Product Data Sheet:
WBSCR11 Rabbit Polyclonal Antibody

Cat. No:	ABN19871
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human GTF2IRD1. AA range:71-120
Reactivity:	Human,Mouse,Rat
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:50-1:200,ELISA 1:10000-1:20000
Molecular Weight:	106kDa
Purification:	Affinity purification
Synonyms:	GTF2IRD1; CREAM1; GTF3; MUSTRD1; RBAP2; WBSCR11; WBSCR12; General transcription factor II-I repeat domain-containing protein 1; GTF2I repeat domain-containing protein 1; General transcription factor III; MusTRD1/BEN; Muscle TFII-I repeat do
Background:	The protein encoded by this gene contains five GTF2I-like repeats and each repeat possesses a potential helix-loop-helix (HLH) motif. It may have the ability to interact with other HLH-proteins and function as a transcription factor or as a positive transcriptional regulator under the control of Retinoblastoma protein. This gene plays a role in craniofacial and cognitive development and mutations have been associated with Williams-Beuren syndrome, a multisystem developmental disorder caused by deletion of multiple genes at 7q11.23. Alternative splicing results in multiple transcript variants. [provided by RefSeq, Nov 2010],developmental stage:Highly expressed in developing and regenerating muscles, at the time of myofiber diversification.,disease:Haploinsufficiency of GTF2IRD1 may be the cause of certain cardiovascular and musculo-skeletal abnormalities observed in Williams-Beuren syndrome (WBS), a rare developmental disorder. It is a contiguous gene deletion syndrome involving genes from chromosome band 7q11.23.,domain:The N-terminal half may have an activating activity.,function:May be a transcription regulator involved in cell-cycle progression and skeletal muscle differentiation. May repress GTF2I transcriptional functions, by preventing its nuclear residency, or by inhibiting its transcriptional activation. May contribute to slow-twitch fiber type specificity during myogenesis and in regenerating muscles. Binds troponin I slow-muscle fiber enhancer (USE B1). Binds specifically and with high affinity to the EFG sequences derived from the early enhancer of HOXC8.,PTM:Phosphorylated upon DNA damage, probably by ATM or ATR.,similarity:Belongs to the TFII-I family.,similarity:Contains 5 GTF2I-like repeats.,subunit:Interacts with the retinoblastoma protein (RB1) via its C-terminus.,tissue specificity:Highly expressed in adult skeletal muscle, heart, fibroblast, bone and fetal tissues. Expressed at lower levels in all other tissues tested.,
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA 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.

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