

Recombinant Human PHF6

Catalog No: #GP10724



Package Size: #GP10724-1 100ug

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Description

Product Name	Recombinant Human PHF6
Brief Description	Recombinant Protein
Immunogen Description	Fusion protein corresponding to a region derived from 63-312 amino acids of human PHF6
Target Name	PHD finger protein 6
Other Names	BFLS; BORJ; CENP-31
Accession No.	Swissprot:Q8IWS0Gene Accession:BC005994
Uniprot	Q8IWS0
GeneID	84295;
Storage	-20~-80°C, pH 7.6 PBS

Background

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.?

References

Note: For in vitro research use only, not for diagnostic or therapeutic use. This product is not a medical device.

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