



Human FXN peptide (DAG-P0511)

This product is for research use only and is not intended for diagnostic use.

PRODUCT INFORMATION

| Antigen Description | This nuclear gene encodes a mitochondrial protein which belongs to FRATAXIN family. The protein functions in regulating mitochondrial iron transport and respiration. The expansion of intronic trinucleotide repeat GAA results in Friedreich ataxia. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Jun 2009] |
|-----------------------|---|
| Specificity | Expressed in the heart, peripheral blood lymphocytes and dermal fibroblasts. |
| Purity | 70 - 90% by HPLC. |
| Conjugate | Unconjugated |
| Sequence Similarities | Belongs to the frataxin family. |
| Format | Liquid |
| Preservative | None |
| Storage | Shipped at 4°C. Upon delivery aliquot and store at -20°C or -80°C. Avoid repeated freeze / thaw cycles. Information available upon request. |

GENE INFORMATION

| Gene Name | FXN frataxin [Homo sapiens (human)] |
|-----------------|---|
| Official Symbol | FXN |
| Synonyms | FXN; frataxin; FA; X25; CyaY; FARR; FRDA; frataxin, mitochondrial; Friedreich ataxia protein; |
| Entrez Gene ID | <u>2395</u> |
| mRNA Refseq | NM 000144.4 |
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| Protein Refseq | NP_000135.2 |
|---------------------|--|
| 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; Mitochondrial Iron-Sulfur Cluster Biogenesis, organism-specific biosystem; Mitochondrial Protein Import, organism-specific biosystem; |
| Function | 2 iron, 2 sulfur cluster binding; ferric iron binding; ferrous iron binding; ferroxidase activity; iron chaperone activity; iron-sulfur cluster binding; protein binding; |