PIPADOR REVISION

MRCOG Part 1 Revision Notes

Proteins: Essential Amino Acids

BIOCHEMISTRY • LECTURE 223

VISUAL EXAMINATION REVISION NOTES

2026 Edition

MRCOG PART 1 REVISION NOTES | BIOCHEMISTRY

223 Proteins: Essential Amino Acids

Essential and conditionally essential amino acids, their products, phenylketonuria and maternal PKU, and other inborn errors

9	Phe → Tyr	120–360	Day 5
Essential amino acids	Tyrosine becomes essential in PKU	Target maternal Phe (µmol/L) in PKU	Newborn bloodspot screening

■ KEY FACTS: Learning objectives

- List the essential, conditionally essential and non-essential amino acids.
- Describe the metabolic products of key essential amino acids.
- Describe inborn errors of essential amino acid metabolism — especially phenylketonuria and maternal PKU — and newborn screening.

223.1 Classification

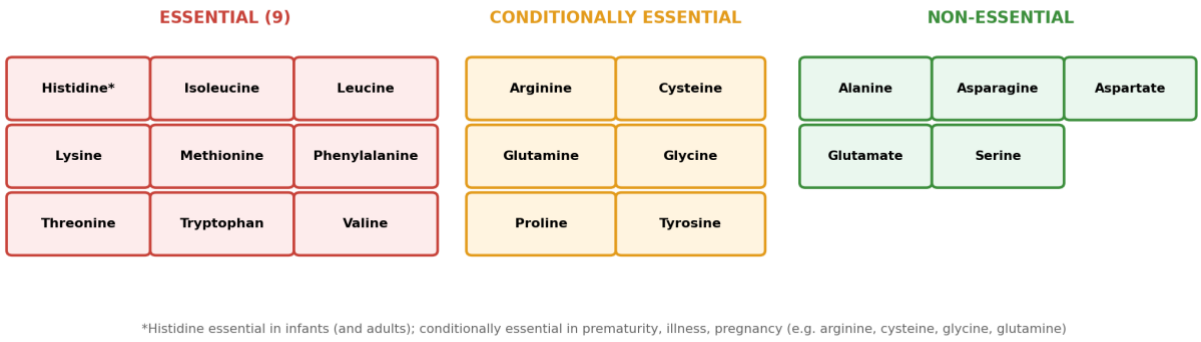


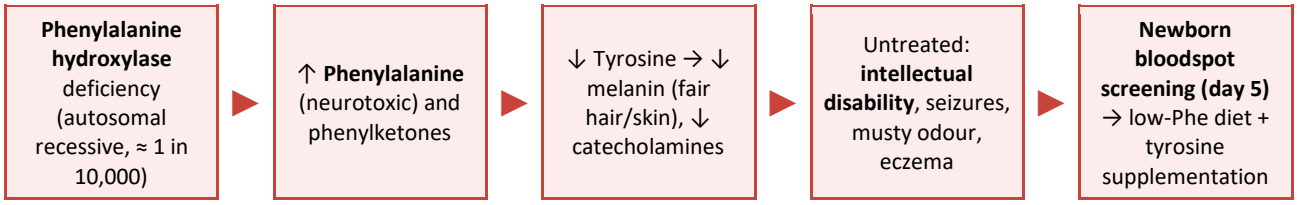
Figure 223.1 — Essential, conditionally essential and non-essential amino acids

Essential amino acids cannot be synthesised (carbon skeleton) by humans and must come from the diet. **Conditionally essential** amino acids become essential when synthesis cannot meet demand: **prematurity** (cysteine, tyrosine, arginine, glycine — immature enzymes), illness/catabolism (glutamine), **pregnancy** (rapid growth), and specific enzyme defects (**tyrosine in PKU**).

223.2 Important products of essential amino acids

Amino acid	Key products / roles
Phenylalanine	→ Tyrosine (phenylalanine hydroxylase, BH4) → DOPA → dopamine → noradrenaline → adrenaline; thyroid hormones; melanin
Tryptophan	→ Serotonin (5-HT) → melatonin; → niacin (B_3) (pellagra if deficient — carcinoid, Hartnup)
Methionine	→ SAM (universal methyl donor — DNA methylation, epigenetics) → homocysteine (→ cysteine via B_6; → methionine via B_{12}/folate); start codon
Lysine	Collagen cross-links (hydroxylysine), carnitine synthesis, histone acetylation
Leucine, isoleucine, valine (BCAAs)	Muscle fuel and protein synthesis (leucine → mTOR); maple syrup urine disease
Histidine	→ Histamine; Hb buffering; essential in infants
Threonine	Mucins, glycine synthesis

223.3 Phenylketonuria (PKU)



■ **RED FLAGS**

- **Maternal PKU syndrome**: high maternal phenylalanine **crosses the placenta (active transport — fetal levels higher than maternal)** and is teratogenic → **microcephaly, intellectual disability, congenital heart disease, FGR**, dysmorphism — even when the fetus does not have PKU.
- **Pre-conception** return to a strict low-Phe diet, target **phenylalanine 120–360 µmol/L before conception and throughout pregnancy**, with frequent (weekly) monitoring by a specialist metabolic team; sapropterin in responsive cases.

223.4 Other disorders

Disorder	Defect	Features / O&G relevance
Maple syrup urine disease	Branched-chain ketoacid dehydrogenase	Neonatal encephalopathy, sweet-smelling urine; bloodspot screening
Homocystinuria	Cystathionine β-synthase (B_6-dependent)	Marfanoid habitus, lens dislocation (downward), intellectual disability, arterial and venous thrombosis (↑ in pregnancy/postpartum — LMWH), screening
Tyrosinaemia type 1	Fumarylacetoacetate hydrolase	Liver failure, renal Fanconi
Alkaptonuria	Homogentisate oxidase	Dark urine on standing, ochronosis, arthritis
Albinism	Tyrosinase	Absent melanin
Hartnup disease	Neutral amino acid transporter (tryptophan)	Pellagra-like rash

UK newborn bloodspot screening (≈ day 5) includes **PKU, MCADD, MSUD, isovaleric acidaemia, glutaric aciduria type 1, homocystinuria**, plus sickle cell disease, congenital hypothyroidism and cystic fibrosis (check current programme).

223.5 Must-know summary

■ **EXAM TIP**

- 9 essential amino acids — **PVT TIM HaLL**; conditionally essential in preterm/illness/pregnancy; **tyrosine essential in PKU**.
- Phe → Tyr → catecholamines, thyroid hormone, melanin; Trp → serotonin, melatonin, niacin; Met → SAM, homocysteine.
- PKU: PAH deficiency, bloodspot screening; **maternal PKU** → microcephaly, CHD — Phe 120–360 µmol/L pre-conception.
- Homocystinuria: lens dislocation, **thrombosis**.

■ **MNEMONIC / MEMORY AID**

Essential amino acids — 'PVT TIM HaLL': Phenylalanine, Valine, Threonine, Tryptophan, Isoleucine, Methionine, Histidine, Leucine, Lysine.