Phenylketonuria (PKU)

Phenylketonuria is an **autosomal recessive**disease (8-10 cases / 100,000 individuals) due to the **absence** or **phenylalanine hydroxylase activity**. It physiologically catalyzes the **hydroxylation** of Phe to Tyr. In the case of an enzyme defect, there is an alternative degradation of Phe - phenylpyruvate (transamination), phenylacetate, phenylacetate or phenylethylamine is formed. These substances **accumulate in tissues** and body fluids and cause a typical **urine odour**. Some of them cause severe **brain damage**.

Phenylketonuria was the first human genetic defect in amino acid metabolism to be discovered and is currently one of the diseases screened in all newborns. If we can recognize it at this age, we can **prevent brain damage** with a strict Phe-restricted diet.

🔎 *For more information see Phenylketonuria.*

'