

## **Rabbit Anti-HFE antibody**

SL12335R

Product Name:	HFE
Chinese Name:	遗传性血色病蛋白相关蛋白1抗体
Alias:	dJ221C16.10.1; Hemochromatosis; Hemochromatosis protein; Hereditary hemochromatosis protein; Hereditary hemochromatosis protein HLA H; HFE 1; HFE;
	HFE_HUMAN; HFE1; HH; High Fe; HLA H; HLA-H; HLAH; MGC:150812; MGC10379; MGC103790; MHC class I like protein HFE; MVCD7; TFOTL2.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,Cow,Horse,Sheep,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800Flow-Cyt=1µg/TestICC=1:100-
	500IF=1:100-500 (Paraffin sections need antigen repair)
	not yet tested in other applications.
	optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	38kDa
<b>Cellular localization:</b>	The cell membrane
Form:	Lyophilized or Liquid
<b>Concentration:</b>	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from Human HFE/Hemochromatosis:262- 348/348 <extracellular></extracellular>
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized
	antibody is stable at room temperature for at least one month and for greater than a year
	when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of
	antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	The features of hemochromatosis include cirrhosis of the liver, diabetes, hypermelanotic
	pigmentation of the skin, and heart failure. Since hemochromatosis is a relatively easily
	treated disorder if diagnosed, this is a form of preventable cancer. The HFE protein,

which is defective in hereditary hemo-chromatosis, normally is expressed in crypt enterocytes of the duodenum where it has a unique, predominantly intracellular localization. In placenta, the HFE protein co-localizes with and forms a stable association with the transferrin receptor (TfR), providing a link between the HFE protein and iron transport. Immunocytochemistry shows that the HFE protein and TfR both are expressed in the crypt enterocytes. Western blots show that, as is the case in human placenta, the HFE protein in crypt enterocytes is physically associated with the TfR and with  $\beta$ 2-microglobulin. It is proposed that HFE has two mutually exclusive activities in cells: inhibition of uptake or inhibition of release of iron and that the balance between serum transferrin saturation and serum transferrin-receptor concentrations determines which of these functions predominates. The gene which encodes HFE maps to human chromosome 6p21.3.

## **Function:**

Binds to transferrin receptor (TFR) and reduces its affinity for iron-loaded transferrin.

## Subunit:

Binds TFR through the extracellular domain in a pH-dependent manner.

Subcellular Location: Membrane; Single-pass type I membrane protein.

**Tissue Specificity:** Expressed in all tissues tested except brain.

## **DISEASE:**

Defects in HFE are a cause of hemochromatosis (HFE) [MIM:235200]. A disorder of iron metabolism characterized by iron overload. Excess iron is deposited in a variety of organs leading to their failure, and resulting in serious illnesses including cirrhosis, hepatomas, diabetes, cardiomyopathy, arthritis, and hypogonadotropic hypogonadism. Severe effects of the disease usually do not appear until after decades of progressive iron loading. Defects in HFE are associated with variegate porphyria (VP) [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. VP 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. Note=Iron overload due to HFE mutations is a precipitating or exacerbating factor in variegate porphyria. Defects in HFE are associated with susceptibility to microvascular complications of diabetes type 7 (MVCD7) [MIM:612635]. These are pathological conditions that develop in numerous tissues and organs as a consequence of diabetes mellitus. They include diabetic retinopathy, diabetic nephropathy leading to end-stage renal disease, and diabetic neuropathy. Diabetic retinopathy remains the major cause of new-onset blindness

among diabetic adults. It is characterized by vascular permeability and increased tissue ischemia and angiogenesis.
Similarity: Belongs to the MHC class I family. Contains 1 Ig-like C1-type (immunoglobulin-like) domain.
SWISS: Q30201
Gene ID: 3077
Database links:
Entrez Gene: 3077 Human
<u>Omim: 613609</u> Human
SwissProt: Q30201 Human
Unigene: 233325 Human
Important Note:
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
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