Blotting, PCR, MLPA and Sanger sequencing.
Application of DNA-based testing for genome mapping, linkage and mutation detection.	Understand the principles and application of next generation sequencing (NGS) technologies including targeted panels, clinical exome sequencing, whole exome sequencing and whole genome sequencing,
Potential application of new DNA technologies	Interpret the large data set created from NGS using basic bioinformatics, filtering techniques, clinical and functional data.
	Know OMIC technologies and their current and future applications
	Be aware of the Human Genome Variation (HGVS) nomenclature for single gene variants.
Sensitivity and specificity of laboratory tests	Understand the sensitivity and specificity of laboratory tests
Investigative approach to biochemical diagnosis of inborn errors of metabolism (via experience gained at metabolic disease clinics.	Investigate inborn errors of metabolism through liaison with metabolic disease colleagues and the genetic laboratory.
The operation of local and national antenatal and newborn genetic disease screening programmes.	Be aware of the operation of local and national antenatal and newborn genetic disease screening programmes.

Skills	**Skills**
Interpretation of clinical consequences of abnormal karyotypes, enzyme deficiencies and molecular test results.	Interpret results of cytogenetic, molecular cytogenetic, molecular genetic and biochemical tests.
Liase with molecular and cytogenetic scientists in analysis of test results.	Liaise with laboratory scientists and bioinformaticians in the analysis of test results.
Provide advice to the laboratory on the wording of reports to referring clinicians.	Provide advice to genetic laboratory colleagues on the wording of reports to referring clinicians
Genetic risk calculation based on laboratory test results (eg MLINK, Bayesian analysis)	Undertake genetic risk calculation based on laboratory test results (incorporation of genetic test results into Bayesian calculations)
	Use databases including ENSEMBL, USCS and locus-specific databases for interpretation of results
Behaviours	**Behaviours**
Awareness of the importance of informed consent that arise in relation to storage of DNA samples and cell lines	Develop awareness of the importance of informed consent in relation to storage of DNA and cell lines
Willingness to liaise with colleagues to interpret laboratory results	Show willingness to liaise with laboratory colleagues to interpret laboratory results
Recognise the value of clear reports and the importance of communicating results clearly to families	Recognise the importance and impact of genetic test results for families and communicate implications of results clearly to them.
	Demonstrate awareness of the potential for incidental findings in genomic analysis and the complexity of these from the patient perspective
	Be able to take informed consent when undertaking genomic analyses
	Be able to adapt to new techniques and tests as they arise and incorporate them into clinical practice appropriately.

Generic content

Section	**2010 Clinical Genetics (2013 amendments)**	**2010 Clinical Genetics (2016 amendments)**
3.3 Good Medical Practice (and syllabus)	3.3 Good Medical Practice	3.3 Good Medical Practice This section has been updated and the content of learning has been mapped to 2013 version of GMP
5.3 Assessment methods	Examinations and Certificates • The Certificate Examination of Clinical Genetics (CE)	Examinations and Certificates • The Certificate Examination - Medical Genetics (CE)

		This section has been updated with current information about the specialty examination
5.3 Assessment methods		The Multiple Consultant Report (MCR) has been added to assessment methods with a description
5.5 ARCP Decision Aid	5.5 ARCP Decision Aid	5.5 ARCP Decision Aid Updated to include mandatory ES report and MCR requirement and to reflect the title of the examination
9. Equality and diversity	9. Equality and diversity	9. Equality and diversity This section has been updated in line with policy and legislation

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