

## Phenylketonuria

---

### Description:

*Phenylketonuria (or PKU) is an inherited disorder, that, left untreated, changes cells inside the brain and can lead to mental retardation. This dramatic change in behaviour can be traced to a tiny mutation in a single gene on chromosome 12. Fortunately, through routine newborn screening, almost all affected newborns are now diagnosed and treated early, allowing them to grow up with normal intelligence.*

*Due to this missing or deficient enzyme, children with PKU cannot process a part of the protein called phenylalanine, which is present in nearly all foods. Without treatment, phenylalanine builds up in the bloodstream and causes brain damage and mental retardation.*

*All newborn babies are tested for PKU, using a heel-prick blood test taken during the first few days of life. About one in 10,000 newborn babies are affected with PKU.*

Phenylketonuria