A patient who has a glutamine synthetase deficiency would have all of the following EXCEPT:

A. Glutamate amination to glutamine is compromised
B. Transport of ammonia from most tissues to liver is hindered
C. Toxic levels of ammonia may accumulate in the patient's tissues and/or blood
D. Transport of ammonia from muscle cells to the liver is not affected
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A new born who refuses feeding has been diagnosed with cystathionine-β- synthase deficiency. What is the diagnosis of his condition?

A. Albinism
B. Homocystinuria
C. Maple syrup urine disease
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Protein digestion is continued in small intestine after partial digestion begins in stomach. Celiac disease is a malabsorption disease due to immune-mediated damage of intestinal cells. Which of the following describes protein digestion and absorption at the small intestine?

A. Pancreatic trypsin and chymotrypsin are exopeptidases that cleave the N-terminus of a polypeptide
B. The intestinal aminopeptidase cleaves the C-terminus of a polypeptide
C. The resulting free amino acids are absorbed by an H^+-linked transport system
D. The di- and tripeptides are absorbed by an Na^+-linked transport system
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Which of the following is the INCORRECT match?

Amino acid	Catabolic intermediate	Glucogenic and/or ketogenic
A. Tyrosine	Fumarate	Glucogenic and ketogenic
B. Proline	α-ketglutarate	Glucogenic
C. Serine	Glutamate	Ketogenic
D. Arginine	α-ketglutarate	Glucogenic
E. Threonine	Succinyl-CoA	Glucogenic

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A patient with Lesch-Nyhan syndrome has visited your clinic. He has a lesion on his lip. In this syndrome, nucleic acid metabolism is compromised. Which of the following describes his condition?

A. Purine denovo synthesis is reduced but purine salvage is increased

B. HGPRT enzyme deficiency results in an inability to salvage guanine and hypoxanthine

C. APRT enzyme deficiency results in an inability to salvage adenine

D. Purine degradation is decreased resulting in hypouricemia

E. AMP, GMP and IMP levels are all reduced

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Phosphorylation of nucleoside monophosphates to nucleoside triphosphates occurs as follows:

A. Base-specific nucleoside monophosphate kinases phosphorylate nucleoside monophosphates in a two-step reaction

B. Base-specific nucleoside diphosphate kinase phosphorylates nucleoside monophosphates

C. General nucleoside monophosphate kinases phosphorylate nucleoside monophosphates followed by the base-specific nucleoside diphosphate kinase

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All of the following are inhibitors of purine or pyrimidine denovo synthesis <u>EXCEPT</u>:

A. PRPP

B. 5-FdUMP

C. Methotrexate

D. GMP

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