abcam

Product datasheet

Recombinant Human DNA Polymerase gamma protein ab114519

1 Image

Overview

Product name	Recombinant Human DNA Polymerase gamma protein
Protein length	Protein fragment

Description

Nature	Recombinant
Source	Wheat germ

Amino Acid Sequence

Accession	P54098
Species	Human
Sequence	CISIHDEVRYLVREEDRYRAALALQITNLLTRCMFAYKLGLNDLPQSVAF FSAVDIDRCLRKEVTMDCKTPSNPTGMERRYGIPQGEALDIYQIIELTKG SLEKRSQPGP
Molecular weight	38 kDa including tags
Amino acids	1130 to 1239

Specifications

Our Abpromise guarantee covers the use of **ab114519** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	ELISA SDS-PAGE Western blot
Form	Liquid
Additional notes	Protein concentration is above or equal to 0.05 mg/ml. Best used within three months from the date of receipt.

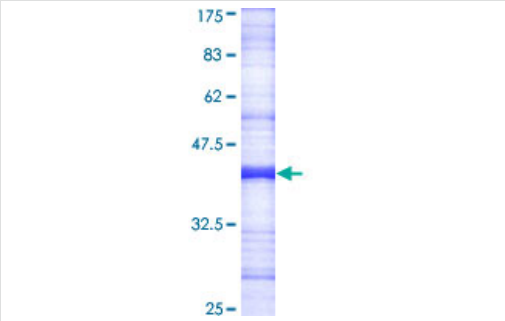
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.79% Tris HCl, 0.3% Glutathione

General Info

Function	Involved in the replication of mitochondrial DNA.
Involvement in disease	Defects in POLG are the cause of progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal dominant type 1 (PEOA1) [MIM:157640]. Progressive external ophthalmoplegia is characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. Defects in POLG are a cause of progressive external ophthalmoplegia with mitochondrial DNA deletions autosomal recessive (PEOB) [MIM:258450]. PEOB is a severe form of progressive external ophthalmoplegia. It is clinically more heterogeneous than the autosomal dominant forms. Can be more severe. Defects in POLG are a cause of sensory ataxic neuropathy dysarthria and ophthalmoparesis (SANDO) [MIM:607459]. SANDO is a clinically heterogeneous systemic disorder with variable features resulting from mitochondrial dysfunction. It shares phenotypic characteristics with autosomal recessive progressive external ophthalmoplegia and mitochondrial neurogastrointestinal encephalopathy syndrome. The clinical triad of symptoms consists of sensory ataxic, neuropathy, dysarthria, and ophthalmoparesis. Defects in POLG are a cause of Alpers-Huttenlocher syndrome (AHS) [MIM:203700]; also called Alpers diffuse degeneration of cerebral gray matter with hepatic cirrhosis. AHS is an autosomal recessive hepatocerebral syndrome. The typical course of AHS includes severe developmental delay, intractable seizures, liver failure, and death in childhood. Refractory seizures, cortical blindness, progressive liver dysfunction, and acute liver failure after exposure to valproic acid are considered diagnostic features. The neuropathological hallmarks of AHS are neuronal loss, spongiform degeneration, and astrocytosis of the visual cortex. Liver biopsy results show steatosis, often progressing to cirrhosis. Defects in POLG are a cause of mitochondrial neurogastrointestinal encephalopathy syndrome (MNGIE) [MIM:603041]; also known as myoneurogastrointestinal encephalomyopathy. MNGIE is an autosomal recessive disease associated with multiple deletions of skeletal muscle mitochondrial DNA (MtDNA). It is clinically characterized by onset between the second and fifth decades of life, ptosis, progressive external ophthalmoplegia, gastrointestinal dysmotility (often pseudoobstruction), diffuse leukoencephalopathy, thin body habitus, peripheral neuropathy, and myopathy. Defects in POLG are a cause of Leigh syndrome (LS) [MIM:256000]. LS is a severe neurological disorder characterized by bilaterally symmetrical necrotic lesions in subcortical brain regions.
Sequence similarities	Belongs to the DNA polymerase type-A family.
Cellular localization	Mitochondrion.

Images



SDS-PAGE - DNA Polymerase gamma protein (ab114519)

12.5% SDS-PAGE analysis of DNA Polymerase gamma protein (ab114519). Stained with Coomassie Blue.

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