Early, accurate diagnosis and treatment can sometimes prevent further complications of the disorder. Family education and counseling may prevent the appearance of the disorder in future family members.

PKU (phenylketonuria)

PKU is an inherited condition that, if untreated, results in mental retardation and disturbances of behavior. Infants with PKU appear healthy and normal at birth. Within a few months on a normal diet, however, symptoms of the disorder begin to appear. Symptoms include an abnormal growth rate and abnormal development, such as the inability to walk or talk.

The name of the disorder, *phenylketonuria*, refers to chemicals known as phenylketones, which appear in the urine of the affected individuals. In a normal person, enzymes convert phenylalanine, one of the 20 amino acids present in many foods, to the amino acid tyrosine. However, individuals homozygous for PKU lack the gene that codes for the enzymes to convert phenylalanine. In their place are recessive alleles that do not code for the enzyme. Thus, in individuals with PKU, phenylalanine cannot be converted to tyrosine. The phenylalanine in the diet is instead broken down to phenylketones and other derivatives that end up in the urine. Still other derivatives accumulate in the body, where they are toxic (poisonous) to the nervous system and result in mental retardation.

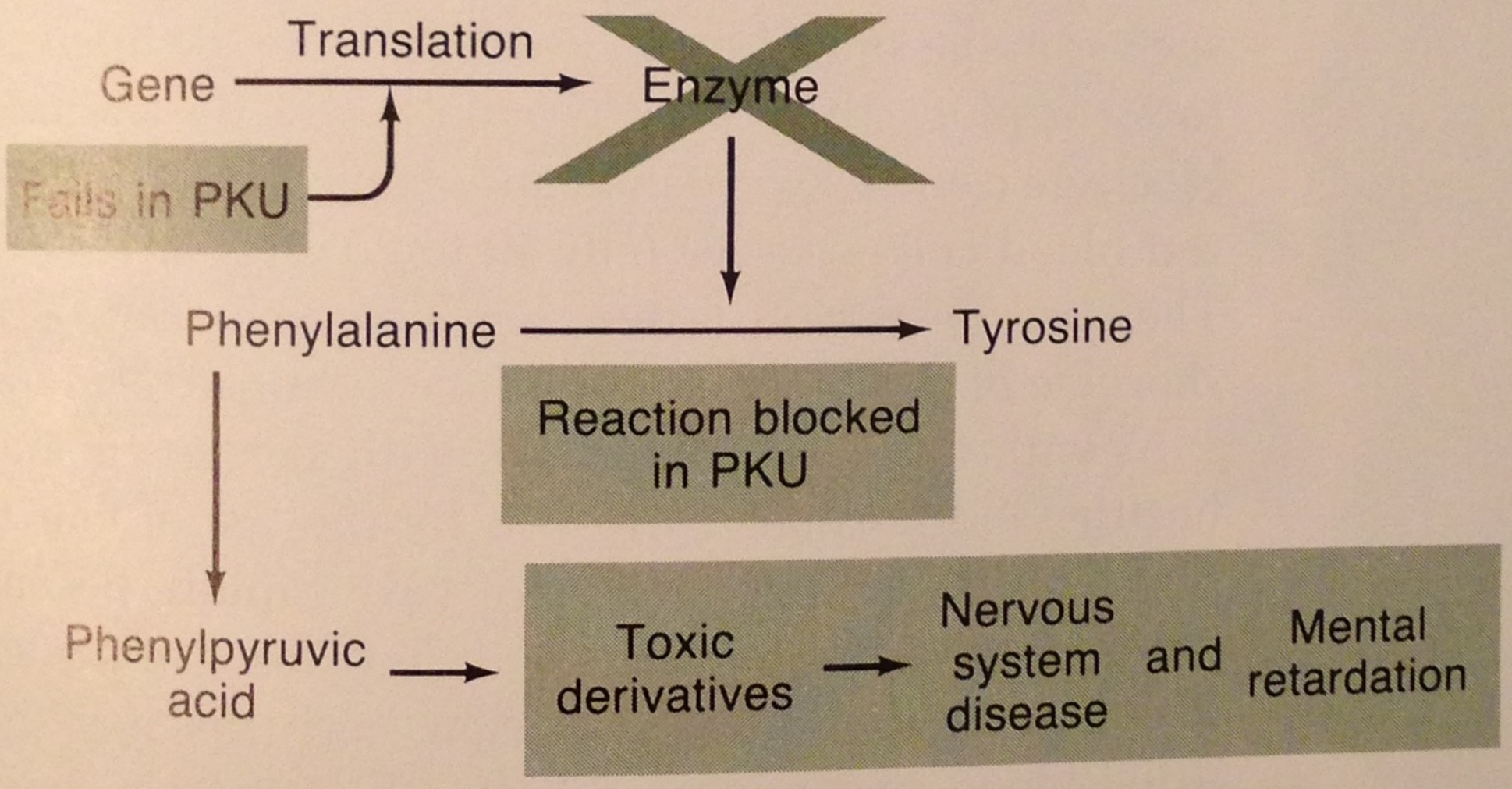

Fig. 22–2. Individuals with PKU lack genes that code for the synthesis of an enzyme needed to convert phenylalanine to tyrosine.

In addition to the harmful accumulation of the breakdown products of phenylalanine, the affected individual suffers from a lack of tyrosine. Tyrosine is an essential amino acid that the body needs for many of its functions, including the production of the pigment melanin.

With early detection, infants can be placed on a special diet low in phenylalanine and supplemented with tyrosine. To be effective, this diet must be started within the first weeks of life. Therefore, early diagnosis of PKU is essential.

Human geneticists have developed and continue to develop tests to diagnose genetic disorders and detect carriers. Application of such tests to populations at high risk or to the population in general is called *screening*. Newborn screening programs test for several genetic disorders, including

PKU and sickle-cell anemia (see page 398). Early detection of such disorders is a benefit of modern biology to humans.

PKU testing is an example of the success of newborn screening programs. There have been no PKU patients institutionalized with mental retardation since such programs started in New York in 1965. Now that women who have received treatment for PKU have reached reproductive age, it has been found that the elevated phenylalanine in their blood is toxic to their fetus during their pregnancy. This makes continuation of the controlled diet even more important.

The lifelong cost of maintaining a PKU patient in an institution can exceed one million dollars. The cost of the newborn screening program is a few dollars per child. Maintenance of the special diet costs about $3,000 a year. From a strictly financial point of view, this public health screening program is "cost effective." Among the issues that still remain are confidentiality of the screening programs and access to good treatment for PKU patients.