

Cork Institute of Technology

Masters in Biomedical Sciences - Award

(NFQ – Level 9)

Summer 2007

BM5003 - The Molecular Basis of Disease

(Time: 3 Hours)

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Answer 4 Questions

Use a Separate Answer Book for Each Question

- Q1. Describe the mechanisms of acute rejection of solid organ transplants with reference to methods of preventing rejection.

- Q2. Discuss the process of mapping the locus and identifying the gene leading to an autosomal dominant condition in a family with many affected and unaffected individuals.

- Q3. Discuss the pathophysiology of Duchenne muscular dystrophy and Becker muscular dystrophy.

- Q4. Explain briefly what ion channels are and how they work. With reference to EITHER sudden cardiac death or cystic fibrosis, briefly explain how ion channel mutations can lead to disease.
- Q5. A 39 year old female presents with a spontaneous lower limb deep vein thrombosis. Outline the investigations required to assess for acquired and inherited thrombophilic disorders, and discuss how the results may influence the management of the thrombotic event.
- Q6. Discuss the influence of genetic and environmental factors on the development of type 2 diabetes.
- Q7. Discuss the interaction of genetic and non-genetic factors in tumour development.
- Q8. 'Botulism and tetanus may be considered intoxications rather than infections'. Discuss this statement, supporting your argument with an outline of the cellular and molecular basis of these diseases.
- Q9. Discuss the mechanisms known to cause cell death in Parkinson disease, including how genetic studies have helped identify these pathways.
- Q10. Discuss the regulation of imprinting of the *IGF2* – *H19* locus and outline the types of genetic and epigenetic abnormalities that result in Beckwith-Wiedemann syndrome.