

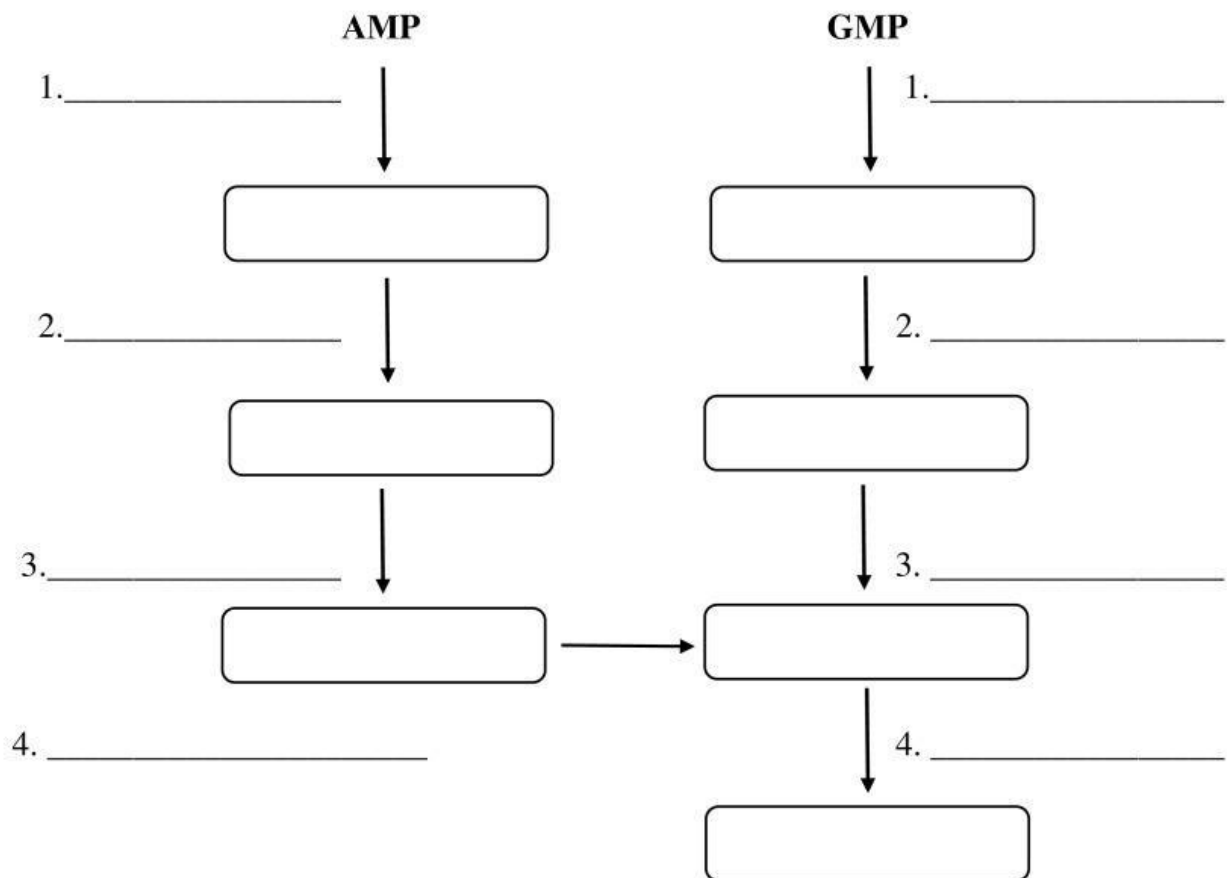
Topic 29. Biosynthesis and catabolism of purine nucleotides. Hereditary disorders of nucleotide metabolism

Theoretical questions:

1. Purine nucleotide catabolism. Formation of uric acid.
2. De novo biosynthesis of purine nucleotides and its regulation.
3. Origin of purine ring atoms.
4. Biosynthesis of purine nucleotides by salvage pathway.
5. Hereditary disorders of purine nucleotide metabolism: a) hyperuricemia, b) gout, c) Lesch-Nyhan syndrome.

Study Questions and Tasks

1. Catabolism of purine nucleotides. Write a scheme of catabolism of AMP, GMP. Indicate metabolites and enzymes.

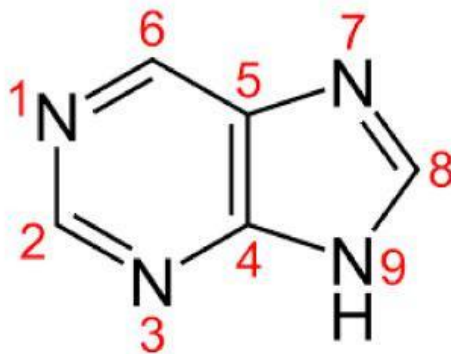


2. Uric acid, its properties, excretion from human organism

3. Specify the origin of the atoms of a purine ring:

Nitrogen

1. _____
3. _____
7. _____
9. _____

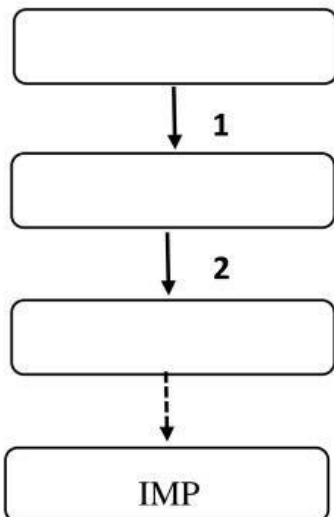


Carbon

2. _____
4. _____
5. _____
6. _____
8. _____

4. *De novo* biosynthesis of purine nucleotides

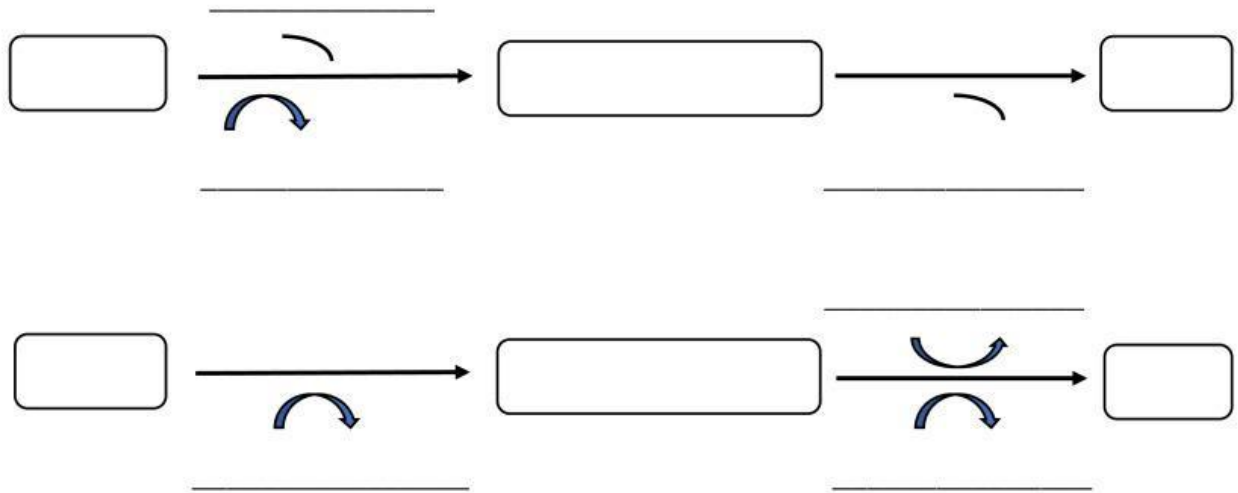
a. Scheme of IMP synthesis



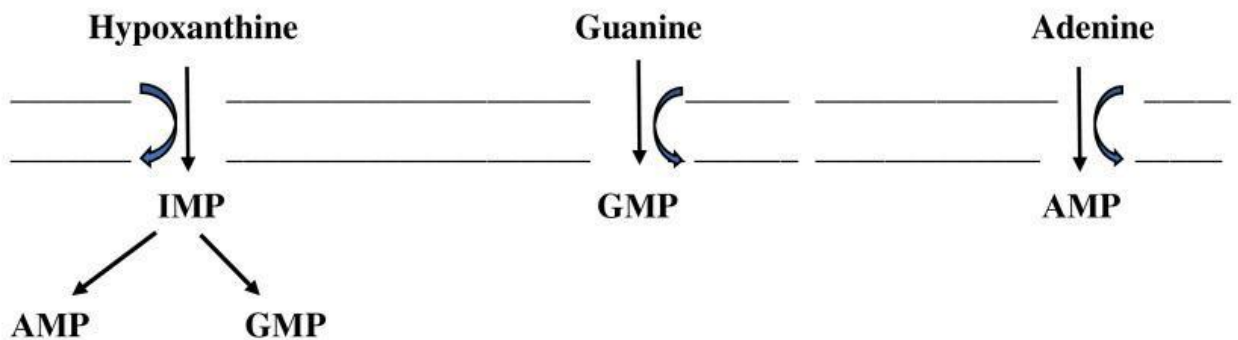
Enzymes

1. _____
2. _____

b. Show conversion of inosinic acid (IMP) to AMP and GMP.



5. Salvage pathway of purine nucleotide biosynthesis.



6. Hereditary metabolic disorders of purine nucleotides. Clinical and biochemical characteristics of:

a) hyperuricemia

b) gout

c) Lesch-Nyhan syndrome

Case tasks

1. A 48-year-old patient consulted a doctor with complaints of severe pain, swelling, and redness in the joints. Based on laboratory analysis, the diagnosis of gout was confirmed. What analysis was performed to make the diagnosis? Write a scheme for the formation of the analyzed metabolite.

2. A 53-year-old man was diagnosed with urolithiasis with elevated urate levels. He was prescribed allopurinol. Allopurinol is an inhibitor of which enzyme, and what is its mechanism of action?

3. An 8-year-old boy with signs of cerebral palsy has an increased concentration of uric acid in his blood. The boy has impaired thinking. He is aggressive and self-mutilates. What hereditary disease does the boy have? What enzyme defect is the cause of this disease? List the reactions and indicate the site of action of the enzyme.

Textbooks:

1. Biochemistry 5th Edition Ch.22, pp. 292-301.
2. Prasad textbook of biochemistry OCR. Topic 19, pp.405-415.