Experiment (2):

1-What are the defects that causing congenital Hyperammonemia?

Type 1: Deficiency of the enzyme ornithine carbamoyltransferase
Type 2, due to deficiency of the enzyme carbamoyl phosphate synthetase

2-What are the defects that causing acquired Hyperammonemia?

Liver diseases, such as viral hepatitis, or excessive alcohol consumption.

3-What causes Reye's syndrome?

The exact cause of Reye's syndrome is unknown, although several factors may play a role in its development. Some cases of Reye's syndrome are presented with different degrees of encephalopathy, hyperammonemia and hypoglycemia.

Experiment (3):

1- Do either of the patients suffer from celiac disease, and if so which one?

That is according to your sample and given normal range

2- In the function of the upper small intestine normal in these patients?

That is according to your sample and given normal range

3- How was the interference of other reducing sugars minimized in your test?

That by measuring at different wave length, because both pentose and hexose react with o-toluidine. Pentose developing a brown color shows maximum absorbance at 475 nm, while hexoses develop a green color with maximum absorbance at 630nm.

Experiment (5):

1-What is the Familial (LCAT) deficiency? And how it effects the level of cholesterol in blood?

That one type of Lecithin cholesterol acyltransferase deficiency, LCAT catalyzes the formation of cholesterol esters in lipoproteins

This disease as two forms

- *Familial LCAT deficiency in which there is complete LCAT deficiency.*
- *Fish eye disease in which there is a partial deficiency.*

Both are autosomal recessive disorders caused by mutations of the LCAT gene located on chromosome 16q22

Effect of lacking this enzyme:
A deficiency of LCAT causes accumulation of un-esterified cholesterol in certain body tissues. Cholesterol effluxes from cells as free cholesterol and is transported in HDL as esterified cholesterol LCAT is the enzyme that esterifies the free cholesterol on HDL to cholesterol ester and allows the maturation of HDL. LCAT deficiency does not allow for HDL maturation resulting in its rapid catabolism of circulating apoA-1 and apoA-2. The remaining form of HDL resembles nascent HDL

2-What is the Familial (LPL) deficiency?

Familial Lipoprotein Lipase Deficiency presents in childhood and is characterized by very severe hypertriglyceridemia. Clearance of chylomicrons from the plasma is impaired, causing triglycerides to accumulate in plasma and the plasma to have a milky appearance

Experiment (7):

Why should PKU patients avoid aspartame?

Aspartame is a methyl ester of the aspartic acid/phenylalanine dipeptide because its breakdown products include phenylalanine, aspartame must be avoided by people with the genetic condition phenylketonuria (PKU).

Experiment (8):

What is the importance of Glutathione in our body?

i. *It is the major endogenous antioxidant produced by the cells, as well as maintaining exogenous antioxidants such as vitamins C and E in their reduced (active) forms.*
ii. *Regulation of the nitric oxide cycle is critical for life, but can be problematic if unregulated.*

iii. *It is used in metabolic and biochemical reactions such as DNA synthesis and repair, protein synthesis, prostaglandin synthesis, amino acid transport, and enzyme activation.*
iv. *Glutathione (GSH) participates in leukotriene synthesis and is a cofactor for the enzyme glutathione peroxidase.*
v. *Glutathione is also needed for the detoxification of methylglyoxal, a toxin produced as a byproduct of metabolism.*

2. Discuss the problems you encountered during this experiment?

The standard reading was it valid, for some reason.

3. Can you explain the assay basic principle?

As given in your handout

4. What Precautions you must take while doing this experiment?

- *collect the sample in heparinized tube*
- *Keep glutathione solution and DNTB reagent in dark bottle*
- *use fresh solutions*

Experiment (9):

1-What is the EC number of arginase? And in which group of enzymes it belongs to?
Arginase *(EC 3.5.3.1) ,belongs to hydrolyses enzyme.*

2-From previous studies find the kinetics of human arginase; optimum PH , optimum temperature and the km for arginase to arginine.

Optimal pH: *9.4* ***Optimal temperature:*** *37◦C*

Km Value:

Different studies on Wild-type human arginase I has shown a different values of Km of arginas to arginine, However the arginase in human km to arginine exhibits KM values for L-arginine hydrolysis of 0.08 mM at pH 8.5 and 0.02 mM at pH 9.5. In comparison, rat arginase I exhibits a KM value of 1.4 mM at pH 9.0 .

3-Although Arginine is present in many tissues, urea synthesis occurs in the liver only, why?
Because Arginase is mostly found in Liver, while the rest of enzyme(four) of urea cycle are also present in another tissue m for this reason Arginine synthesis may occur to varying degree in many tissues .But only Liver can ultimately produce urea.

2- What was the purpose of the manganese sulfate, and perchloric acid used in this experiment?
In the applied protocol, these chemical wasn't used.

3- Deficiency in the liver Arginase activity leads to low levels of serum urea, but low levels of serum urea is not necessary associated with low arginase activity, explain.
Because low levels of serum urea could be due to other than ARG 1 deficiency, it could be due to renal diseas.

4- What is the most important clinical complication of Arginase deficiency?
Toxicity of CNS, lethargy, liver failure, coma.

Experiment (10):

1-Why are time, temperature and pH important in the initial stagesof the isolation of glycogen, but not in the latter stages?

To prevent the hydrolysis of glycogen in the sample.

2-What would be the effect of substituting β-amylase for α-amylase?
Less amount of reducing sugar.