Achievable

USMLE STEP 1 DUMP SHEET

Biochemistry and metabolism

Enzymes and energy

- **Km:** [S] at $\frac{1}{2}V_{max}$; low Km, high affinity
- **Competitive:** Km up, Vmax same (statins)
- **Noncompetitive:** Vmax down, Km same
- **ΔG < 0:** spontaneous; enzymes leave ΔG unchanged

Electron transport chain

- **I:** rotenone. **III:** antimycin A. **IV:** cyanide, CO
- **V (ATP synthase):** oligomycin
- **Uncouplers:** 2,4-DNP, high-dose aspirin, UCP1: heat
- **ROS:** superoxide dismutase, catalase

Glycolysis, TCA, gluconeogenesis

- **Glucokinase:** high Km; liver, β cells (MODY)
- **PFK-1** (rate-limiting): + AMP, F2,6-BP; − ATP, citrate
- **Arsenate:** no ATP at phosphoglycerate kinase
- **PDH, α-KG dehydrogenase:** B1, lipoic acid, CoA, FAD, NAD
- **Isocitrate dehydrogenase:** TCA rate-limiting; 1 NADH
- **Per acetyl-CoA:** 3 NADH, 1 FADH2, 1 GTP
- **Bypasses:** pyruvate carboxylase (biotin), PEPCK, fructose 1,6-bisphosphatase, glucose 6-phosphatase
- **Glucose from pyruvate:** 4 ATP, 2 GTP; not from leucine, lysine

Glycogen storage diseases

Type	Enzyme	Features
I Von Gierke	Glucose 6-phosphatase	Severe fasting hypoglycemia, lactic acidosis, high urate and lipids, big liver
II Pompe	Lysosomal acid maltase	Cardiomegaly, hypotonia, big tongue; glucose normal
III Cori	Debranching enzyme	Milder fasting hypoglycemia, limit dextrins, ketosis
IV Andersen	Branching enzyme	Polyglucosan; cirrhosis and liver failure in infancy
V McArdle	Muscle phosphorylase	Exercise cramps, myoglobinuria; lactate fails to rise

Lysosomal storage diseases

Disease	Enzyme	Stored	Clues
Gaucher	Glucocerebrosidase	Glucocerebroside	Most common; hepatosplenomegaly, bone, Gaucher cells
Tay-Sachs	Hexosaminidase A	GM2 ganglioside	Neurodegeneration, cherry-red macula, no organomegaly
Niemann-Pick	Sphingomyelinase	Sphingomyelin	Cherry-red macula, hepatosplenomegaly, foam cells
Krabbe	Galactocerebrosidase	Galactocerebroside	Globoid cells, demyelination
Metachromatic	Arylsulfatase A	Sulfatides	Demyelination, ataxia, dementia
Fabry (X-linked)	α-Galactosidase A	Ceramide trihexoside	Angiokeratomas, burning limb pain, renal failure
Hurler	α-L-Iduronidase	Heparan, dermatan	Corneal clouding. **Hunter** (X-linked): no clouding

Amino acid disorders

- **PKU:** phenylalanine hydroxylase; musty odor, fair skin
- **Malignant PKU:** dihydropteridine reductase (BH4)
- **MSUD:** branched-chain α-ketoacid dehydrogenase
- **Alkaptonuria:** dark urine, ochronosis, arthritis
- **Homocystinuria:** lens dislocation, thrombosis

Urea cycle and porphyrias

- **OTC deficiency** (X-linked): high orotic acid
- **CPS I, NAGS:** orotic acid normal or low
- **Orotic aciduria:** megaloblastic, ammonia normal
- **Lead:** ALA dehydratase, ferrochelatase; stippling
- **AIP:** PBG deaminase; abdominal pain; glucose, heme
- **PCT:** uroporphyrinogen decarboxylase; blisters

Sugar and fat disorders

- **Galactosemia:** uridyltransferase; cataracts, E. coli sepsis
- **Fructose intolerance:** aldolase B; hypoglycemia
- **MCAD:** hypoketotic hypoglycemia with fasting
- **Lesch-Nyhan:** HGPRT; self-mutilation, gout

Lipoproteins

- **Apo B-48:** chylomicrons. **C-II:** activates LPL
- **Type I:** LPL or apo C-II. **IIa:** LDL receptor
- **Abetalipoproteinemia:** MTP. **Tangier:** ABCA1