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HFE Rabbit pAb

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Catalog # AP54828

Product Information

Primary Accession	Q30201
Reactivity	Human
Predicted	Rat, Dog, Mouse, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	40108
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from Human HFE/Hemochromatosis
Epitope Specificity	262-348/348
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Membrane; Single-pass type I membrane protein.
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.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	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 β2-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.

Additional Information

Gene ID	3077
Other Names	Hereditary hemochromatosis protein, HLA-H, HFE, HLAH
Dilution	Flow-Cyt=1 □g/Test
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

Protein Information

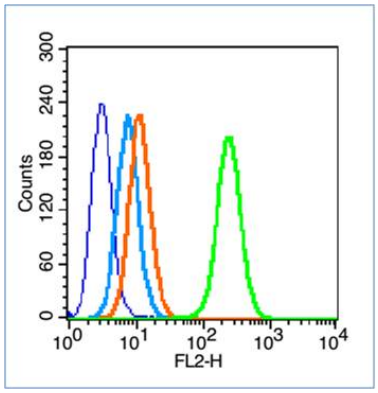
Name	HFE
Synonyms	HLAH
Function	Binds to transferrin receptor (TFR) and reduces its affinity for iron-loaded transferrin.
Cellular Location	Cell membrane; Single-pass type I membrane protein
Tissue Location	Expressed in all tissues tested except brain.

Background

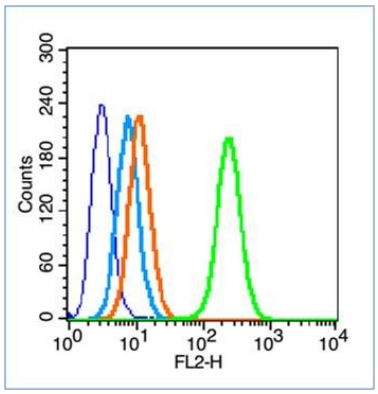
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Images

Blank control (blue line): HL60(fixed with 70% ethanol Overnight at 4℃).
Primary Antibody (green line): Rabbit Anti-iHFE antibody (AP54828),Dilution: 0.2 µg /10^6 cells;
Isotype Control Antibody (orange line): Rabbit IgG .



Secondary Antibody (white blue line): Goat anti-rabbit IgG-PE,Dilution: 1 μg /test.



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