# Rapid Analysis for the Diagnosis of PKU

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**Using the Biochrom 30 Amino Acid Analyser, phenylalanine levels can be measured using a rapid, accurate programme for the diagnosis of PKU. Routine newborn screening is necessary to allow affected infants to be treated and monitored appropriately.**

Phenylketonuria (PKU) is a genetic inborn error of metabolism, that is detectable during the first days of life with appropriate blood testing. The absence or deficiency of an enzyme that is responsible for processing the essential amino acid phenylalanine characterizes PKU.

Without treatment, most infants with PKU develop mental retardation. Early detection of PKU can allow patient treatment to prevent mental retardation, as well as other associated problems. Treatment consists of a carefully-controlled phe-restricted diet begun during the first days or weeks of life. Frequent blood monitoring of PKU patients is necessary, especially during infancy and early childhood when it is important to keep the blood phenylalanine levels at the safest level. Biochrom Amino Acid Analysers are widely used for the routine analysis of phenylalanine.

Clinical laboratories have a continuous requirement for analyses of large numbers of samples. Rapid, accurate analysis is critical to meet the demand of the screening programmes. Improvements in peak resolution, accuracy, run times, and quantitation by using a dedicated instrument has enabled clinical laboratories to screen large numbers of samples per week.

Using the Biochrom 30 Amino Acid Analyser phenylalanine can be rapidly and accurately quantified using norleucine as internal standard. The separation is carried out using a 20 cm x 4.6 mm high performance physiological column using buffer DII predominantly. The program enables 25 analyses to be performed a day. In addition tyrosine and homocystine are also well resolved thus enabling screening for the other metabolic diseases such as Tyronisis and Homocystinuria.

**Reference:** Biochrom Application Note 57

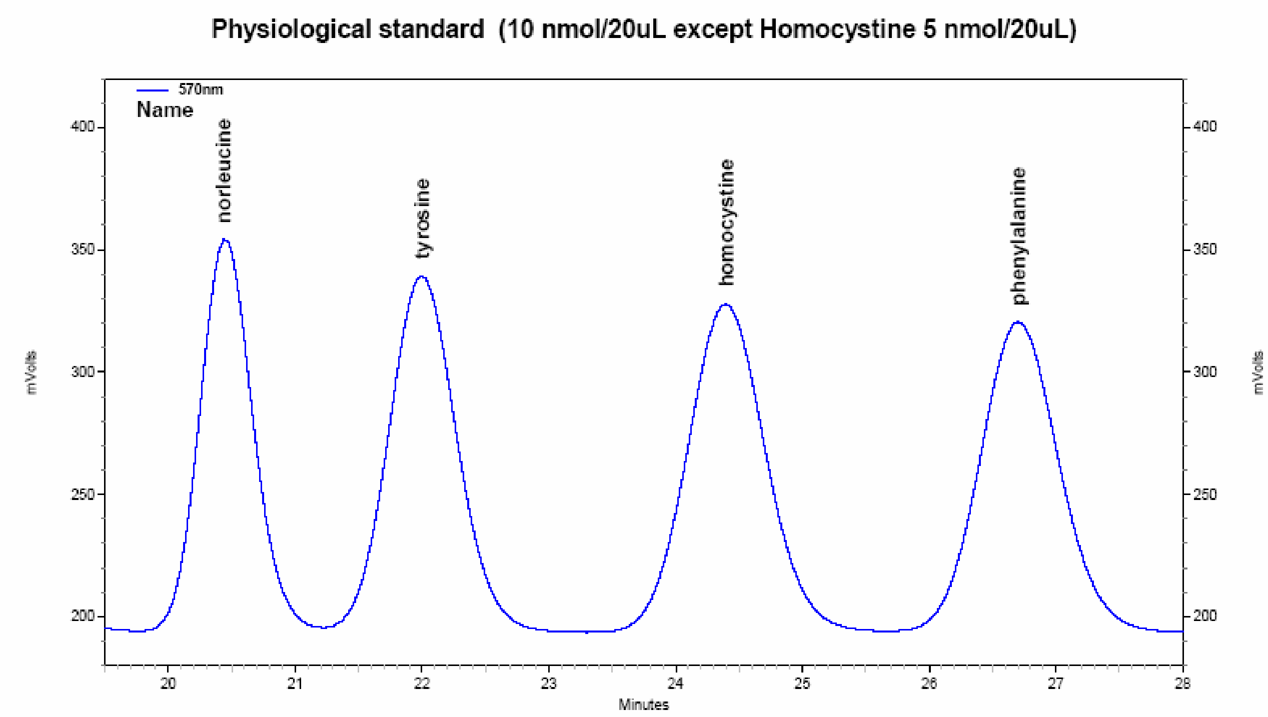


***Figure 1.*** *PKU screening:*

# BioSys

Sample: Physiological Fluid std | Amount Loaded: 10 nmol

Column Type: Peek | Column Number: U-1085 | Resin Batch: 07216

Bed Length (mm): 200 | Diameter (mm): 4.6 | Instrument Serial Number: 88352

| | Buffer | Nin |
|---|---|---|
| Flow Rate (ml/h): | 25 | 20 |

| | Buffer | Molarity | pH | Batch No. |
|---|---|---|---|---|
| Buffer 3 - | Lithium Citrate Buffer CII | 0.50 | 3.15 | 11570 |
| Buffer 4 - | Lithium Citrate Buffer DII | 0.90 | 3.50 | 11830 |
| Buffer 5 - | Lithium citrate pH 3.55 | 1.65 | 3.55 | 11858 |
| Buffer 6 - | Lithium hydroxide Solution | 0.30 | | |
| Reagent | Ninhydrin | | | 11884 |
| | Ultrosolve | | | 11839 |

**Title:** **PKU**

Nin Flow Rate: 20.0 ml/h

| No. | Time | Temp | Buffer | Pump | Nin | Rec | Commands |
|---|---|---|---|---|---|---|---|
| 1 | 01:00 | 75°C | 4 | 25.0ml/h | ON | OFF | |
| 2 | 00:00 | 75°C | 4 | 25.0ml/h | ON | OFF | Reset |
| 3 | 01:00 | 75°C | 4 | 25.0ml/h | ON | OFF | Load |
| 4 | 15:00 | 75°C | 4 | 25.0ml/h | OFF | ON | |
| 5 | 07:00 | 75°C | 4 | 25.0ml/h | ON | ON | |
| 6 | 05:00 | 75°C | 5 | 25.0ml/h | ON | ON | |
| 7 | 05:00 | 80°C | 6 | 25.0ml/h | ON | ON | |
| 8 | 03:00 | 80°C | 3 | 25.0ml/h | OFF | OFF | |
| 9 | 15:00 | 75°C | 3 | 31.0ml/h | OFF | OFF | |
| 10 | 04:00 | 75°C | 4 | 25.0ml/h | ON | OFF | |