chemistry books, visit our website www.ejaypee.com, for detailed information on biochemistry books, visit our website www.ejaypee.com, for detailed information on biochemistry books

diginerve®
A Jaypee Initiative
• Video Lectures • Dr. Wise AI Chatbot
• Question Bank • Notes
Download App

Early Clinical Exposure
A Case Based Approach in
Clinical Biochemistry

As per the Revised Competency-based Medical Education Curriculum (NMC)



Anita Chalak
Priti Puppalwar
Yogesh Pawade

Foreword
Vedprakash Mishra

2nd Edition

JAYPEE

Contents

1. COVID-19 1

Section 1: Carbohydrates

2. Lactose Intolerance 5
3. Hereditary Fructose Intolerance 6
4. Hurler Syndrome (MPS-I) 8
5. Sanfilippo Type A Syndrome (MPS-III) 11
6. Classical Galactosemia 13
7. Von Gierke's Disease 15
8. G6PD Deficiency 17
9. Type 1 DM 20
10. Type-II Diabetes Mellitus 22
11. Diabetic Ketoacidosis 24
12. Gestational Diabetes Mellitus 27
13. Neonatal Hypoglycemia 31
14. Renal Glucosuria 34
15. Diabetic Neuropathy 36
16. Diabetic Retinopathy 39
17. Pompe's Disease 42

Section 2: Lipids

18. Steatorrhea 45
19. Phrynoderma 47
20. Acute Respiratory Distress Syndrome 49
21. Zellweger's Syndrome 51
22. Alcoholic Fatty Liver Disease 53
23. Non-alcoholic Fatty Liver Disease 55
24. Atherosclerosis 57
25. Myocardial Infarction 60
26. Primary Familial Hypercholesterolemia (Type-II A Hyperlipoproteinemia) 62

Section 3: Proteins and Amino Acids

27. Carbamoyl Phosphate Synthetase-1 Deficiency 65
28. Ornithine Transcarbamylase Deficiency 68
29. Phenylketonuria 70
30. Alkaptonuria 74
31. Albinism 77
32. Parkinson's Disease 80
33. Cystinuria 83
34. Hartnup's Disease (Pellagra-like Dermatosis) 86
35. Classical Homocystinuria 89
36. Maple Syrup Urine Disease 93

Section 4: Nucleotides

37. Gout 95
38. Lesch-Nyhan Syndrome 97
39. Orotic Aciduria 99

Section 5: Vitamins

40. Vitamin A Deficiency 101
41. Vitamin K Deficiency 104
42. Rickets 106
43. Osteomalacia 109
44. Beriberi 111
45. Wernicke-Korsakoff Syndrome 113
46. Riboflavin (Vitamin B_2) Deficiency 115
47. Pellagra 117
48. Pyridoxin (Vitamin B_6) Deficiency 119
49. Pernicious Anemia (Vitamin B_{12}) Deficiency 121
50. Folic Acid Deficiency 124
51. Scurvy 126

Section 6: Minerals

52. Goiter 129
53. Graves' Disease (Hyperthyroidism) 132
54. Pheochromocytoma 136
55. Iron Deficiency Anemia 139
56. Tetany 141
57. Wilson's Disease 144

Section 7: Heme and Hemoglobinopathies

58. Acute Intermittent Porphyria 147
59. Porphyria Cutanea Tarda 149
60. Physiological Jaundice 152
61. Hemolytic Jaundice 156
62. Hepatic Jaundice 159
63. Obstructive Jaundice 162
64. Sickle Cell Anemia 165
65. Thalassemia 169

Section 8: Acid Base and Water Electrolyte Balance

66. Dehydration 173
67. Metabolic Acidosis 176
68. Metabolic Alkalosis 180
69. Respiratory Acidosis 183
70. Respiratory Alkalosis 185

Section 9: Nutrition

71. Kwashiorkor 187
72. Marasmus 190
73. Obesity 193

Section 10: Genetic Disorders

74. Down Syndrome 197
75. Turner Syndrome 200
76. Klinefelter Syndrome 204

Section 11: Miscellaneous

77. Metabolic Syndrome 209
78. Starvation 212
79. Cushing's Syndrome 216
80. Myxedema 219
81. Nephrotic Syndrome 221
82. Multiple Myeloma 224
83. Bacterial Meningitis 227
84. Viral Meningitis 230
85. Acute Pancreatitis 232
86. Chronic Kidney Disease 235

G6PD Deficiency

8
CHAPTER

CASE STUDY

A 30-year-old male was suffering from malaria and was put on the drug, Primaquine. During the treatment, he developed signs of anemia (pallor, fatigue, breathlessness). He gave history of dark colored urine since the episode of illness. Laboratory tests indicated reduced blood hemoglobin levels, raised serum bilirubin levels and the liver enzymes (Sr. AST and ALT) showed mild rise.

QUESTIONS

1. What is the probable diagnosis? Justify.

Ans: The probable diagnosis is hemolytic jaundice due to deficiency of enzyme glucose-6-phosphate dehydrogenase (G6PD).

- **Key factors in case scenario:** Suffering from malaria, on treatment with primaquine, pallor, fatigue, breathlessness, dark colored urine
- **Key factors in examination/investigations:** Reduced blood hemoglobin levels, raised serum bilirubin levels and mild rise in liver enzymes (Sr. AST and ALT)

2. What is the incidence of the disorder?

Ans: G6PD deficiency is an X-linked recessive disease and thus it affects males.

3. Explain the biochemical basis of the disorder.

Ans:

- It affects all the cells but red blood cells (RBCs) are the most affected, because HMP shunt is the only source of NADPH in the RBC.
- G6PD (rate limiting enzyme of HMP shunt) deficiency leads to less production of NADPH from HMP shunt resulting in lowered levels of reduced glutathione, which makes the erythrocytes susceptible to hemolysis.
- G6PD converts NADP to its reduced form, NADPH, in the pentose phosphate pathway (HMP shunt) **(Fig. 8.1)**.
- Reactive oxygen species (ROS) are continuously produced in cells. They are capable of damaging all the cells of body, including RBCs. Glutathione in its reduced form counter-attacks these ROS and thus maintains the membrane integrity of RBCs. NADPH formed in pentose phosphate pathway, converts oxidized glutathione to reduced glutathione. Thus, NADPH plays an important role in eliminating the free radicals from the cell **(Fig. 8.2)**.
- G6PD deficiency occurs with increased frequency throughout Africa, Asia, the Mediterranean, and the Middle East. Prevalence of deficiency is correlated with geographical distribution of malaria, as persons with G6PD deficiency are protected from malaria caused by *Plasmodium falciparum*.

JAYPEE BROTHERS

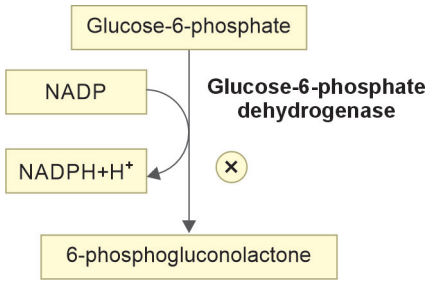

Fig. 8.1: A step in pentose phosphate pathway.

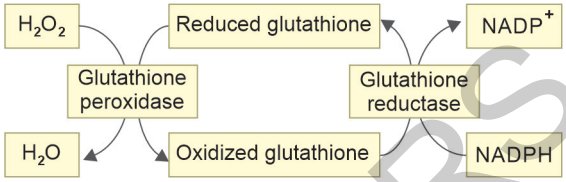

Fig. 8.2: Formation of reduced glutathione.

- The oxidation of hemoglobin leads to the formation of Heinz bodies. Removal of Heinz bodies by spleen leads to formation of "Bite cells" (RBCs that have lost a peripheral portion of the cell).

4. What are the characteristic clinical features of the disorder?

Ans:

- **Hemolytic anemia:** It is the most common characteristic feature of G6PD deficiency. It occurs due to destruction of red blood cells. This leads to jaundice, fatigue and shortness of breath.
- **Kernicterus:** Hemolytic anemia may lead to kernicterus in newborn resulting in reduced psychomotor development, neurosensory hypoacousia, absence of speech and poor reflex of the pupil to light.

5. What are the triggering factors for G6PD deficiency?

Ans:

- Ingestion of fava beans, certain legumes
- Drugs, such as antimalarial drug primaquine, dapsone, sulfonamides, aspirin, nonsteroidal anti-inflammatory drugs, nitrofurantoin
- Miscellaneous: Vitamin K (water-soluble form), doxorubicin, methylene blue, nalidixic acid, furazolidone, niridazole, phenazopyridine
- Naphthalene impregnated clothes

6. Why fava beans ingestion leads to hemolytic crisis in G6PD deficiency patients?

Ans: The oxidants in Vicia fava are two glycosides whose aglycones, when auto-oxidized, produce oxygen free radicals. The incidence of favism is highly variable due to variations in concentration, in absorption, or in metabolism of the aglycones.

7. What investigations are required to diagnose the condition?

Ans:

- G6PD screening tests: Fluorescent spot test, detecting the generation of NADPH from NADP. The test is positive if the blood spot fails to fluoresce under ultraviolet light.
- Quantitative estimation of G6PD: Normal level of G6PD: 7.9–16.3 U/g Hb
- Reticulocyte count and mean corpuscular hemoglobin

8. How can this patient be managed?

Ans:

- Avoidance of oxidative stressors is the main treatment for G6PD deficiency.
- Rarely, anemia may be severe enough to warrant a blood transfusion.

JAYPEE BROTHERS

- Adequate urine flow should be maintained if hemoglobinuria develops during an acute hemolytic episode.
- Hemolytic episodes can be prevented by warning patients about risks posed by oxidant drugs and fava beans and by prompt treatment of infections.

BIBLIOGRAPHY

1. Jameson J, Fauci AS, Kasper DL, Hauser SL, Longo DL, Loscalzo J. Harrison's Principles of Internal Medicine, 12th edition. New York: McGraw Hill; 2018.
2. Vasudevan DM, Sreekumari S, Vaidyanathan K. Textbook of Biochemistry for Medical Students, 8th edition. New Delhi: Jaypee Brothers Medical Publishers (P) Ltd; 2019.

JAYPEE BROTHERS

for detailed information on biochemistry books, visit our website www.ejaypee.com, for detailed information on biochemistry books, visit our website www.ejaypee.com, for detailed infor

Early Clinical Exposure: A Case Based Approach in

Clinical Biochemistry

Salient Features

- Fully aligned with the "Early Clinical Exposure (ECE)" guidelines as outlined in the 2024 Competency-Based Medical Education (CBME) curriculum by the National Medical Commission (NMC).
- Includes commonly encountered clinical cases presented in a question–answer format, aiding in conceptual clarity and examination readiness.
- Provides balanced and appropriate case distribution across various content areas, ensuring comprehensive curriculum coverage.
- Features carefully curated illustrations to enhance the understanding of clinico-biochemical correlations in real-life contexts.
- Enriched with concise photographs and informative graphics to promote visual learning and deeper retention of key concepts.

Anita Chalak (Kale) MD (Biochemistry) MHPE PhD (HPE) GSMC-FAIMER is a Professor and Head in Department of Biochemistry at Seth GS Medical College (GSMC) and King Edward Memorial (KEM) Hospital, Mumbai, Maharashtra, India, and has a noticeable qualitative experience of more than 26 years in Clinical Biochemistry. Her excellence in Medical Education is evident from her accolades being MHPE and PhD (HPE). She has been a forerunner in framing student learning objectives (SLOs) for competencies in the subject of Biochemistry for CBME curriculum. She has more than 50 national and international publications, 9 copyrights and 5 books to her honor. With her extensive experience in the subject, she has articulated the flow of the content methodically, making it more lucid and persuasive.

Priti Puppalwar MD (Biochemistry) is a Professor and Head in Department of Biochemistry at Karmaveer Ma Sa Kannamwar Government Medical College, Chandrapur, Maharashtra, India. She has almost 19 years of extensive academic experience in Clinical Biochemistry and Medical Education, having formally trained with GSMC-FAIMER International Fellowship. She has a research track with 18 national and international publications. Her in-depth and comprehensive subject knowledge has essentially made this compilation of clinical cases more ingeniously credible.

Yogesh Pawade MD DNB (Biochemistry) PGDHHCM MNAMS is a Professor in Department of Biochemistry at All India Institute of Medical Sciences (AIIMS), Nagpur, Maharashtra, India. Besides having wide experience in Clinical Biochemistry, he is trained in Medical Education and CBME, with ACME and GSMC-FAIMER International Fellowship. He holds recognition for more than 27 publications, 7 copyrights and 3 books to his position. He contributed to this book substantially by edging the content of the book graphically, assembling it into a credibly resourceful package.

Shelving Recommendation
BIOCHEMISTRY

ISBN 978-93-6616-722-0

9 789366 167220

Buy from ejaypee

JAYPEE BROTHERS