

## Human Adenosine deaminase Reference Standard

**Cat. No.** ADA-9H    **Lot. No.** (See product label)

### SPECIFICATION

#### Product Overview

Each sample is in lyophilised form and equivalent to about 1 mL of adenosine deaminase (ADA 1) from Human erythrocytes. The preparation has been stabilised by incorporation in a matrix of 50 mmol/L Tris/HCl buffer pH=7.4 and human serum albumin 30 g/L. No contamination, as assessed by measurement of their catalytic activity, has been detected for the following enzymes: acid phosphatase, acetylcholinesterase, glutamate dehydrogenase, glucose-6-phosphate dehydrogenase and adenosine deaminase isoenzyme 2. L-lactate dehydrogenase, alanine aminotransferase and aspartate aminotransferase were found in trace amounts 0.39%, 0.01% and 0.09%, respectively (% of total adenosine deaminase catalytic activity). The material is kept under dry nitrogen in rubber stoppered vials. The residual moisture mass fraction of the sample had a value of (0.74±0.21 %). The intended use of the material is to validate, to calibrate or to assess the performance of adenosine deaminase catalytic concentration measurement procedures. The user must assure that the transfer procedure is adequate.

#### Species

Human

#### Description

This gene encodes an enzyme that catalyzes the hydrolysis of adenosine to inosine. Various mutations have been described for this gene and have been linked to human diseases. Deficiency in this enzyme causes a form of severe combined immunodeficiency disease (SCID), in which there is dysfunction of both B and T lymphocytes with impaired cellular immunity and decreased production of immunoglobulins, whereas elevated levels of this enzyme have been associated with congenital hemolytic anemia.

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|                             |                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         |
|-----------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Usage</b>                | It is not recommended that a portion of the lyophilised material contained in an ampoule be used. The entire content of the ampoule must be reconstituted. To make it ready for use, the material has to be reconstituted according to the procedure described in Chapter 9 of the certification report. The commutability of the material with routine in vitro diagnostic devices has not been assessed. If the material is used for the calibration of in vitro diagnostic devices the commutability has to be assessed by the user. |
| <b>Quality Control Test</b> | Using adenosine as substrate and glutamate dehydrogenase as auxiliary enzyme measurements are performed at 37 °C (Bota A, Gella FJ, Canalias F. Optimization of adenosine deaminase assay by response surface methodology. Clin Chim Acta 2000;290:145-57).                                                                                                                                                                                                                                                                             |
| <b>Notes</b>                | The serum material was produced from blood from healthy blood donors. Each portion of serum was tested negative for Anti-HIV-1&2, Anti-HTLV-I&II. However, the material is of human origin and should be handled with adequate care. For in vitro analysis only.                                                                                                                                                                                                                                                                        |
| <b>Storage</b>              | Upon arrival at the laboratory the material shall be stored at -20 °C. However, the European Commission cannot be held responsible for changes that happen during storage of the material at the customer's premises, especially of opened samples.                                                                                                                                                                                                                                                                                     |
| <b>Concentration</b>        | Human Adenosine Deaminase (ADA 1): 2.55 ukat/L                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          |

## GENE INFORMATION

|                        |                                                     |
|------------------------|-----------------------------------------------------|
| <b>Gene Name</b>       | ADA adenosine deaminase [ Homo sapiens ]            |
| <b>Official Symbol</b> | ADA                                                 |
| <b>Synonyms</b>        | ADA; adenosine deaminase; adenosine aminohydrolase; |

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|----------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Gene ID</b>             | 100                                                                                                                                                                                                                                                                                                                                                                      |
| <b>mRNA Refseq</b>         | NM_000022                                                                                                                                                                                                                                                                                                                                                                |
| <b>Protein Refseq</b>      | NP_000013                                                                                                                                                                                                                                                                                                                                                                |
| <b>MIM</b>                 | 608958                                                                                                                                                                                                                                                                                                                                                                   |
| <b>UniProt ID</b>          | P00813                                                                                                                                                                                                                                                                                                                                                                   |
| <b>Chromosome Location</b> | 20q13.12                                                                                                                                                                                                                                                                                                                                                                 |
| <b>Pathway</b>             | C-MYB transcription factor network, organism-specific biosystem; Metabolic pathways, organism-specific biosystem; Metabolism, organism-specific biosystem; Metabolism of nucleotides, organism-specific biosystem; Primary immunodeficiency, organism-specific biosystem; Primary immunodeficiency, conserved biosystem; Purine metabolism, organism-specific biosystem; |
| <b>Function</b>            | adenosine deaminase activity; adenosine deaminase activity; hydrolase activity; metal ion binding; protein binding; purine nucleoside binding; zinc ion binding; zinc ion binding;                                                                                                                                                                                       |