Clinical Biochemistry

Assignment-3

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BT-C

(1) Importance of clinical Biochemistry.

It is used in clinical diagnosis, manufacture of various biological products, treatment of diseases.

clinical Biochemistry has revealed the abnormalities in their metabolism and their relationship to various diseases.

(2) List the clinical features of Lesch Nyhan Syndrome.

- Neurological symptoms:

Severe neurologic disease, characterized by self multilating behaviors such as lip and finger biting or head banging.

- Gout
- Involuntary muscle movement.

(3) Write a note on Cori's disease

It is a Type III Glycogen storage disease caused due to Glycogen debranching enzyme. Deposition of abnormal glycogen storage. It affects the Liver and muscle.

Clinical features.

About 15% affect liver only. Hypoglycaemia, poor growth, hepatomegaly, moderate progressive myopathy

Symptoms can regress with age.

Treatment is protein supplements for muscle disorder.

(4) Write the vision related clinical conditions in Albinism.

In Ocular albinism only the eyes lack pigment.

Rapid involuntary back and forth movement of the eyes.

Extreme near sightedness or farsightedness

Sensitivity to light.

(5) State the recent methodological issues in clinical biochemistry.

Most recent methodological issues in clinical biochemistry are all associated with high volume testing: laboratory automation and workflow management, and computational issues.

(6) List the clinical features of Nieman Pick disease.

- Type A usually begins in the first few months of life.

- Abdominal swelling within 3-6 months.
- cherry red spot in the eye
- Feeding difficulties

- Type B symptoms are usually milder.
- They occur in late childhood or the teenage years
- Abdominal swelling may occur in young children.

- Type C affects school aged children.

- Enlarged spleen
- Enlarged liver.
- Learning difficulties

(7) Give the name and symptoms of the condition where galactose accumulates in blood.

Galactosemia is the condition where galactose accumulates in blood

symptoms :

- Intellectual disability
- Hepatomegaly
- Hepatic failure
- Renal failure.

(8) What is the cause of Hers Disease ?

It is caused due to increased deposits of normal glycogen in liver or in red or white blood cells.

liver phosphorylase enzyme is affected.

(9) Write a note on Fructosuria ?

Fructosuria is a autosomal recessive genetic disorder arises as a result of deficiency of the hepatic enzyme fructokinase. Essential fructosuria is characterized by the presence of fructose in the urine after ingesting fructose. This disorder is mild and it probably remains undiagosed in many people.

(10) Write a note on Addison's disease

Addison's disease occurs when your body doesn't produce enough of certain hormones. Adrenal gland produce little cortisol and aldosterone

Symptoms.

- Steadily worsening fatigue.
- Abdominal pain
- Nausea and vomiting.
- Diarrhea.

(11) Name the hormone, popularly called as stress hormone?

Cortisol is hormone released by the adrenal gland in response to stress or lowered levels of blood glucocorticoids and is involved in stimulating glucose synthesis and anti stress and anti inflammatory processes

(12) Write the role of aldosterone in mineral metabolism?

Aldosterone regulates the salt and water balance of the body by increasing the retention of Na^+ and water and the excretion of potassium by the kidneys It also has a limited effect on the metabolism of fats, carbohydrates and proteins.

(13) Give the pathway for the conversion of testosterone to Estrogen hormone.

Testosterone

↓ Aromatase

↓ NADPH

β-Estradiol (estrogen hormone)

Testosterone is converted to β Estradiol by a microsomal enzyme aromatase which brings about aromatisation of ring A. It brings about 3 successive hydroxylations of testosterone and the final hydroxylated product loses C_{19} non enzymatically and converted to β estradiol.

(14) What are the outcomes of hyperthyroidism?

Hyperthyroidism in adults is associated with excessive heat generation, metabolic wasting, cardiac dysfunction (tachycardia), tremors and anxiety.

(15) Recall the hormone responsible for breast cancer.

Estrogen and Progesterone are the hormones responsible for breast cancer. Being exposed to long time of these hormones are responsible for breast cancer.

(16) Name the hormone, popularly called as stress hormone.

Cortisol. is called stress hormone.

(17) Write the role of aldosterone in mineral metabolism.

Ib regulates the Na^+ and water and excretes K^+ by the kidneys.

(18) Importance of thyroid hormone.

Thyroid hormone helps with brain development and function.

Regulates the metabolic rate of the body.

Muscle control and Bone health.

Protein synthesis.

) Types of Goiter.

Colloid goiter

Nontoxic goiter

Toxic goiter.

(21) Define monoamine hormones.

The monoamine hormones are hormones derived from aromatic amino acids such as phenylaline, tyrosine and tryptophan and are involved in neurotransmission.

eg. Catecholamines. Adrenaline. Dopamine.

(22) List out the naturally occuring Estrogens in Human.

Estrone (E_1)

Estradiol (E_2)

Estriol (E_3)

Estetrol (E_4) are the 4 naturally occuring estrogens in women.