

PHD finger protein 6 (PHF6) (NM_032335) Human Untagged Clone

ACCN: NM_032335



OTI Disclaimer:	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:	<ol style="list-style-type: none"> 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.
Note:	Plasmids are not sterile. For experiments where strict sterility is required, filtration with 0.22um filter is required.
RefSeq:	<u>NM_032335.2</u> , <u>NP_115711.2</u>
RefSeq Size:	1236 bp
RefSeq ORF:	939 bp
Locus ID:	84295
UniProt ID:	<u>Q8IWS0</u>
Cytogenetics:	Xq26.2
Protein Families:	Druggable Genome, Transcription Factors

Gene Summary:

This gene is a member of the plant homeodomain (PHD)-like finger (PHF) family. It encodes a protein with two PHD-type zinc finger domains, indicating a potential role in transcriptional regulation, that localizes to the nucleolus. Mutations affecting the coding region of this gene or the splicing of the transcript have been associated with Borjeson-Forssman-Lehmann syndrome (BFLS), a disorder characterized by cognitive disability, epilepsy, hypogonadism, hypometabolism, obesity, swelling of subcutaneous tissue of the face, narrow palpebral fissures, and large ears. Alternate splicing results in multiple transcript variants, encoding different isoforms. [provided by RefSeq, Jun 2010]

Transcript Variant: This variant (3) uses an alternate termination site, resulting in a distinct C-terminus (isoform 2).