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

Part 1 examination

Clinical Cytogenetics/Genetics/Molecular Genetics: First paper

Tuesday 22 March 2016

Candidates must answer FOUR questions ONLY

Time allowed: Three hours

1. You have been asked by your local obstetricians to give a presentation on current prenatal genetic testing and what will impact on service provision over the next 5 years. Include current and future testing options, including the techniques, their advantages and disadvantages.

2. Whole genome sequencing (WGS) can be used for the diagnosis of rare genetic disorders. Discuss the reasons why WGS may not establish a genetic diagnosis in a Mendelian genetic disorder drawing from specific disease examples where possible. How can these limitations be overcome currently and how do you predict this will change in the next five years?

3. Describe how molecular testing has transformed the diagnosis and management of two of the following neoplasms: Gliomas, lung cancer, colorectal cancer, rhabdomyosarcoma.

4. Review the current range of genetic diagnostic services for diseases where epigenetic mechanisms play a role in disease pathogenesis.

5. How does your quality management system contribute to the quality of results obtained in the laboratory?

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Part 1 examination

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.