

PHF6 抗原(重组蛋白)

中文名称: PHF6 抗原(重组蛋白)

英文名称: PHF6 Antigen (Recombinant Protein)

别 名: BFLS; BORJ; CENP-31

储 存: 冷冻(-20℃)

相关类别: 抗原

概述

Fusion protein corresponding to a region derived from 63-312 amino acids of human PHF6

技术规格

Full name:	PHD finger protein 6
Synonyms:	BFLS; BORJ; CENP-31
Swissprot:	Q8IWS0
Gene Accession:	BC005994
Purity:	>85%, as determined by Coomassie blue stained SDS-PAGE
Expression system:	Escherichia coli
Tags:	His tag C-Terminus, GST tag N-Terminus
Background:	This gene is a member of the plant homeodomain (PHD)-like fing er (PHF) family. It encodes a protein with two PHD-type zinc fing er domains, indicating a potential role in transcriptional regulation , that localizes to the nucleolus. Mutations affecting the coding re gion of this gene or the splicing of the transcript have been asso ciated with Borjeson-Forssman-Lehmann syndrome (BFLS), a disor der characterized by cognitive disability, epilepsy, hypogonadism, hypometabolism, obesity, swelling of subcutaneous tissue of the f ace, narrow palpebral fissures, and large ears. Alternate splicing re sults in multiple transcript variants, encoding different isoforms.