

Product datasheet for **SC308016**

Plectin (PLEC) (NM_201381) Human Untagged Clone

Product data:

Product Type:	Expression Plasmids
Product Name:	Plectin (PLEC) (NM_201381) Human Untagged Clone
Tag:	Tag Free
Symbol:	Plectin
Synonyms:	EBS1; EBSMD; EBSND; EBSO; EBSOG; EBSPA; HD1; LGMD2Q; LGMDR17; PCN; PLEC1; PLEC1b; PLTN
Vector:	<u>pCMV6 series</u>

Fully Sequenced ORF: >NCBI ORF sequence for NM_201381, the custom clone sequence may differ by one or more nucleotides

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Restriction Sites:

Please inquire

ACCN:

NM_201381

OTI Disclaimer: Due to the inherent nature of this plasmid, standard methods to replicate additional amounts of DNA in E. coli are highly likely to result in mutations and/or rearrangements. Therefore, OriGene does not guarantee the capability to replicate this plasmid DNA. Additional amounts of DNA can be purchased from OriGene with batch-specific, full-sequence verification at a reduced cost. Please contact our customer care team at custsupport@origene.com or by calling 301.340.3188 option 3 for pricing and delivery.

Our molecular clone sequence data has been matched to the reference identifier above as a point of reference. Note that the complete sequence of our molecular clones may differ from the sequence published for this corresponding reference, e.g., by representing an alternative RNA splicing form or single nucleotide polymorphism (SNP).

OTI Annotation: This TrueClone is provided through our Custom Cloning Process that includes sub-cloning into OriGene's pCMV6 vector and full sequencing to provide a non-variant match to the expected reference without frameshifts, and is delivered as lyophilized plasmid DNA.

Components: The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water).

Reconstitution Method:

1. Centrifuge at 5,000xg for 5min.
2. Carefully open the tube and add 100ul of sterile water to dissolve the DNA.
3. Close the tube and incubate for 10 minutes at room temperature.
4. Briefly vortex the tube and then do a quick spin (less than 5000xg) to concentrate the liquid at the bottom.
5. Store the suspended plasmid at -20°C. The DNA is stable for at least one year from date of shipping when stored at -20°C.

RefSeq: [NM_201381.1](#), [NP_958783.1](#)

RefSeq Size: 14646 bp

RefSeq ORF: 13548 bp

Locus ID: 5339

UniProt ID: [Q15149](#)

Cytogenetics: 8q24.3

Protein Families: Druggable Genome

Gene Summary: Plectin is a prominent member of an important family of structurally and in part functionally related proteins, termed plakins or cytolinkers, that are capable of interlinking different elements of the cytoskeleton. Plakins, with their multi-domain structure and enormous size, not only play crucial roles in maintaining cell and tissue integrity and orchestrating dynamic changes in cytoarchitecture and cell shape, but also serve as scaffolding platforms for the assembly, positioning, and regulation of signaling complexes (reviewed in PMID: 9701547, 11854008, and 17499243). Plectin is expressed as several protein isoforms in a wide range of cell types and tissues from a single gene located on chromosome 8 in humans (PMID: 8633055, 8698233). Until 2010, this locus was named plectin 1 (symbol PLEC1 in human; Plec1

in mouse and rat) and the gene product had been referred to as "hemidesmosomal protein 1" or "plectin 1, intermediate filament binding 500kDa". These names were superseded by plectin. The plectin gene locus in mouse on chromosome 15 has been analyzed in detail (PMID: 10556294, 14559777), revealing a genomic exon-intron organization with well over 40 exons spanning over 62 kb and an unusual 5' transcript complexity of plectin isoforms. Eleven exons (1-1j) have been identified that alternatively splice directly into a common exon 2 which is the first exon to encode plectin's highly conserved actin binding domain (ABD). Three additional exons (-1, 0a, and 0) splice into an alternative first coding exon (1c), and two additional exons (2alpha and 3alpha) are optionally spliced within the exons encoding the acting binding domain (exons 2-8). Analysis of the human locus has identified eight of the eleven alternative 5' exons found in mouse and rat (PMID: 14672974); exons 1i, 1j and 1h have not been confirmed in human. Furthermore, isoforms lacking the central rod domain encoded by exon 31 have been detected in mouse (PMID:10556294), rat (PMID: 9177781), and human (PMID: 11441066, 10780662, 20052759). The short alternative amino-terminal sequences encoded by the different first exons direct the targeting of the various isoforms to distinct subcellular locations (PMID: 14559777). As the expression of specific plectin isoforms was found to be dependent on cell type (tissue) and stage of development (PMID: 10556294, 12542521, 17389230) it appears that each cell type (tissue) contains a unique set (proportion and composition) of plectin isoforms, as if custom-made for specific requirements of the particular cells. Concordantly, individual isoforms were found to carry out distinct and specific functions (PMID: 14559777, 12542521, 18541706). In 1996, a number of groups reported that patients suffering from epidermolysis bullosa simplex with muscular dystrophy (EBS-MD) lacked plectin expression in skin and muscle tissues due to defects in the plectin gene (PMID: 8698233, 8941634, 8636409, 8894687, 8696340). Two other subtypes of plectin-related EBS have been described: EBS-pyloric atresia (PA) and EBS-Ogna. For reviews of plectin-related diseases see PMID: 15810881, 19945614. Mutations in the plectin gene related to human diseases should be named based on the position in NM_000445 (variant 1, isoform 1c), unless the mutation is located within one of the other alternative first exons, in which case the position in the respective Reference Sequence should be used. [provided by RefSeq, Aug 2011]

Transcript Variant: This variant (7) differs in the 5' UTR and the 5' coding region compared to variant 1. The resulting protein (isoform 1d) contains a distinct N-terminus compared to isoform 1c. Plectin isoform 1d has been found associated with Z-disks in skeletal muscle (PMID: 17389230).