Biochemistry

| System | Biochemical Processes | Biochemical Disorders |
|---|---|---|
| **General Biochemistry** | - DNA structure, replication, repair - RNA transcription, processing, translation - Protein folding, post-translational modifications - Enzyme kinetics and regulation - Energy metabolism (ATP production, oxidative phosphorylation) - Carbohydrate, lipid, amino acid, nucleotide metabolism - Signal transduction pathways (MAPK, JAK-STAT, GPCR) - pH regulation and buffer systems | - Inborn errors of metabolism (e.g., phenylketonuria, homocystinuria) - Lysosomal storage diseases (e.g., Gaucher, Tay-Sachs, Niemann-Pick) - Mitochondrial disorders (e.g., MELAS, Leigh syndrome) |
| **Hematopoietic & Lymphoreticular** | - Heme biosynthesis and degradation - Iron metabolism and transport (ferritin, transferrin) - Purine and pyrimidine metabolism (nucleotide synthesis) | - Hemoglobinopathies (sickle cell disease, thalassemia) - Porphyria (acute intermittent porphyria, erythropoietic porphyria) - G6PD deficiency - Hyperuricemia and gout |
| **Central & Peripheral Nervous** | - Neurotransmitter biosynthesis and degradation (dopamine, serotonin, GABA, glutamate) - Brain energy metabolism (glucose, ketone bodies) - Blood-brain barrier and transport systems | - Neurodegenerative disorders (Alzheimer's, Parkinson's, Huntington's) - Phenylketonuria, maple syrup urine disease - Tay-Sachs, Krabbe, metachromatic leukodystrophy - Wilson disease (copper metabolism defect) |
| **Skin & Connective Tissue** | - Collagen and elastin biosynthesis - Keratinization and skin barrier function - Melanin biosynthesis (tyrosinase, albinism) | - Ehlers-Danlos syndrome, Marfan syndrome - Osteogenesis imperfecta - Scurvy (vitamin C deficiency) - Albinism, vitiligo |
| **Musculoskeletal** | - Muscle contraction biochemistry (actin, myosin, troponin) - Bone mineralization and calcium metabolism - ATP production in muscle (glycolysis, oxidative phosphorylation) | - Duchenne muscular dystrophy - Rickets, osteomalacia - Gout (uric acid metabolism disorder) - McArdle disease (glycogen storage disorder) |
| **Respiratory** | - Oxygen transport and hemoglobin function - Acid-base balance and $CO_2$ buffering - Surfactant synthesis and lung biochemistry | - Chronic obstructive pulmonary disease (COPD) - Respiratory acidosis/alkalosis - Neonatal respiratory distress syndrome (deficiency of surfactant) |
| **Cardiovascular** | - Lipid metabolism (cholesterol, triglycerides, lipoproteins) - Coagulation cascade biochemistry - Nitric oxide and vascular regulation | - Familial hypercholesterolemia - Atherosclerosis and dyslipidemias - Hypertension (renin-angiotensin-aldosterone system) - Homocystinuria (vascular complications) |
| **Gastrointestinal** | - Digestion and absorption of macronutrients - Bile acid metabolism and enterohepatic circulation - Pancreatic enzyme biochemistry | - Lactose intolerance - Celiac disease (gluten metabolism) - Wilson disease (copper transport disorder) - Gilbert syndrome (bilirubin metabolism defect) |
| **Renal/Urinary** | - Electrolyte and fluid homeostasis - Acid-base balance and renal buffering - Urea cycle and nitrogen excretion | - Renal tubular acidosis - Fanconi syndrome - Cystinuria (amino acid transport defect) - Hyperammonemia (urea cycle disorders) |

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| **Reproductive** | - Steroid hormone biosynthesis (estrogen, testosterone) - Follicular and luteal phase hormonal regulation - Sperm metabolism and capacitation | - Congenital adrenal hyperplasia - Aromatase deficiency - Klinefelter and Turner syndromes - Polycystic ovarian syndrome (PCOS) |
| **Endocrine** | - Hormone synthesis, secretion, receptor signaling - Insulin and glucose metabolism - Thyroid hormone biosynthesis - Parathyroid hormone and calcium metabolism | - Diabetes mellitus (Type 1 and Type 2) - Cushing syndrome (cortisol excess) - Addison disease (adrenal insufficiency) - Hypothyroidism and hyperthyroidism |