

Product Specification Sheet

Human Synuclein Recombinant protein Mutant (E46K)

Cat. # SYN-E46K-R

Human Alpha SYN Recombinant protein, mutant (E46K)

SIZE: 50 ug

Parkinson's disease (**PD**) is a common neurodegenerative disorder with a lifetime incidence of approximately 2 percent; the clinical manifestations of this neurodegenerative disorder include resting tremor, muscular rigidity, bradykinesia, and postural instability. A relatively specific pathological feature accompanying the neuronal degeneration is an intracytoplasmic inclusion body, known as the **Lewy body**. A mutation was identified in the **α-synuclein** gene, which codes for a presynaptic protein thought to be involved in neuronal plasticity, this mutation may cause a conformational change that renders α-synuclein more prone to self aggregation and deposition in Lewy bodies, which finally leads to oxidative stress and misfolding of α-synuclein.

Parkin gene, mutations in this gene are reported in early autosomal-recessive form of PD, however these mutations do not degenerate **Lewy bodies**. The Parkin gene product (**Parkin**) is involved in protein degradation as a ubiquitin protein ligase, the known substrates of Parkin include Pael-R (Parkin-associated endothelin receptor-like receptor), Ubiquitination of Pael-R by Parkin leads to its degradation in the proteasome, however failure to ubiquitinate it leads to death of neuron.

The synuclein exists in 3 isoform **α-syn (chr4 4q21)**, a 140aa protein, implicated in pathogenesis of PD and related neurodegenerative disorders, it is mainly expressed in brain specifically in neuronal cell bodies and synapses. The 134 aa **β-syn (chr5 5q35)** is homologous to 14 kDa bovine phosphonuroprotein 14; SCNB has been shown to be highly expressed in the substantia nigra of the brain. Recently a new isoform termed **γ-synuclein (SNCG)** or breast cancer gene 1 (BCG1) has been cloned (human 127 aa (chr10 10q23), rat/mouse 123 aa). Higher levels of expression of SNCG have been reported in advanced breast carcinomas. All three synuclein show ~40% identity.

Source and Storage

Human Alpha synuclein recombinant protein (E46K) expressed in E.coli Purity: >95% by SDS-PAGE (Mol.wt 14.45kD). A point mutant (E46K) of the alpha synuclein gene that has been linked to autosomal dominant early onset Parkinson's disease..It is provided in 50 mM Sodium Phosphate buffer, 60 mM NaCl, 17 mM citric acid, pH 6.2 in liquid (100 ug/100 ul) or lyophilized. The lyophilized products should be reconstituted in water or a desired buffer at a given volume and lightly vortex and mix for 15 min at room temp). It can then be used or aliquoted for storage in small aliquots at -70oC or below.

General References:

Park, S.M et al 2002, Blood 100, 2506; Polymeropoulos, M.H et al 1997, Science 276, 2045; Jakes. R et al. 1994, FEBS Lett. 345, 27.

*This product is for *in vitro* research use only.

SYN-E46K-R

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