The present technique for screening for phenylalaninaemia was introduced by Guthrie (1960) and is now in common use throughout the world. The Guthrie technique involves a simple scraping of skin from the heel of the newborn infant, followed by drying and application of a filter paper disc. The infant is then dried and returned to the mother. The filter paper is incubated at 37°C for several hours, and the resulting disc is dried. The disc is then sent to a central laboratory where it is treated with a chemical reagent to develop the test. The disc is then scored for coloration of the test area, which is a reflection of the amount of phenylalanine present in the specimen. The score is then compared to a standard curve to determine the concentration of phenylalanine in the specimen. The test is highly specific and sensitive, and it can detect even very small amounts of phenylalanine in the specimen. It has been shown to have a sensitivity of 99.9% and a specificity of 100%.

The incidence of phenylketonuria (PKU) is estimated to be 1 in 20,000 to 30,000 births in the United States. The condition is caused by a deficiency of the enzyme phenylalanine hydroxylase, which is responsible for the conversion of phenylalanine to tyrosine. This deficiency leads to the accumulation of phenylalanine in the body, which can cause a variety of serious health problems, including mental retardation, seizures, and vision and hearing loss.

The early detection of PKU is critical to prevent these complications. Therefore, all newborns in the United States are screened for PKU using the Guthrie test. If the test is positive, further testing is performed to confirm the diagnosis and to determine the severity of the condition. Treatment is then initiated as soon as possible to prevent or minimize the effects of the condition. The treatment typically involves a diet that is low in phenylalanine and rich in tyrosine. The diet is usually started within the first few days of life and continued for life.

The Guthrie test is highly effective in detecting PKU, but it is also capable of detecting a wide range of other conditions that can be caused by deficiencies in enzymes or other conditions that affect amino acid metabolism. These conditions include tyrosinemia, hypermethioninemia, and hyperphenylalaninemia. The Guthrie test can also be used to screen for other conditions, such as galactosemia, which is caused by a deficiency of the enzyme galactose-1-phosphate uridyltransferase, and maple syrup urine disease, which is caused by a deficiency of the enzyme branched-chain 2-oxoacid dehydrogenase complex.

The Guthrie test is a simple, rapid, and highly effective screening test that is widely used to detect PKU and other amino acid disorders. It is an essential component of newborn screening programs in the United States and in many other countries around the world. The test has saved thousands of lives and has greatly improved the quality of life for those with PKU. It has also helped to reduce the cost of medical care by identifying and treating these conditions early. The Guthrie test is a testament to the power of science and technology in improving the health of newborns and infants.