

# Masters Comprehensive – Class of 2018

## Biochemistry Objectives

1. Define anion gap and explain its usefulness in diagnosing metabolic acidosis
2. Describe the regulation of oxygen binding and release by hemoglobin
3. Explain the basis of collagenopathies such as Osteogenesis imperfecta and Ehlers-Danlos syndrome
4. List the types of enzyme inhibition, compare the kinetics of competitive and noncompetitive inhibition and provide examples of each type
5. Compare and contrast the action of uncouplers and inhibitors of electron transport chain
6. Explain the signaling by and the oncogenic mutation of the Ras protein
7. Explain the function of GLUT transporters and their properties
8. Explain the integration of glucose in glycolysis, gluconeogenesis and glycogenolysis and their subcellular locations
9. Describe the role of TCA cycle and the importance of its intermediates
10. Explain the etiology and presentation of Glycogen Storage Diseases
11. Explain the clinical presentation resulting from a G6PD deficiency
12. Explain the basis of mucopolysaccharidoses (MPS) with examples
13. Explain the two phases of  $\beta$ -oxidation of a typical saturated fatty acid and its regulation
14. Provide examples of sphingolipidoses and identify the enzyme defects associated with each
15. Explain the function of the different lipoproteins and the LDLR and how their deficiencies can lead to hypercholesterolemia and hypertriglyceridemias
16. Explain the importance of arginine in Metabolism
17. Describe the catabolism of heme to bilirubin and its metabolism
18. Explain the basis of inherited disorders of nucleotide metabolism
19. Compare and contrast Type 1 and Type 2 diabetes mellitus
20. Describe the effects of insulin and glucagon on carbohydrate metabolism
21. Explain the role of fat-soluble/water-soluble vitamins in metabolism
22. Describe the diagnostic parameters of iron-deficiency anemia and hemochromatosis

## Medical Genetics Objectives

1. Explain the process of prokaryotic/eukaryotic DNA replication including proofreading
2. Describe the diseases associated with defects in DNA repair pathways
3. Describe the steps in eukaryotic transcription
4. Explain the various types of mutations that affect the genetic code
5. Describe the mechanism of action of the following classes of inhibitors: Aminoglycosides; Tetracyclines; Chloramphenicol; Macrolides; Lincosamides; Oxazolidinones; Diphtheria toxin
6. Describe the mechanisms for alternative eukaryotic posttranscriptional regulation: Alternative splicing; Alternative poly A signals
7. Describe the process of genomic imprinting and give examples of diseases associated with it

8. Describe the different types of chromosomal translocations and provide examples of diseases associated with them
9. Explain the use of oligonucleotide microarrays as a tool in molecular medicine
10. Calculate the gene/genotype frequency of an autosomal recessive disorder in a given population
11. Define oncogenes and tumor suppressor genes and describe the concept of loss of heterozygosity
12. Describe general and targeted chemotherapeutic approaches to treatment of cancer
13. Determine the mode of inheritance from a given pedigree
14. Explain the calculation of recurrence risk of single gene disorders
15. Explain how genetic variations of both drug metabolizing enzymes and drug targets affect individual responses to drug treatment
16. Describe the methods of prenatal diagnosis and provide examples of prenatal screening
17. Describe treatment strategies that manipulate metabolism and/or increase the function of the affected gene or protein