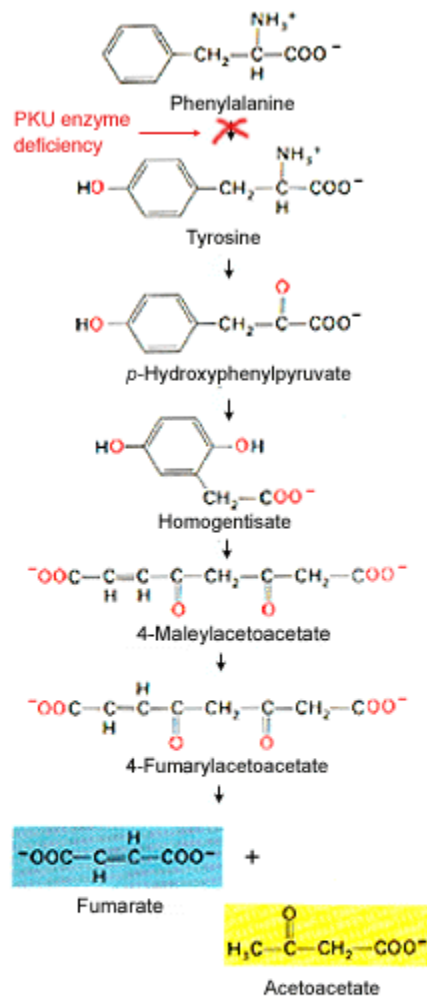




## Nutritional and Metabolic Diseases



Surplus amino acids are broken down to make metabolic energy. The first step in the degradation pathway of phenylalanine and tyrosine requires the enzyme phenylalanine hydroxylase. Individuals lacking this enzyme suffer from phenylketonuria (PKU), an inborn error of metabolism. [Modified from Stryer, L. (1988) Biochemistry 3rd edn, W.H. Freeman and Co., with permission.]

Metabolism is the means by which the body derives energy and synthesizes the other molecules it needs from the fats, carbohydrates and proteins we eat as food, by enzymatic reactions helped by minerals and vitamins.

This global statement masks the complicated network of enzyme- catalyzed reactions that occurs in cells. Although this page is devoted to diseases caused by errors in metabolic processes, there is actually a significant level of tolerance of errors in the system: often, a mutation in one enzyme does not mean that the individual will suffer from a disease. A number of different enzymes may compete to modify the same molecule, and there may be more than one way to achieve the same end result for a variety of metabolic intermediates. Disease will only occur if a critical enzyme is disabled, or if a control mechanism for a metabolic pathway is affected.

Here, we highlight the diseases of metabolism for which a gene has been identified, cloned and mapped. Many of these are inborn errors of metabolism: inherited traits that are due to a mutation in a metabolic enzyme; others involve mutations in regulatory proteins and in transport mechanisms.

## Diseases

Adrenoleukodystrophy

Diabetes, type 1

Gaucher disease

Glucose galactose malabsorption

Hereditary hemochromatosis

Lesch-Nyhan syndrome

Maple syrup urine disease

Menkes syndrome

Niemann-Pick disease

Obesity

Pancreatic cancer

Phenylketonuria

Prader-Willi syndrome

Porphyria

Refsum disease

Tangier disease

Tay-Sachs disease

Wilson's disease

Zellweger syndrome