

Phenylalanine Dehydrogenase

Features/Benefits

- Neonatal screening enzyme
- · Phenylketonuria screening (Pku) test
- · Lower false positives than other tests
- · Lower total costs than Guthrie method
- Simple and rapid

Phenylketonuria (PKU) is a genetic metabolic disorder in which individuals are unable to metabolise phenylalanine. If left undiagnosed, it can lead to brain and developmental impairments. However, with early diagnosis and proper treatment, individuals can develop normally. Newborn screening for PKU has been common practice in the Western world for many years, and such programs are expanding globally.

Historically, the Guthrie microbial test has been the standard screening method. However, it is increasingly being replaced by a more affordable, faster, and more sensitive enzyme-based test using phenylalanine dehydrogenase. This newer method has demonstrated superior sensitivity and reliability compared to previous approaches, such as the Guthrie and fluorometric tests, and it produces fewer false positives due to its low cross-reactivity.

Specification

| Minimum Activity | 6 units of phenylalanine dehydrogenase per mg solid |
|---------------------------|--|
| Specific Activity | > 20 units/mg protein |
| Unit Definition | 1 unit converts 1 μM of NAD+ in the presence of phenylalanine at pH 10.5 and 30°C |
| Biological Source | Sporosarcina sp. |
| Form | Lyophilised off-white powder |
| Km | 5 mM (on NAD) |
| IUB No & Type | 11.4.1.20 / L-phenylalanine: NAD oxido-reductase |
| Optimum pH Range | 10.0 - 11.0 |
| Optimum Temperature Range | 35 - 40°C |

Phenylalanine dehydrogenase (PheDH) is an enzyme used in neonatal screening for PKU. It converts phenylalanine into phenylpyruvate with the help of NAD+, which acts as an electron acceptor. The reduced form of NAD+ (NADH) then transfers electrons to a substance called lodonitrotetrazolium hydrochloride (INT) via the enzyme diaphorase, producing a red formazan dye. This dye can be measured using a spectrophotometer to determine phenylalanine levels.

Blood samples from newborns are taken through a heel prick and placed on filter paper. In commercial kits, the blood spot is treated with methanol to preserve proteins, and the amino acids are then washed off into water. The sample is mixed with PheDH, NAD+, diaphorase, and INT, and the colour change is measured using a spectrophotometer. Elevated phenylalanine levels suggest PKU.

To ensure accurate results, the optimum amounts of enzyme, buffers, and reaction conditions must be determined in studies. The indicated optimum temperature and pH conditions were determined as measured by the Biocatalysts' assay procedure. More information on PKU and the screening process can be found in the references below.

Dooley K.C., 1992, Clin. Biochem., 25, pp 271 - 275. Campbell R.S., Hollifield R.D., Varsani H., Milligan T.P., Brearley G. and Price C.P., 1994, Anal. Clin. Biochem., 31, pp 140-146. Campbell R.S., Brearley G., Varsani H., Morris H.C., Milligan T.P., Halls K. et. al., Clin. Chimica. Acta., 1992, 210, pp197 - 210. Wendel U., Hummel W. and Langenbeck U., 1989, Anal. Biochem., 180, pp 91- 94.

Health & Safety

Always read the Material Safety Datasheet (MSDS) before use and retain. If you are in any doubt about recommended product handling and safety, please contact Biocatalysts before use. Generally, when using enzymes avoid contact with the skin and eyes and do not breathe dusts or aerosols containing them.

GM Status

This product does not contain GMMs or genetically modified material.

Food Status

This product has not been produced as a food grade product but has been manufactured under ISO 9001 accreditation.

Quality

- 1. Good Manufacturing Practice (GMP) The Company's integrated management system encompasses Total Quality, Health and Safety, Food Safety and GMP.
- 2. Biocatalysts Ltd is certified to ISO9001, ISO14001, ISO45001 and FSSC 22000.

Availability

Available in 1,000- and 10,000-unit packs.

Storage

This product is fully active when stored desiccated at -20°C.

