ETHIOPIAN INTERNATIONAL JOURNAL OF MULTIDISCIPLINARY RESEARCH

eISSN: 2349-5715 pISSN: 2349-5707

Volume: 12, Issue 01, January-2025 SJIF 2019: 4.702 2020: 4.737 2021: 5.071 2022: 4.919 2023: 6.980 2024: 7,662

Mamatyaqubova Malohat Sharofjon qizi

Andijan State medical Institute

Assistant Department of medical biology and histalogy

ETIOLOGY,PATHOGENESIS AND BIOCHEMISTRY,DIAGNOSIS,TREATMENT METHODS OF PHENYLKETANURIA DISEASE

Annotation: This article presents several facts about the causes of Origin,diagnosis,biochemistry,treatment methods of Phenylketanuria, which is considered a chromosomal disease.Phenylketanuria is a recessive chromosome disease.The conversion of phenylalanine to tyrosine is caused when disturbed.Treatment for patients with phenylketonuria is continuous and consists of following a strict diet from the moment of diagnosis. Completely eliminating phenylalanine from the diet and compensating for amino acid deficiency with the help of special dietary supplements allows you to maintain normal metabolism in the body. The article presents the causes, consequences, types, methods of treatment, physical therapy; diet for phenylketonuria.

Keywords: Phenylalanine,diet,pathology,tyrosine,phenylketanuria,intelligence,reflex,brain,genotype, mutant,gene,hypertrophy,,therapy, enzyme.

Annotatsiya: Ushbu maqolada xromosoma kasalligi hisoblangan Fenilketanuriyaning kelib chiqish sabablari,diagnozi,biokimyosi,davolash usullari haqida bir qancha ma’lumotlar keltirilgan.Fenilketanuriya retsissiv xromosoma kasalligi hisoblanadi.Fenilalaninning tirozinga aylanishi buzilganda kelib chiqadi.Fenilketonuriya bilan og'rigan bemorlarni davolash uzluksiz bo'lib, tashxis qo'yilgan paytdan boshlab qat'iy dietaga rioya qilishdan iborat. Fenilalaninni dietadan to'liq chiqarib tashlash va aminokislotalar yetishmovchiligini maxsus xun takviyeleri yordamida qoplash tanadagi normal metabolizmni saqlashga imkon beradi. Maqolada kasalikning sabablari, oqibatlari, turlari, davolash usullari, jismoniy terapiya; fenilketonuriya uchun dieta keltirilgan.

Kalitso’zlar: Fenilalanin,dieta,patologiya,tirozin,fenilketanuriya,aql,miya,refleks,genotip,mutant,gen, gipertofiya,terapiya,ferment.

Аннотация:В этой статье представлена некоторая информация о причинах,диагностике,биохимии,лечении фенилкетанурии, которая считается хромосомным заболеванием.Фенилкетанурия считается рецессивным хромосомным заболеванием.Возникает при нарушении превращения фенилаланина в тирозин.Лечение больных фенилкетонурией является непрерывным и заключается в соблюдении строгой диеты с момента постановки диагноза. Полное исключение фенилаланина из рациона и восполнение дефицита аминокислот с помощью специальных пищевых добавок позволяет поддерживать нормальный обмен веществ в организме. В статье представлены причины, последствия, виды, методы лечения заболевания, лечебная физкультура; диета при фенилкетонурии.

Ключевые:Слова:фенилаланин,диета,патология,тирозин,фенилкетанурия,интеллект,мозг, генотип,мутант,ген,гипертония,рефлекс,терапия,фермент.

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Phenylketanuria biochemistry:

Phenylketonuria (phenylpyruvic oligophrenia, Felling's disease) is a congenital metabolic pathology that leads to a violation of the reactions of the conversion of essential amino acids phenylalanine to tyrosine. An increase in phenylalanine and its metabolic products in the blood leads to damage to the central nervous system, which is manifested by mental disorders and nervous defects.

There is a 2xil reason for the origin of phenylketanuria.They include the following causes, with a defect in the enzyme phenylalanine hydroxylase and with a defect in the substance tetrahydrobiopterin, which is considered its coferment.Phenylketanuria is divided into 2xyl species.Classical phenylketanuria and variant phenylketanuria.

Classical phenylketanuria is caused by a defect in the enzyme phenylalanine hydroxylase.Phenylalanine becomes non-tyrosine and increases in the body.This condition prevents the brain from passing other amino acids such as tryptophan amino acid.As a result,the synthesis of dopamine,adrenaline, norepinephrine and methionine decreases.Newborns experience cases of microcephaly,lagging behind mental and physical development.The frequency of this pathology among newborns is small. This figure does not exceed 1: 7000. Therefore, this disease is classified as an orphan disease - rare congenital or acquired nosologies, which are characterized by a chronic course of a pathological process with a tendency to develop, degenerative changes in organs and early disability of the child.

Causes of phenylketonuria:

Phenylketonuria is an autosomal recessive hereditary disease caused by a mutation in the gene responsible for phenylalanine metabolism. According to the Basic Rules of inheritance, this disease can occur in a child from a phenotypically healthy parent. In the study of the genetic material of such people, a recessive gene is found, the expression of which is suppressed by the dominant allele. The genotype of the carrier parents is represented by "Aa", where "a" is the dominant allele and "a" is the recessive allele. If a recessive gene enters both gametes, A "aa" genotype zygote is formed. This combination is very important for the unborn child and becomes a disease of "phenylketonuria". Pathology manifests itself equally in female and male children.

Pathology develops only when there are two recessive genes in the chromosome set of somatic cells in a child. If both healthy parents are carriers of the mutant gene, then the probability of having a child with this enzymopathy is the same as the probability of having a completely healthy child-25%. In 50% of cases, a healthy carrier of the recessive phenylketonuria gene is born.

Genetic causes of the disease contribute to impaired amino acid metabolism. Phenylalanine is an important amino acid that enters the child's body with food. Using phenylalanine 4-hydroxylase, this amino acid is converted to tyrosine, which is then used by the body for protein synthesis. As a result of the absence of this enzyme, phenylalanine undergoes a pathological transformation, which leads to an increase in the blood level of phenylalanine and abnormal amino acid exchange products, which negatively affects lipid metabolism in the brain, causing mental retardation. Due to the lack of tyrosine, the synthesis of neurotransmitters that regulate the functioning of the nervous system is disrupted.

Types of phenylketonuria:

There are three types of the disease:

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Type 1 phenylketonuria is characterized by a mutation in the gene responsible for the synthesis of the (classical) - phenylalanine-4-hydroxylase enzyme. This type of disease accounts for 98% of all diagnosed enzymatic cases.

Type 2 phenylketonuria (atypical) occurs when chromosome 4, which is responsible for the synthesis of dihydropyridine reductase, breaks down.

Develops with a deficiency of Type 3 (atypical) - tetrahydrobiopterin.

Depending on the level of phenylalanine in the blood, three forms of this disease are conditionally distinguished: mild; moderate; severe.

Symptoms of phenylketonuria:

The first signs of this disease appear after starting breastfeeding. The enzyme system of a sick child cannot withstand the processing of phenylalanine, which enters the child's body with breast milk. The increase in the level of this amino acid and the products of its pathological metabolism leads to a delay in the psychomotor development of the child: the newborn is inactive, slowly affects color and sound stimuli. Objective signs of phenylketonuria include:

- muscle hypertension and hyperreflexia (the child sits with his legs under him, when walking, his legs bend at the knees and thigh joints);

- skin rash (papules, vesicles), redness of the skin;

-other skin symptoms: dermographism, dermatitis, eczema;

- slow attitude to the environment;

- swallowing disorder;

- spontaneous convulsions;

- the smell of" Mouse " coming from the skin and urine;

- decreased intelligence.

Most children with this condition have white hair and blue eyes (signs of albinism). If you do not follow a diet, the clinical picture of fermentopathy increases and can lead to mental retardation or stupidity.

Diagnosis of phenylketonuria:

Determining the level of phenylalanine in the blood is included in the screening program for newborns, which provides an early diagnosis of pathology. According to the results of the screening test, the child is prescribed an additional blood test using colorimetry or sent to a specialized medical institution where genetic analysis is carried out. At a meeting with a geneticist, a family tree is formed in which it is possible to determine the transmission of a recessive mutant gene.

Methods for determining phenylketonuria:

1.Harty test-blood test for phenylalanine levels;

2.Felling test-checking urine by adding acetic acid to it;

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3.colorimetric methods;

4.DNA Diagnostics-identification of a mutant gene.

5.For differential diagnosis purposes, EEG and MRI are performed.

Methods of treatment of phenylketonuria

Treatment of children with this pathology involves limiting the consumption of foods containing phenylalanine. Compliance with the recommended diet ensures the normal intellectual development of the child and prevents the development of the disease. Diet for phenylketonuria includes: restriction of natural proteins (breast milk, meat, fish, dairy products); replenishment of amino acids, energy, vitamin deficiency with the help of therapeutic nutrition.

The introduction of complementary foods for children with phenylketonuria begins with a few drops of juice (apple, pear). At 6 months, you can give vegetable puree. The second additional food is low-protein porridge.

Consequences of phenylketonuria:

Complications of this disease are associated with damage to the central nervous system and the development of mental neurological diseases:

mental retardation (mental retardation, insanity;

a neurological defect that manifests as irreversible dystrophic changes (convulsions).

So, when checking the diagnosis early and prescribing a diet, the prognosis is conditionally favorable (the disease cannot be completely cured, but the toxic effects of phenylalanine and its derivatives can be avoided).

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