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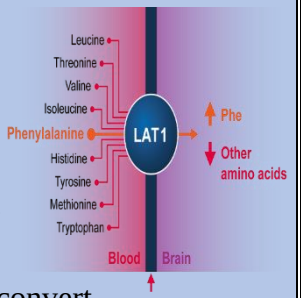
Phenylketonuria (PKU)

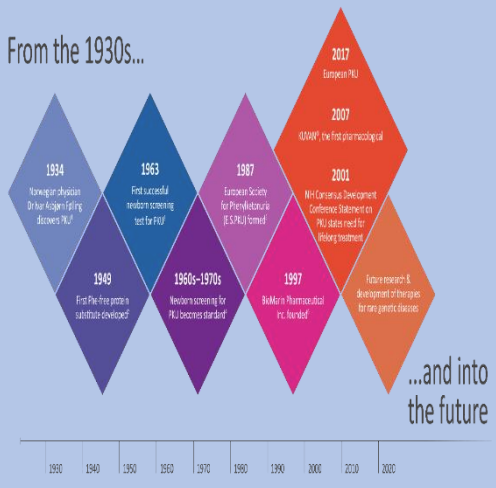
- Phenylketonuria (PKU) is a rare genetic condition that causes the accumulation of an amino acid called phenylalanine (Phe) in the body.
- Amino acids are compounds of protein bulding.
- Phenylalanine is found in all proteins and some artificial sweeteners.



- Phenylalanine Hydroxylase (PAH) is an enzyme used by the body to convert Phenylalanine into tyrosine, which your body needs to create neurotransmitters such as epinephrine, norepinephrine and dopamine.
- PKU is caused by a defect in the gene that helps to create Phenylalanine Hydroxylase (PAH).
- When this enzyme is missing, your body can't break phenylalanine, which causes the accumulation of phenylalanine in the body.

Incidence of the disease

continent	Cases per live birth
Africa	1:3,000
Europe	1:4,000–1:100,000
Middle East	1:6,250
North America	19,000–1:13,500–1:19,000
Latin America	1:25,000–1:50,000
Asia	7,11,14:327,000 1:11,500–1:327,000
Australia	1:10,000



References: 1. The united states of The United States of The government's policy of "supporting the government' in the country is to ensure that the government's policy is to ensure that the government's work is to ensure that the government's work is to ensure that the government's work is conduct Bremen: UNI-MED, 2016. 2. Alonso-Fernández JR, Cólon C. *J Med Screen* 2009;16(4):205–211. 3. Guthrie R, Susi A. *Pediatrics*

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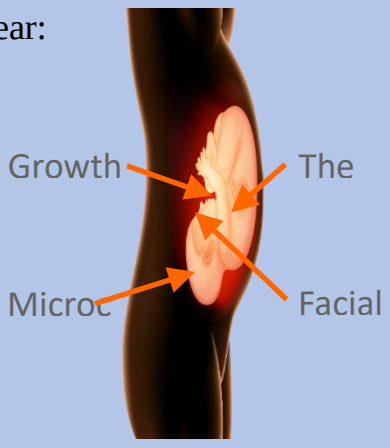
Causes of Phenylketonuria disease

- PKU is a rare genetic disease caused by a defect in the PAH gene.
- The PAH gene helps to form the Phenylalanine Hydroxylase (PAH) enzyme which is responsible for cracking the amino acid phenyl alanine.
- A dangerous build-up of phenylalanine can occur when someone eats protein-rich foods, such as eggs and meat.
- The disease is passed on to children when both parents are transfering an infected version of the PAH gene for their child.

Symptoms of Phenylketonuria disease

- PKU symptoms can range from mild to severe.
- The most intense form of this disturbance is known as the classic PKU.
- An infant with classic PKU may be seen Normal in the first few months of life, However, if the infected child has not been diagnosed and treated during the early period, the following symptoms will start to appear:
- Cramps.
- Tremors or hands spasm.
- Growth development delayed.
- Hyperactivity.
- Eczema.
- An unpleasant smell of breath, skin and urine.



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What happens if PKU is not diagnosed at birth and rapid treatment doesn't begin:

- Permanent brain damage and intellectual disability during the first few months of life.
- Behavioral problems for older children.
- Once a specific diet and the necessary treatments (whether the pills or subcutaneous injections, depending on the condition and age of the patient) the symptoms will begin to decrease *(ask your doctor about the available treatments).*
- In general, there are no abnormal symptoms in people with PKU who manage their diet and receive treatment correctly.

Challenges Facing people with phenylketonuria

- The long-term outcomes for people with PKU who has followed a diagnostic and therapeutic plan (whether tablets, powder or subcutaneous injections depending on the condition and age of the patient and the shortly starting after birth) are very good (ask your therapist about the treatments available).
- When diagnosis and treatment are delayed, un reversible brain damage can occur and this can in turn lead to incurable mental disabilities.
- By the first year of a child's life, untreated PKU can lead to developmental delays, behavioral and emotional problems, including various neurological problems.

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Does Phenyl ketonuria disease has an effect on the mental function of adults?

- Adolescent and adult patients who do not adhere to treatment are at risk of suffering due to low levels of mental ability.
- Patients who are not committed to treatment complain of increased irritability, lack of concentration, which in turn affects both their social and professional lives.
- Some patients suffer from anxiety and depression or in severe cases the patient of phenylketonuria may suffer from paranoia and schizophrenia, although it has been found that all these symptoms improved when the treatment is resumed.

What is the burden of phenyl ketone urea on the daily life of adult?

Adults suffer from the same burdens as children:

- The diet overlaps with their job's requirements.
- Socially, patients tend to live longer at their parents' home and seem to have more difficulty finding a life partner.
- The difficulty of following the diet without parental supervision.
- Congenital and general malformations of the child if the mother is ill and is not committed to treatment.

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Treatment options

People with PKU can relieve their symptoms and prevent complications by following a diet and by taking medications (tablets, powder or subcutaneous injections depending on the patient's condition and the age of the patient) ***(Ask your doctor about available treatments).***

1.Diet:

Boiled egg
~600 mg Phe/100 g

Apples
~2 mg Phe/100 g

- It is a special diet that limits foods containing phenylalanine where infants with PKU can be fed breast milk.
- When your baby is old enough to eat solid foods, you should avoid protein-rich foods including **(eggs, cheese, nuts, milk, beans, chicken, beef and fish).**
- To make sure they still get enough protein, children with PKU need special milk that contains all the amino acids the body needs except phenyl alanine.
- PKU patients will have to follow these dietary restrictions and follow phenylalanine free diet throughout their whole lives to control the complications of the disease.
- PKU patients need to work closely with a doctor or dietitian to maintain the proper balance of nutrients while reducing the intake of phenylalanine.
- Diet is not the ideal way to control the disease.

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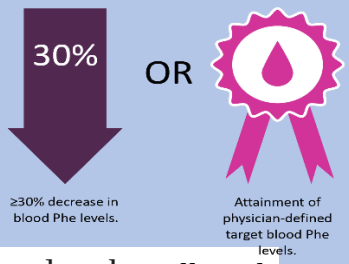
2. Pharmacotherapy



Tablets or powder:

- U.S. Food and Drug Administration (FDA) recently approved pku pills and powder to help lower levels of phenylalanine.
- This drug should be used in combination with pku's own meal plan; however, it does not work with all people with PKU.
- It is considered most effective in children with mild cases of PKU

(Ask your treating doctor if this treatment is right for you).

Subcutaneous injections:

- The U.S. Food and Drug Administration (FDA) and the European Union (EMA) recently approved subcutaneous injection for pku patients over 16 years of age.
- These injections are used to lower levels of phenylalanine in blood in adults who have irregular blood levels above 600 µmol/L (10 mg/dL) on their current treatment.

(Ask your therapist if this treatment is right for you, and you should discuss the potential benefits and risks with your healthcare provider).

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