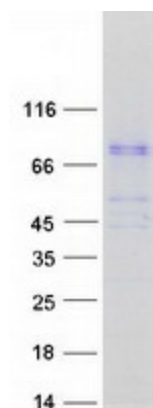


RefSeq ORF:	2016
Synonyms:	BBS2L1
Locus ID:	55212
UniProt ID:	Q8IWZ6
Cytogenetics:	4q27

Summary: This gene encodes one of eight proteins that form the BBSome complex containing BBS1, BBS2, BBS4, BBS5, BBS7, BBS8, BBS9 and BBIP10. The BBSome complex is believed to recruit Rab8(GTP) to the primary cilium and promote ciliogenesis. The BBSome complex assembly is mediated by a complex composed of three chaperonin-like BBS proteins (BBS6, BBS10, and BBS12) and CCT/TRiC family chaperonins. Mutations in this gene are implicated in Bardet-Biedl syndrome, a genetic disorder whose symptoms include obesity, retinal degeneration, polydactyly and nephropathy; however, mutations in this gene and the BBS8 gene are thought to play a minor role and mutations in chaperonin-like BBS genes are found to be a major contributor to disease development in a multiethnic Bardet-Biedl syndrome patient population. Two transcript variants encoding distinct isoforms have been identified for this gene.[provided by RefSeq, Oct 2014]

Product images:



Coomassie blue staining of purified BBS7 protein (Cat# [TP307504]). The protein was produced from HEK293T cells transfected with BBS7 cDNA clone (Cat# [RC207504]) using MegaTran 2.0 (Cat# [TT210002]).