PHENYLKETONURIA (PKU)

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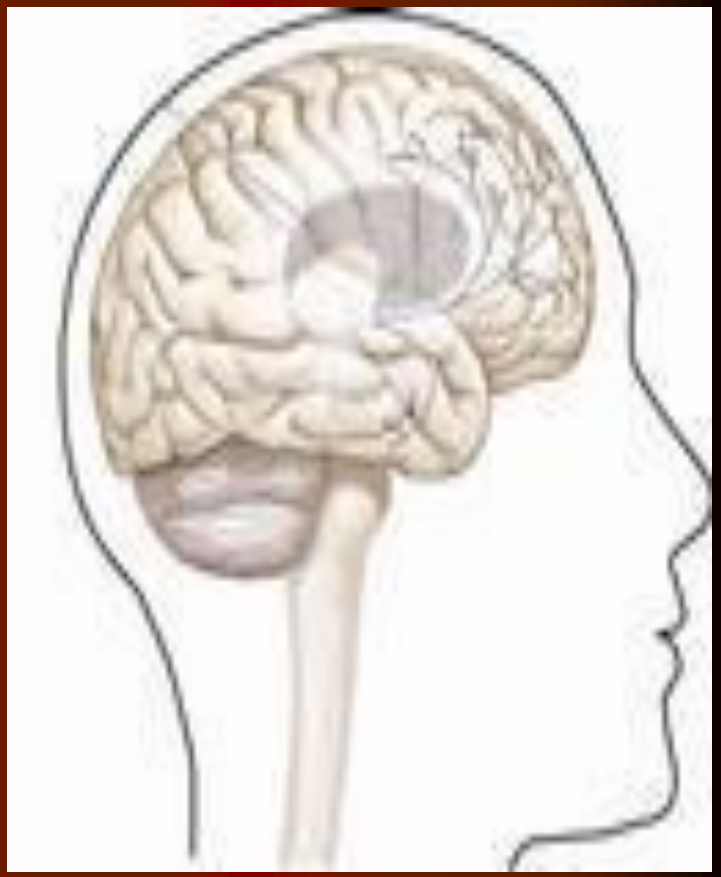
(PKU)

- What is (PKU)?
- Who does it affect?
- What are the origins of PKU?
- What are the treatments for PKU?
- What are the precautions of PKU?

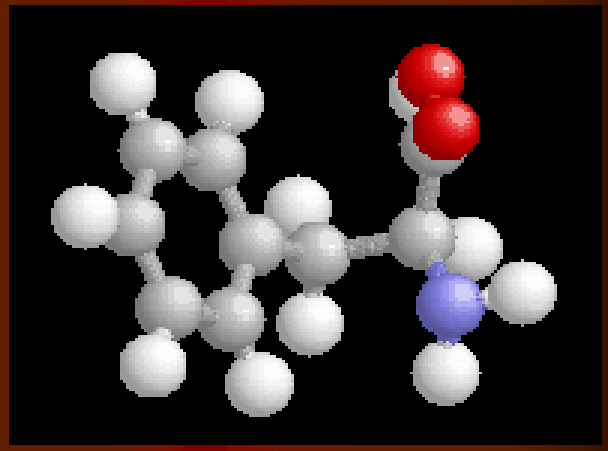
WHAT IS PKU?

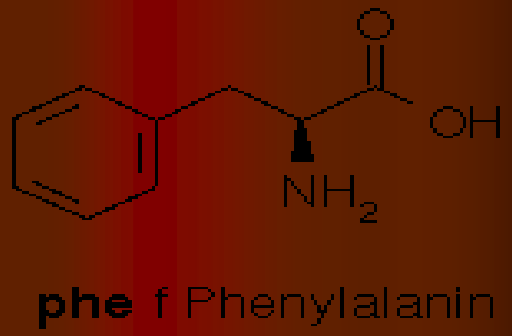
PKU, in its "classic" form, is a rare, inherited metabolic disease that results in mental retardation and other neurological problems when treatment is not started within the first few weeks of life. .



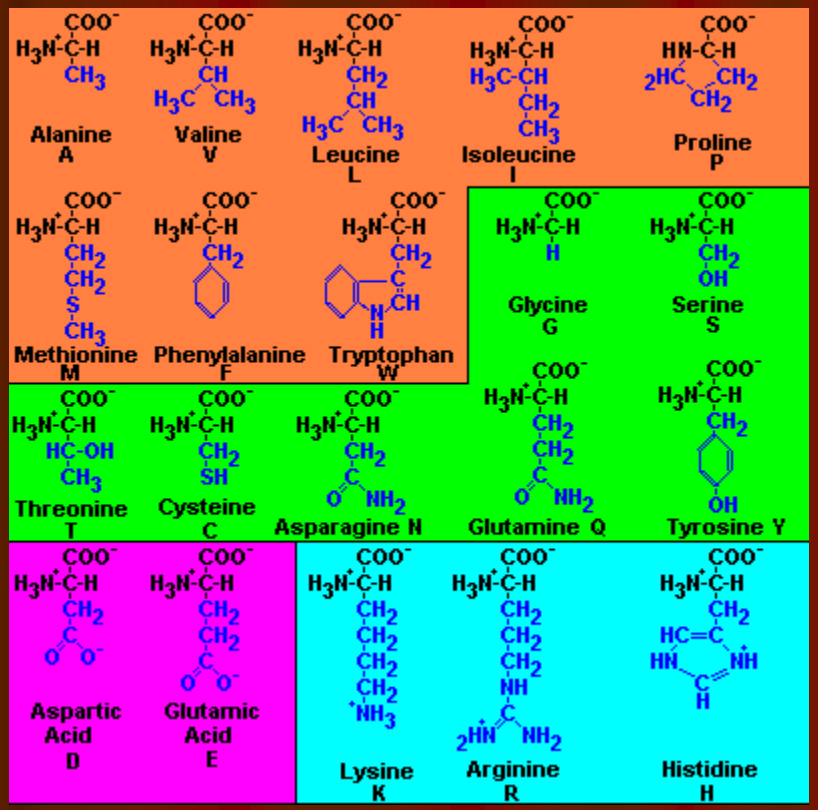
PHENYLALANINE

PKU is characterized by the inability of the body to utilize the essential amino acid (phenylalanine).





WHATS THE DIFFERENCE BETWEEN ESSENTIAL AND NON-ESSENTIAL AMINO ACIDS?



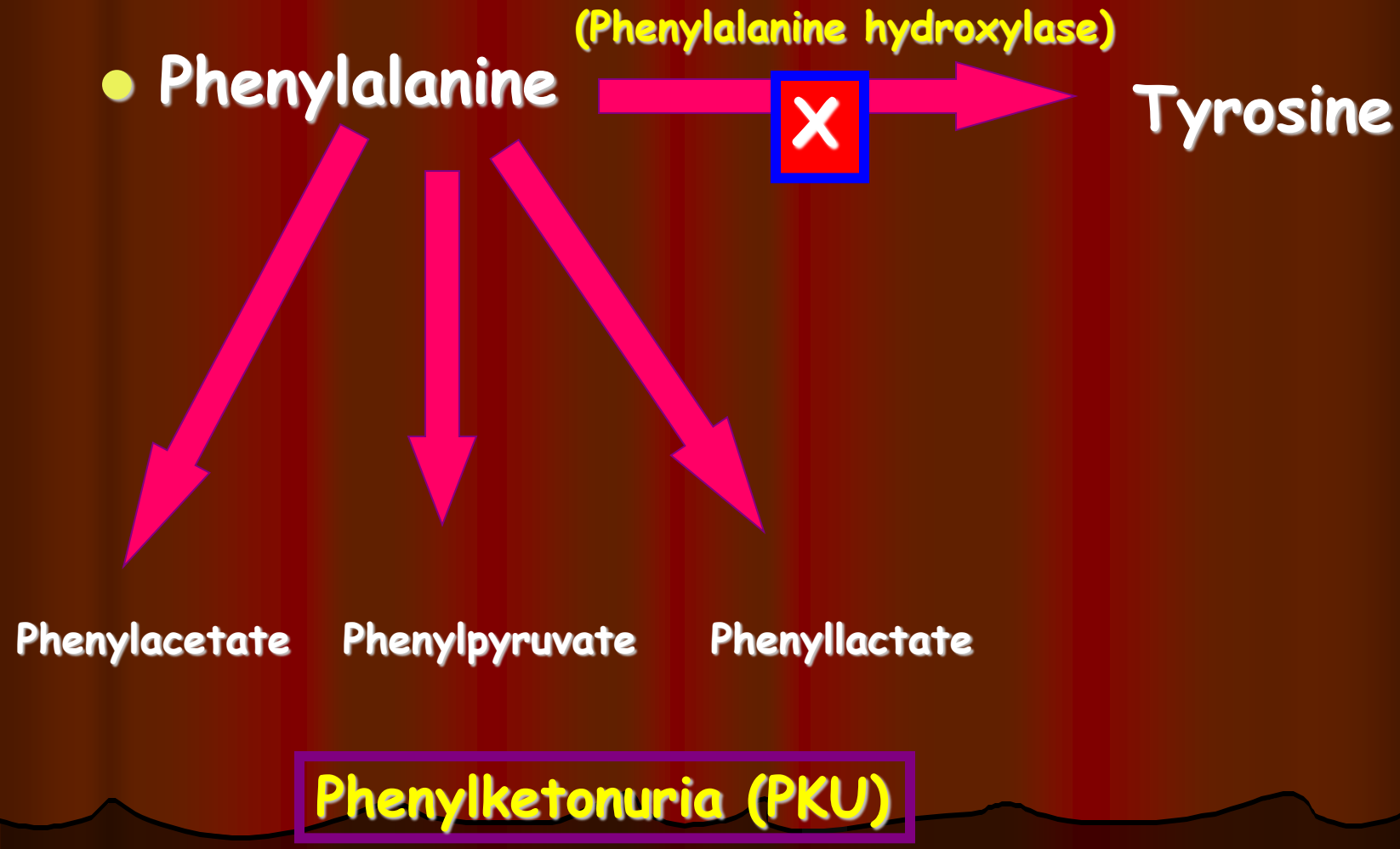
Amino acids are the building blocks for body proteins.

Essential amino acids can only be obtained from the food we eat as our body does not normally produce them.

ENZYMATIC ACTIVITY

- In cases of PKU, the enzyme that breaks down phenylalanine, phenylalanine hydroxylase, is completely or nearly completely deficient.
- This enzyme normally converts phenylalanine to another amino acid, tyrosine, which is utilized by the body.
- When this enzyme, phenylalanine hydroxylase, is absent or deficient, phenylalanine and its breakdown chemicals from other enzyme routes, accumulate in the blood and body tissues.

❖Metabolism of phenylalanine in state of (PKU)



LEVELS OF BLOOD PHENYLALANINE

- A normal blood phenylalanine level is about 1mg/dl.
- In cases of PKU, levels may range from 6-80mg/dl, but are usually greater than 30mg/dl.

WHAT HAPPENS WHEN THERE IS TOO MUCH BLOOD PHENYLALANINE?

Chronically, high levels of phenylalanine and some of its breakdown products can cause significant brain problems. There are other disorders of hyperphenylalaninemia, but classic PKU is the most common cause of high levels of phenylalanine in the blood.

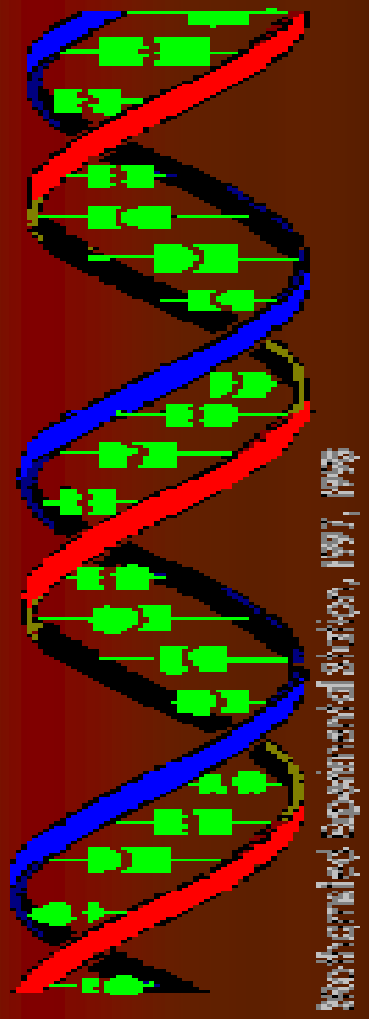
WHAT HAPPENS WHEN THERE IS TOO LITTLE BLOOD PHENYLALANINE?

It is important to remember that some phenylalanine is needed to maintain normal body activities.

Insufficient phenylalanine intake may cause mental and physical sluggishness, loss of appetite, anemia, rashes, and diarrhea.

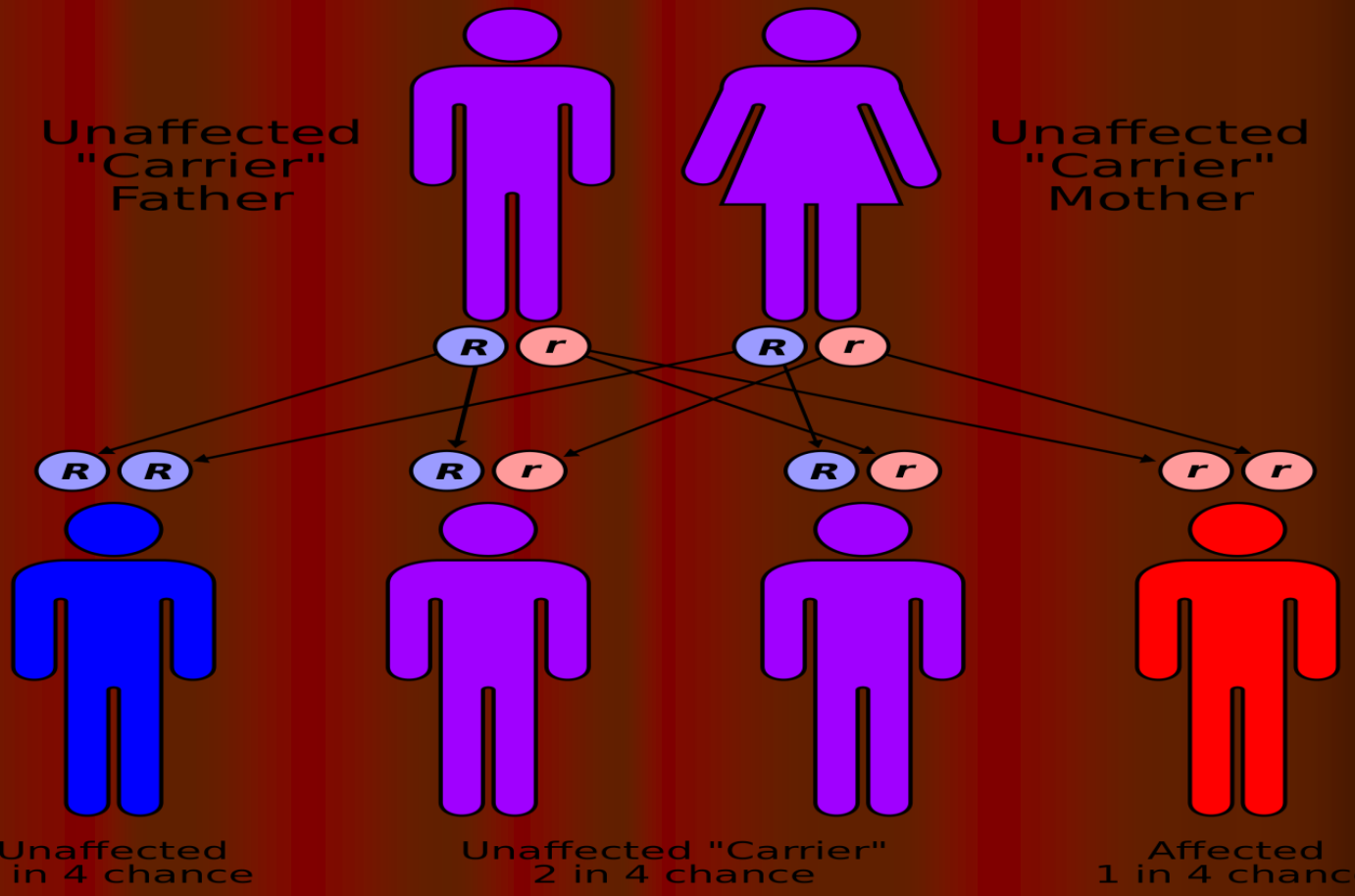
WHO DOES PKU EFFECT?

PKU is inherited as an autosomal recessive trait.



- Two people who conceive a child must both be the carriers of the defective gene in order for their child to have the disorder.
- The “carrier” for PKU does not have the symptoms.

Inheretance of PKU:



WOMEN WITH PKU

It is recommended that women with PKU who are of child bearing age, closely adhere to the low-phenlyalanine levels before conception and throughout pregnancy. The risk of miscarriage, mental retardation, microcephaly, and congenital heart disease in the child is high if the mother's blood phenylalanine is poorly controlled.

INCIDENCE OF PKU

- PKU affects about one out of every (10,000 - 20,000) Caucasian or oriental births. The incidence in African Americans is far less.

- The PKU disorder is as frequent in men as it is in women.

WHAT ARE THE SYMPTOMS OF PKU?

(About 50% of untreated infants have the following early symptoms)

Vomitting
Irritability
Eczema-like rash
Unusual odor of urine
Nervous System Problems(increased muscle tone, more active muscle tendon reflexes)
Microcephaly
Decreased body growth

OTHER SYMPTOMS OF PKU

- Prominent cheek and jaw bones widely spaced teeth
- Poor development of tooth enamel.

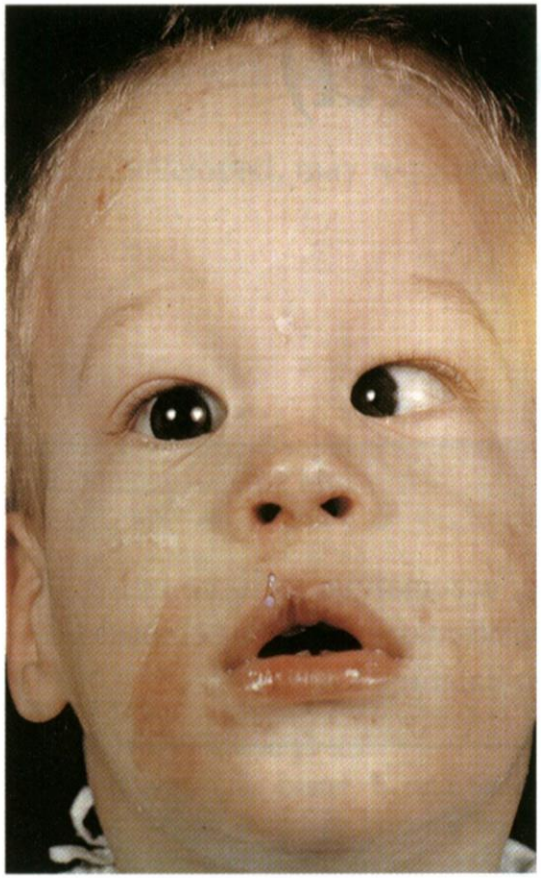


Fig. 19.3 The face of this patient with PKU illustrates the rather subtle eczematoid rash. The brown eyes remind us that not all patients with this disease have blue eyes. In addition, he had epicanthal folds and a left internal strabismus.

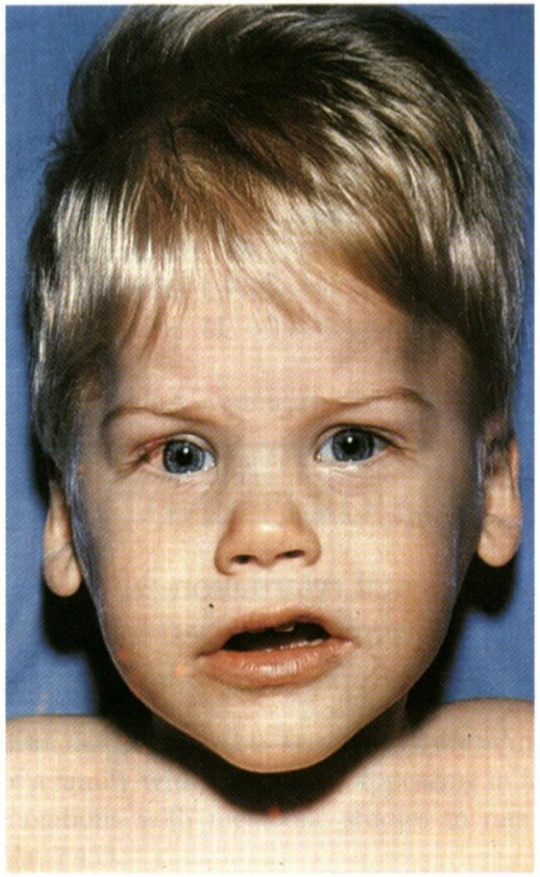


Fig. 19.4 L.S. This patient was diagnosed as having PKU at 10 months of age. The eyes were blue, the skin fair and the hair blond.

WHAT HAPPENS IF PKU GOES UNTREATED?

- If PKU goes untreated or undetected, severe brain problems occur such as seizures and mental retardation.
- This can occur as early as the second month of life, if not detected and treated.

HOW CAN PKU BE TREATED?

- Every state now screens the blood phenylalanine level of all newborns at about 3 days of age.

- This test is one of several newborn screening tests performed before or soon after discharge from the hospital.

- Usually a few drops of blood are obtained by a small prick on the heel, placed on a card and then sent for measurement.
- If the screening test is abnormal, other tests are needed to confirm or exclude PKU.
- Newborn screening allows early identification and early implementation of treatment.

- Ferric chloride + urine of new born baby→ Green colour in the presence of ketone bodies

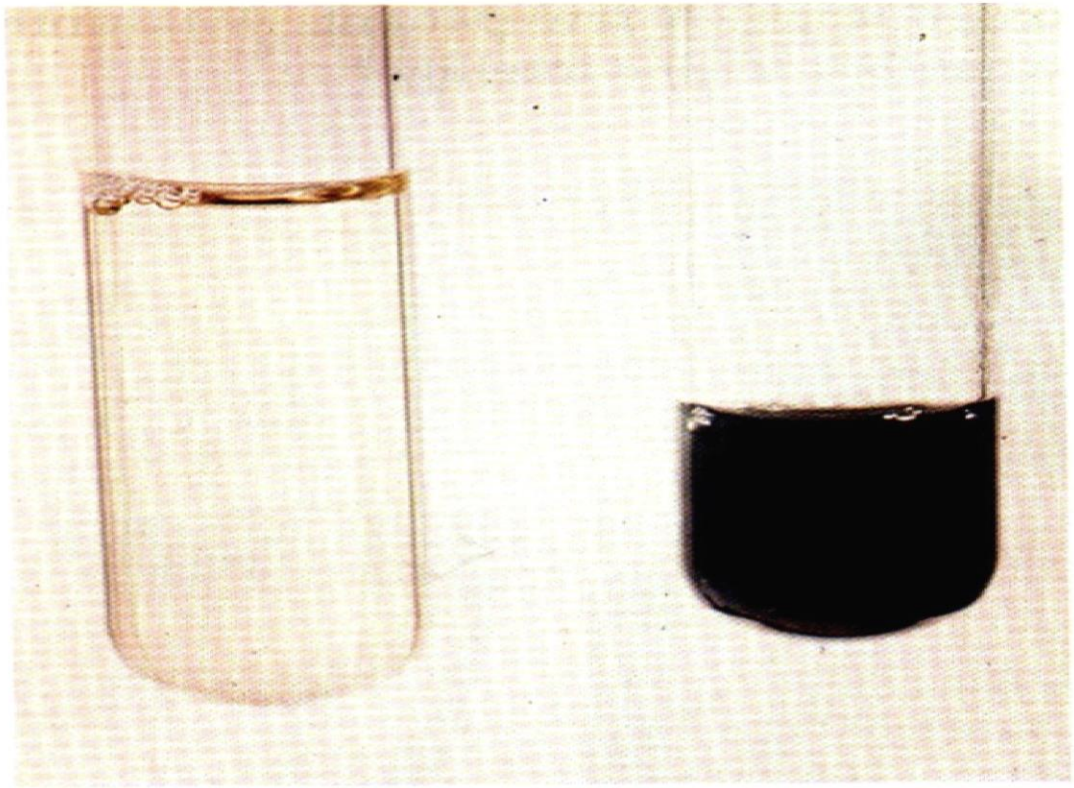
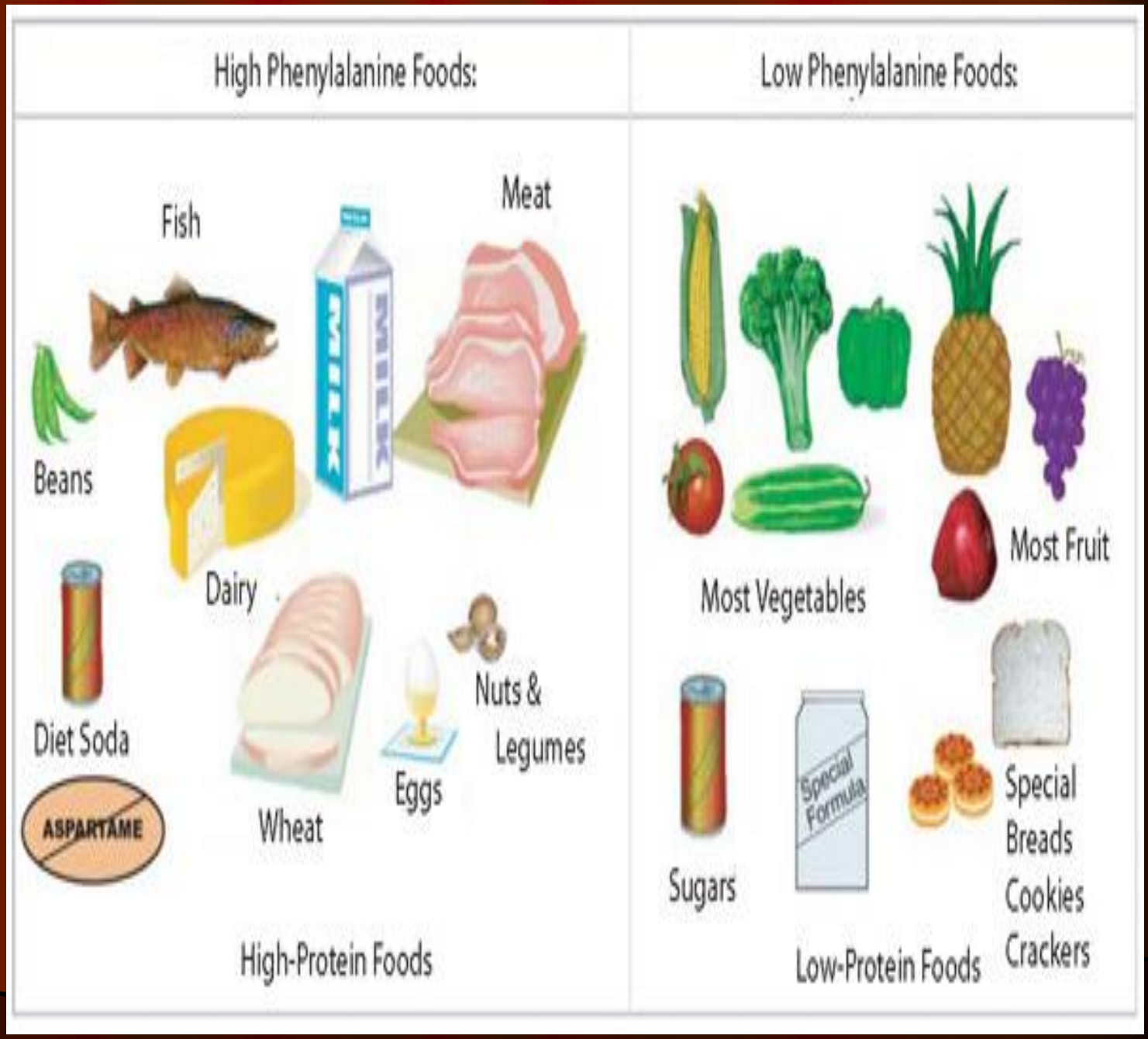


Fig. 19.2 A positive ferric chloride test in a patient with untreated PKU.

WHAT TYPE OF DIET IS SUITABLE FOR SOMEONE WITH PKU?

- The goal of PKU treatment is to maintain the blood levels of pheylalanine between 2 and 10mg/dl.

- Treatment for PKU consists of a diet low in phenylalanine, which is maintained infants with special formulas and in individuals by eliminating meat and using low protein grain products. Measured amounts of cereals, starches, fruit, and vegetables, along with a milk substitute are recommended instead.


High Phenylalanine Foods:
Low Phenylalanine Foods:
Fish
Meat
Beans
Dairy
Diet Soda
ASPARTAME
Wheat
Eggs
Nuts &
Legumes
High-Protein Foods
Most Vegetables
Most Fruit
Sugars
Special Formula
Special
Breads
Cookies
Crackers
Low-Protein Foods

OTHER THINGS TO STAY AWAY FROM

- Individuals with PKU must be alert for food sweetened with aspartame.
- NutraSweet in particular, should be avoided because it is a derivative of phenylalanine.

LIFESTYLE ADJUSTMENTS FOR PKU PATIENTS

- More frequent doctor visits
- Required dietary restrictions that may impact day to day activities.
- Permanent monitoring of blood phenylalanine levels

Thanks for your attention

Special thanks for Dr. Hazim