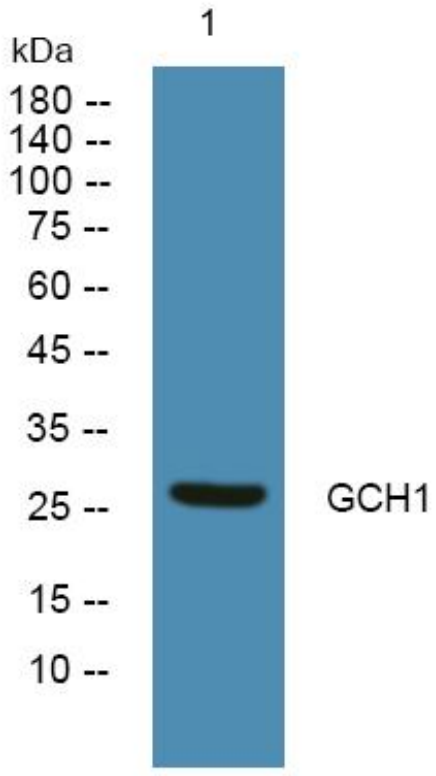
GCH1 Monoclonal Antibody

Catalog No	BYmab-05611
Isotype	IgG
Reactivity	Human;Mouse;Rat
Applications	WB
Gene Name	GCH1 DYT5 GCH
Protein Name	GTP cyclohydrolase 1 (EC 3.5.4.16) (GTP cyclohydrolase I) (GTP-CH-I)
Immunogen	Synthesized peptide derived from part region of human protein
Specificity	GCH1 Monoclonal Antibody detects endogenous levels of protein.
Formulation	Liquid in PBS containing 50% glycerol, and 0.02% sodium azide.
Source	Monoclonal, Mouse,IgG
Purification	The antibody was affinity-purified from mouse antiserum by affinity-chromatography using epitope-specific immunogen.
Dilution	WB 1:500-2000
Concentration	1 mg/ml
Purity	≥90%
Storage Stability	-20°C/1 year
Synonyms	
Observed Band	27kD
Cell Pathway	Cytoplasm . Nucleus .
Tissue Specificity	In epidermis, expressed predominantly in basal undifferentiated keratinocytes and in some but not all melanocytes (at protein level).
Function	catalytic activity:GTP + H(2)O = formate + 2-amino-4-hydroxy-6-(erythro-1,2,3-trihydroxypropyl)-dihydropteridine triphosphate.,disease:Defects in GCH1 are the cause of dystonia type 5 (DYT5) [MIM:128230]; also known as progressive dystonia with diurnal fluctuation, autosomal dominant Segawa syndrome or dystonia-parkinsonism with diurnal fluctuation. DYT5 is a DOPA-responsive dystonia. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DYT5 typically presents in childhood with walking problems due to dystonia of the lower limbs and worsening of the dystonia towards the evening. It is characterized by postural and motor disturbances showing marked diurnal fluctuation. Torsion of the trunk is unusual. Symptoms are alleviated after sleep and aggravated by fatigue and excercise. There is a favorable response to

	L-DOPA without
Background	This gene encodes a member of the GTP cyclohydrolase family. The encoded protein is the first and rate-limiting enzyme in tetrahydrobiopterin (BH4) biosynthesis, catalyzing the conversion of GTP into 7,8-dihydroneopterin triphosphate. BH4 is an essential cofactor required by aromatic amino acid hydroxylases as well as nitric oxide synthases. Mutations in this gene are associated with malignant hyperphenylalaninemia and dopa-responsive dystonia. Several alternatively spliced transcript variants encoding different isoforms have been described; however, not all variants give rise to a functional enzyme. [provided by RefSeq, Jul 2008],
matters needing attention	Avoid repeated freezing and thawing!
Usage suggestions	This product can be used in immunological reaction related experiments. For more information, please consult technical personnel.

Products Images



Western Blot analysis of various cells using GCH1 Monoclonal Antibody