The Royal College of Pathologists
Pathology: the science behind the cure

Part 1 examination

Molecular Genetics: First paper

Tuesday 24 March 2015

Candidates must answer FOUR questions ONLY

Time allowed: Three hours

1. What are the drivers towards integration of pathology genetic services? What are the opportunities and challenges posed by this model?

2. A sample sent in for genetic testing has been identified via analysis to be of a different gender than that reported on the referral card. Describe the procedure you would follow to investigate this, giving both scientific and technical reasons that could explain the discrepancy, considering both molecular and cytogenetic causes.

3. Describe a comprehensive cost efficient testing strategy for developmental delay in children and adolescents. Describe the limitations of your chosen approach and why this strategy might change over the next 5 years.

4. What is the definition of stratified medicine? Use specific examples of conditions where cytogenetic and/or molecular genetic findings are clinically relevant to stratified medicine.

5. You are invited to contribute to a multi-disciplinary meeting to formulate an approach to deal with incidental findings. What are incidental findings, how do they arise, and why are they a problem? In your opinion what should the approach of genetic laboratories be with regard to incidental findings? Justify your answer using examples from both cytogenetic and molecular laboratories.

The Royal College of Pathologists
Pathology: the science behind the cure

Part 1 examination

Molecular Genetics: First paper

Tuesday 25 March 2014

Candidates must answer FOUR questions ONLY

Time allowed: Three hours

1. Non-invasive prenatal testing (NIPT) for aneuploidy and non-invasive prenatal diagnosis (NIPD) for single gene disorders will revolutionise prenatal screening and diagnosis of genetic conditions. Discuss.

2. The validation and verification of methods is a formal requirement for accreditation according to the standards applicable to genetic testing laboratories. Explain the difference between validation and verification, giving examples of how you might go about each of these with respect to specific laboratory testing protocols.

3. Bioinformatics tools and external genetic databases have become increasingly important resources to diagnostic laboratories. Critically evaluate their use and limitations illustrating your answer with examples of each relevant to clinical cytogenetics and molecular genetics.

4. Discuss the impact that next generation sequencing will have on the delivery of genetics services over the next five years. What are the challenges and opportunities associated with the introduction of this technology?

5. What is the definition of a rare disease? Why is it important to have a strategy for diagnosis and treatment of rare disorders, and in what areas will genetic testing impact on this strategy both now and in the future? Illustrate your answer with specific examples.

The Royal College of Pathologists
Pathology: the science behind the cure

Part 1 examination

Molecular Genetics: Second paper

Tuesday 26 March 2013

Candidates must answer FOUR questions ONLY

Time allowed: Three hours

1. Several large scale genomic studies have gained prominence recently. These include the DDD (Deciphering Developmental Disorders), the ENCODE (Encyclopaedia of DNA Elements), and the 1000 genomes projects. Discuss the aims of these studies, and how the results of these, and others, will provide important resources for array, whole genome or exome sequencing-based diagnostic services.

2. Describe Preimplantation Genetic Diagnosis (PGD) and Preimplantation Genetic Screening (PGS). You have been asked to set up laboratory testing for PGD. What testing methodology would you use, and what are the important factors to consider when developing and running the service.

3. Discuss the importance of de novo mutations in human genetic disease and to service provision (do not include sporadic cancer). Include in your discussion: different types of mutation, mechanism, incidence, predisposition, discovery, pathogenicity.

4. What are the standards which a genetic laboratory should meet to ensure it is providing a high quality service? Explain why these are necessary

5. What is the clinical utility of providing a service for detecting acquired mutations in non-small cell lung cancer? Discuss the type of service you would set up and the issues you might encounter.