



CARBOHYDRATE METABOLISM AND ITS GENETIC DISORDERS

Scientific leader : **Khalimova Salomat Asrorovna**
205 pediatrics student: **Ashirova Naila Abror kizi**
Samarkand State Medical University, Department of
Biological Chemistry

Abstract: *This article carbohydrate exchange processes and his/her pedigree violations to study Carbohydrates in the body energy source as used and this processes genetic management under done increases . Carbohydrate exchange violation pedigree genetic mutations , metabolic diseases and other clinical situations with is related . Article carbohydrate exchange of disorders genetic and clinical aspects , as well as this to disruptions against new research and treatment methods seeing comes out . Home Attention, sugar. diabetes , metabolic syndrome , galactosemia diseases with related to disruptions focused .*

Keywords: *Carbohydrate metabolism, hereditary disorders, genetic defects, diabetes mellitus, galactosemia, drug-induced polyuria, aldose reductase, hexokinase, energy metabolism.*

Carbohydrate exchange - this in the body energy sources one to be , to be correct processing to be given vital importance These processes metabolism different in stages , that including glucose , galactose and other monosaccharides such as simple carbohydrates with related fermentative in reactions manifestation This will be of exchange pedigree disorders different kind metabolic to diseases take arrival It is also possible to breed factors and genetic mutations this violations brought release it is possible , this and clinical in practice important importance has . In the article carbohydrate exchange main roads and pedigree of violations impact in detail analysis will be done .

Carbohydrates in the body energy source as main place The most common carbohydrate is glucose. Glucose is delivered to all organs through the blood. The brain is very sensitive to a decrease in glucose concentration in the body, because

the main energy source for the brain is glucose. It has also been proven that the brain's daily glucose requirement is 120 g.

Carbohydrate metabolism consists of several processes:

- *Glycolysis* : The conversion of glucose to pyruvate and lactate under aerobic and anaerobic conditions. ATP and NADH are produced.
- *Glycogenogenesis* : The synthesis of glycogen from glucose, a form of reserve energy.
- *Glycogenolysis* : The breakdown of glycogen into glucose-6-phosphate.
- *Gluconeogenesis* : The resynthesis of glucose from fatty acids, amino acids, or lactate.
- *Pentose phosphate pathway*: Glucose metabolism for nucleotide and NADPH synthesis.
- *Krebs cycle*: The breakdown of pyruvate via oxaloacetate and acetyl-CoA to carbon dioxide and water. A large amount of ATP is produced in this process.

All of this is controlled with the help of various enzymes and coenzymes.

Genetic Disorders: Genes that control carbohydrate metabolism and their mutations can lead to the development of metabolic diseases in the body. Genetic disorders are mainly caused by genetic mutations. These mutations can disrupt metabolic processes and make it difficult for the body's energy production, storage and distribution systems. Diabetes and Galactosemia Metabolic diseases such as diabetes are caused by such genetic disorders.

Carbohydrate exchange violation different kind clinical situations and to diseases take arrival possible . Most wide widespread from disruptions one this is sweet Diabetes is a disease that affects the heart . organism to insulin relatively resistance or insulin production to release stop as a result to the surface coming state . Biochemical point of view from the perspective of , sugary diabetes main the causes are enzyme or hormones deficit with related . Sugary diabetes main from the reasons

one of the hormones insulin lack or organism insulin effective work out Insulin deficiency two main kind of in diabetes observed :

1st type Diabetes : This type in diabetes stomach under beta cells of the pancreas damage see , produce insulin It is not released . It is autoimmune disease as a result to the surface is coming .

Type 2 diabetes : In this type, although insulin production is normal, the cells are unable to use insulin effectively, which is called insulin resistance. Glucagon hormone deficiency or imbalance : Glucagon is a hormone that works in opposition to insulin and is produced by the alpha cells of the pancreas. It stimulates the liver to release glucose to increase blood sugar levels. If glucagon is not produced properly, this can lead to erratic changes in blood sugar levels.

Amylase enzyme: Some cases of diabetes may be affected by an imbalance of enzymes involved in the digestive process, including amylase. Amylase primarily breaks down starches and other carbohydrates into simple sugars such as glucose and maltose. If these enzymes are not working properly, carbohydrates may not be fully broken down, which can lead to high blood sugar levels.

Diabetes clinical signs :

1. Polyuria – high amount of sugar level kidneys through more urine to be released take is coming .
2. Many fluid intake – sugar amount increase because of organism dehydration feeling does and as a result drink desire increases .
3. Many food – in the body energy shortage because of , man permanent accordingly hunger feels and many eats .
4. In health changes – fatigue , fast fatigue , vision of ability decline , wounds slowly completion or liver and in the heart problems increases .
5. Skin to dry and itching – in diabetes skin dry goes and itching possible .
6. Weight gain and loss – some in patients weight increase , in others and fast weight loss is observed .

7. View problems – retinopathy (vision of nerves due to injury) see ability gets worse .

Diagnostics :

1. Measuring blood glucose concentration – in the blood sugar level measurement main diagnostic method is . The following tests with inspections done increased :
 - Fasting blood glucose test – fasting blood sugar level .
 - Oral Glucose Tolerance Test - sugar acceptance since then blood sugar how change to determine .
 - HbA1c test – for the last 2-3 months blood sugar average level to determine .
2. This of tests results as follows interpretation is done :
 - Normal : Fasting blood glucose < 100 mg/ dL , HbA1c < 5.7%
 - Pre- diabetes : Fasting blood glucose 100-125 mg/ dL , HbA1c 5.7-6.4%
 - Diabetes : Fasting blood glucose $\geq$ 126 mg/ dL , HbA1c $\geq$ 6.5%

Treatment :

Diabetes treatment one how many directions own inside takes :

1. With a diet nutrition – diabetes management for correct food very important . Carbohydrates limit , fats and proteins suitable accordingly choose , also , fruit and vegetables with enriched on a diet action to do necessary .
2. Physical activity – regular physical Exercise (running , walking , swimming , cycling) walk to make (blood) sugar control to do help gives .
3. Medication Treatment – insulin therapy in type 1 diabetes need will be , in type 2 diabetes and mouth through acceptance to be done medicines and Sometimes insulin is used . Medications mainly insulin sensitivity increase or insulin production to release to multiply help gives .
4. Blood sugar following go – blood sugar regular accordingly measured stand necessary , this the situation permanent accordingly control to do help gives .

5. Complications prevent to get – diabetes many serious to complications take arrival possible , for example , heart diseases , kidney deficiency , nervousness system problems and of sight Therefore , diabetes permanent accordingly control to do important .

Galactosemia is galactose a ni in metabolism correct I will break it down. not getting because of developing genetic Galactosemia is a disease . autosomali recessive disease become will pass , that is the disease develop for both mutation from both parents to take Galactosemia usually of galactose galactose-1-phosphate uridyltransferase (GALT) enzyme mutation of the gene responsible for the synthesis of because of to the surface Galactose-1-phosphate uridyltransferase (GALT) The enzyme galactose-1-phosphate is an enzyme that converts galactose-1-phosphate to galactose-6-phosphate , which is a precursor to galactose. metabolism control If the GALT enzyme shortage or his/her function broken if , galactose and his/her metabolic products (galactose-1-phosphate) in the body gathered , various kind to injuries take arrival There are 3 main types of galactosemia :

- 1- Classical galactosemia is caused by a deficiency of galactose 1-phosphate uridyl transferase. As a result, galactose 1-phosphate loses its ability to be converted to glucose 1-phosphate, and the level of galactose 1-phosphate in the blood increases. Galactose 1-phosphate is hepatotoxic: it accumulates in the liver, leading to cirrhosis and hepatomegaly. In addition, liver hepatocytes rupture, causing hyperbilirubinemia and, as a result, yellowing of the skin .

If galactose 1-phosphate accumulates in large quantities, vomiting and diarrhea occur .

- 2- Galactosemia is caused by galactokinase deficiency. Now the amount of galactose in the blood increases. As a result, cataracts and cataract swelling are observed. This is because the cataract contains the enzyme aldose reductase, and high levels of galactose in the blood enter the cataract and are converted to galactitol by the action of aldose reductase. Galactitol is osmotically active, drawing water

from the periphery and causing the cataract to swell, and cataracts occur due to the toxicity of galactitol.

Type 3: Galactosemia is rare and has symptoms similar to type 1 galactosemia. Due to UDP galactose kinase 4-epimerase deficiency, the amount of galactose 1-phosphate in the blood is increased .

Treatment: Since this disease mainly occurs in infants, eliminate milk and dairy products from the diet and add lactose-free dairy products.

Treatment methods for hereditary disorders and metabolic diseases:

Genetic testing – helps to identify the presence of mutations, understand the types of disease and the level of risk. If a patient has a hereditary disorder, it is possible to advise descendants about the risk of passing on the disease.

Genetic counseling - providing accurate information to family members about hereditary diseases and creating an understanding of the causes of the disease.

Diet therapy – In diseases that disrupt carbohydrate metabolism, the most effective treatment is nutrient management, particularly carbohydrate management.

diabetes , it is necessary to follow a special diet to limit sugar in food and manage insulin.

galactosemia , galactose and sources of galactose (milk and dairy products) must be completely avoided.

Excessive consumption of milk – In diseases such as galactosemia, there is a problem with metabolizing milk, so special milk-like plant products such as soy are used .

Insulin therapy – Since there is a lack of insulin in type 1 diabetes, it is necessary to receive insulin from an external source .

altered enzymes – galactosemia and fructosemia, enzyme therapy can be performed in some cases, but this is not currently widely used .

Medications – Oral medications or insulin sensitizers are used to control blood sugar in type 2 diabetes .

Increase physical activity – regular exercise, such as walking, swimming, cycling or fitness, helps improve insulin effectiveness, which is important for managing type 2 diabetes .

Maintaining biorhythms and a healthy lifestyle – reducing stress, quitting bad habits (smoking, alcohol), and ensuring regular sleep improves overall health .

Monitoring liver and kidney function – diseases associated with genetic disorders can cause liver and kidney damage. Therefore for this of organs activity regular accordingly checking stand Galactosemia or other metabolic diseases treatment during this organs over permanent control demand is being done .

Conclusion . Inherited metabolic diseases, including diabetes mellitus, galactosemia, and other disorders affecting carbohydrate metabolism, are caused by a disruption in the body's metabolic processes. There are several approaches to treating these diseases: genetic counseling, diet, medication, physical activity, and regular medical monitoring. Early diagnosis, genetic counseling, diet, and, if necessary, medication can help slow the progression of the disease and improve quality of life.

REFERENCES

1. Kahn, SE, & Weir, GC (2011). "Understanding Insulin Resistance and Its Role in Diabetes." *Diabetes Care* , 34(2), 324–331.
2. Galactosemia : Overview and Diagnosis. (2022). National Institute of Child Health and Human Development. Retrieved from <https://www.nichd.nih.gov/>
3. American Diabetes Association. (2020). "Diagnosis and Classification of Diabetes Mellitus." *Diabetes Care* , 43(Supplement 1), S14-S31.
4. Tanzi , RE, & Housman, DE (1992). "Genetic Defects in Human Disease." *Science* , 258(5079), 1385-1393.
5. Voss, P., & Kelly, M. (2021). "Dietary Management of Inherited Metabolic Disorders." *Nutrition in Clinical Practice* , 36(2), 389-397.
6. Baikulov A. K. , Murtazaeva N. K. , Toshbayev F . N . DYNAMICS INFLUENCE LACTATE DEHYDROGENASE PRI EXPERIMENTAL NOM

INFRACRECT MYOCARD //World of Scientific news in Science. - 2024. - T. 2. – no. 3. - S. 244-251.

7. Baikulov A. K., Ubaidullaeva G. B., Eshburieva B. R. Correction of experimental hyperlipoproteinemia with proizvodnymi chitozana // World of Scientific news in Science . - 2024. - T. 2. – no. 2. - S. 937-947.

8. Baykulov AK, Halimova SA, Eshburieva NR Dynamics of the influence of lactate dehydrogenase during experimental myocardial infarction // World of Scientific news in Science. - 2024. - T. 2. – no. 3. - S. 232-238.

9. Kenjayevich BA et al. EXPERIMENTAL INDUCTION OF HYPERHOMOCYSTEINEMIA UNDER OXIDATIVE STRESS //RESEARCH. UZ. – 2024. – T. 40. – No. 1. – P. 25-30.

10. Baykulov AK, Khurramova SG, Ubaidullayeva GB STUDYING OXIDATIVE STRESS OF LIPIDS DURING EXPERIMENTAL MYOCARDIAL INFARCTION IN RATS //Bulletin news in New Science Society International Scientific Journal. - 2024. - T. 1. – no. 5. - S. 181-191.

11. Baykulov AK, Toshboyev FN, Akhmadov JZ BIOCHEMICAL AND PHYSIOLOGICAL CHANGES IN PARASITE PARAMETERS IN THE HOST-PARASITE RELATIONSHIP //Modern Scientific Research International Scientific Journal. - 2024. - T. 2. – no. 1. – S. 257-262.

12. Baykulov AK et al. CORRECTION OF EXPERIMENTAL HYPERLIPOPROTEINEMIA WITH CHITOSAN PRODUCTS // Modern Science and education news international scientific magazine . - 2024. - T. 2. – no. 2. - S. 230-240.

13. Baykulov AK et al. CORRECTION OF EXPERIMENTAL HYPERLIPOPROTEINEMIA WITH CHITOSAN PRODUCTS //International scientific journal of modern science and education news. – 2024. – T. 2. – No. 2. – P. 230-240.