Recombinant Human FXN protein, His-tagged

Cat. No. FXN-19H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human FXN (amino acids 56-210) fussed with His tag at N-terminal was expressed in E. coli.
Species	Human
Source	E.coli
ProteinLength	56-210 a.a.
Description	Frataxin was discovered as defects in the frataxin gene are causative of Friedreich's ataxia, a rare progressive neurological disorder. The defect is autosomal recessive, meaning that both frataxin genes must be defective in a particular individual in order to generate the disease phenotype. The genetic defects are usually localized to the first intron of the frataxin gene, which in normal individuals includes 7-22 copies of tandemly repeated GAA trinucleotides. In affected individuals, there are many more GAA repeats, 200-900 being noted in the original paper, with most individuals having 700-800. This disease is therefore one of the family of diseases associated with various forms of nucleotide repeat amplification, other well known examples being Huntington's disease, some forms of ALS and fragile X syndrome. In the case of Friedreich's ataxia, the repeats result in lowered level of frataxin protein production, and this in turn leads to progressive cell death, particularly in neurons of the spinal cord and peripheral ganglia.The normal function of frataxin is not well understood, but appears to be involved in protecting mitochondria from free radical damage. In affected individuals, there are motor and sensory deficits and a variety of other

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	problems which may include heart disorders and diabetes. The HGNC name for this protein is FXN.
Molecular Mass	27 kDa

GENE INFORMATION

Gene Name	FXN frataxin [Homo sapiens]
Official Symbol	FXN
Synonyms	FA; X25; CyaY; FARR; FRDA; frataxin, mitochondrial; Friedreich ataxia protein
Gene ID	2395
mRNA Refseq	NM_000144
Protein Refseq	NP_000135
MIM	606829
UniProt ID	Q16595
Chromosome Location	9q21.11
Pathway	HIF-2-alpha transcription factor network, organism-specific biosystem; Metabolism, organism-specific biosystem; Metabolism of proteins, organism-specific biosystem
Function	2 iron, 2 sulfur cluster binding; ferric iron binding; ferrous iron binding

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