Product name:	HUWE1 Rabbit Polyclonal Antibody
Cat number:	ABN12290
Conjugate:	Unconjugated
Size:	100μL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	Synthesized peptide derived from part region of human protein
Reactivity:	Human,Mouse
Applications:	IHC 1:50-1:300,ICC/IF 1:50-1:200
Molecular Weight:	481kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

This gene encodes a protein containing a C-terminal HECT (E6AP type E3 ubiquitin protein ligase) domain that functions as an E3 ubiquitin ligase. The encoded protein is required for the ubiquitination and subsequent degradation of the anti-apoptotic protein Mcl1 (myeloid cell leukemia sequence 1 (BCL2-related)). This protein also ubiquitinates the p53 tumor suppressor, core histones, and DNA polymerase beta. Mutations in this gene are associated with Turner type X-linked syndromic mental retardation. [provided by RefSeq, Aug 2013],disease:A chromosomal microduplication involving HUWE1 and HSD17B10 is the cause of mental retardation X-linked type 17 (MRX17) [MIM:300705]; also known as mental retardation X-linked type 31 (MRX31). Mental retardation is characterized by significantly sub-average general intellectual functioning associated with impairments in adaptative behavior and manifested during the developmental period. In contrast to syndromic or specific X-linked mental retardation which also present with associated physical, neurological and/or psychiatric manifestations, intellectual deficiency is the only primary symptom of non-syndromic X-linked mental retardation.,disease:Defects in HUWE1 are the cause of mental retardation syndromic X-linked Turner type (MRXST) [MIM:300706]; also known as mental retardation and macrocephaly syndrome. MRXST shows clinical variability. Associated phenotypes include macrocephaly and variable contractures.,domain:The HECT domain mediates inhibition of the transcriptional activity of p53.,function:E3 ubiquitin-protein ligase which mediates ubiquitination and subsequent proteasomal degradation of target proteins. Regulates apoptosis by catalyzing the polyubiquitination and degradation of MCL1. Also ubiquitinates the p53 tumor suppressor and core histones including H1, H2A, H2B, H3 and H4. Binds to an upstream initiator-like sequence in the preprodynorphin gene. Regulates neural differentiation and proliferation by catalyzing the polyubiquitination and degradation of MYCN. May regulate abundance of CDC6 after DNA damage by polyubiquitinating and targeting CDC6 to degradation.,pathway:Protein modification; protein ubiquitination.,PTM:Phosphorylated on tyrosine; phosphorylation is probably required for its ability to inhibit TP53 transactivation.,PTM:Phosphorylated upon DNA damage, probably by ATM or ATR.,sequence caution:Chimeric cDNA, contains the C-terminal part of ATP5I.,similarity:Belongs to the TOM1/PTR1 family.,similarity:Contains 1 HECT (E6AP-type E3 ubiquitin-protein ligase) domain.,similarity:Contains 1 UBA domain.,similarity:Contains 1 UIM (ubiquitin-interacting motif) repeat.,similarity:Contains 1 WWE domain.,subcellular location:Mainly expressed in the cytoplasm of most tissues, except in the nucleus of spermatogonia, primary spermatocytes and neuronal cells (By similarity). Predominantly cytosolic or perinuclear in some colorectal carcinoma cells.,subunit:Interacts with isoform p14ARF of CDKN2A which strongly inhibits HUWE1 ubiquitin ligase activity. Interacts with MYCN and CDC6.,tissue specificity:Weakly expressed in heart, brain and placenta but not in other tissues. Expressed in a number of cell lines, predominantly in those from colorectal carcinomas.,