Immunotag™ HFE Monoclonal Antibody

Antibody Specification

Catalog No.	ITM0330
Product Description	Immunotag™ HFE Monoclonal Antibody
Size	50 µg, 100 µg
Conjugation	HRP, Biotin, FITC, Alexa Fluor® 350, Alexa Fluor® 405, Alexa Fluor® 488, Alexa Fluor® 555, Alexa Fluor® 594, Alexa Fluor® 647
IMPORTANT NOTE	This product is custom manufactured with a lead time of 3-4 weeks. Once in production, this item cannot be cancelled from an order and is not eligible for return.
Target Protein	HFE
Clonality	Monoclonal
Storage/Stability	-20°C/1 year
Application	WB,IF,ELISA
Recommended Dilution	Western Blot: 1/500 - 1/2000. Immunofluorescence: 1/200 - 1/1000. ELISA: 1/10000. Not yet tested in other applications.
Concentration	1 mg/ml
Reactive Species	Human
Host Species	Mouse
Immunogen	Purified recombinant fragment of human HFE expressed in E. Coli.
Specificity	HFE Monoclonal Antibody detects endogenous levels of HFE protein.
Purification	Affinity purification
Form	Ascitic fluid containing 0.03% sodium azide.
Gene Name	HFE
Accession No.	Q30201 P70387
Alternate Names	HFE; HLAH; Hereditary hemochromatosis protein; HLA-H

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Description	hemochromatosis(HFE) Homo sapiens The protein encoded by this gene is a membrane protein that is similar to MHC class I-type proteins and associates with beta2-microglobulin (beta2M). It is thought that this protein functions to regulate iron absorption by regulating the interaction of the transferrin receptor with transferrin. The iron storage disorder, hereditary haemochromatosis, is a recessive genetic disorder that results from defects in this gene. At least nine alternatively spliced variants have been described for this gene. Additional variants have been found but their full-length nature has not been determined. [provided by RefSeq, Jul 2008],
Protein Expression	Brain,Colon,Placenta,Whole blood,
Subcellular Localization	extracellular space,early endosome,plasma membrane,integral component of plasma membrane,external side of plasma membrane,integral component of membrane,cytoplasmic vesicle,MHC class I protein complex,apical part of cell,basal part of cell,perinuc
Protein Function	Additional isoforms seem to exist,disease:Defects in HFE are a cause of hereditary hemochromatosis (HH) [MIM:235200]. HH is an autosomal recessive inborn disorder of iron metabolism. It is the most common recessive disease in Caucasians. HH is characterized by abnormal intestinal iron absorption and progressive increase of total body iron, which results in midlife in clinical complications including cirrhosis, cardiopathy, diabetes, endocrine dysfunctions, arthropathy, and susceptibility to liver cancer. Since the disease complications can be effectively prevented by regular phlebotomies, early diagnosis is most important to provide a normal life expectancy to the affected subjects.,disease:Defects in HFE are a cause of porphyria variegata (PV) [MIM:176200]. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. PV is the most common form of porphyria in South Africa. It is characterized by skin hyperpigmentation and hypertrichosis, abdominal pain, tachycardia, hypertension and neuromuscular disturbances. High fecal levels of protoporphyrin and coproporphyrin, increased urine uroporphyrins and iron overload are typical markers of the disease.,disease:Defects in HFE are associated with susceptibility to diabetic nephropathy [MIM:612635]; also called susceptibility to microvascular complications of diabetes type 7 (MVCD7) or susceptibility to diabetic proliferative retinopathy. Diabetic nephropathy is a kidney disease and resultant kidney function impairment due to the long standing effects of diabetes on the microvasculature (glomerulus) of the kidney. Features include increased urine protein and declining kidney function.,function:Binds to transferrin receptor (TFR) and reduces its affinity for iron-loaded transferrin.,similarity:Belongs to the MHC class I family.,similarity:Contains 1 Ig-like C1-type (immunoglobulin-like) domain.,subunit:Binds TFR through the extracellular domain in a pH-dependent manner.,tissue specificity:In all tissues tested except brain.,
Usage	For Research Use Only! Not for diagnostic or therapeutic procedures.