

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 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.