

Cat. No:	ABN19231
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human TCOF1. AA range:41-90
Reactivity:	Human,Mouse
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:200-1:1000,ELISA 1:5000-1:20000
Molecular Weight:	152kDa
Purification:	Affinity purification
Synonyms:	TCOF1; Treacle protein; Treacher Collins syndrome protein

Background: This gene encodes a nucleolar protein with a LIS1 homology domain. The protein is involved in ribosomal DNA gene transcription through its interaction with upstream binding factor (UBF). Mutations in this gene have been associated with Treacher Collins syndrome, a disorder which includes abnormal craniofacial development. Multiple transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Sep 2008],disease:Defects in TCOF1 are the cause of Treacher Collins syndrome (TCS) [MIM:154500]. TCS is an autosomal dominant disorder of craniofacial development that occurs with an incidence of 1/50,000 live births. The clinical features of TCS are bilaterally symmetrical and include: (1) abnormalities of the external ears, atresia of the external ear canals, and malformation of the middle ear ossicles, which may result in conductive hearing loss; (2) lateral downward sloping of palpebral fissures, frequently with colobomas of the lower eyelids; (3) hypoplasia of the mandible and zygomatic complex; (4) cleft palate.,function:May be involved in nucleolar-cytoplasmic transport. May play a fundamental role in early embryonic development, particularly in development of the craniofacial complex.,PTM:Phosphorylated upon DNA damage, probably by ATM or ATR.,similarity:Contains 1 LisH domain.,

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.

**For Research use only
IMMUNOLOGICAL SCIENCES**