

Hypobetalipoproteinemia

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Hypobetalipoproteinemia is a rare genetic disorder causing abnormally low levels of LDL cholesterol. It is thought to be caused by mutated apolipoprotein B. The patient can have hypobetalipoproteinemia and simultaneously have high levels of HDL cholesterol. According to The Merck Manual, "no treatment is required." Typically in hypobetalipoproteinemia, plasma cholesterol levels will be around 80-120 mg/dL, LDL cholesterol will be around 50-80 mg/dL, and longevity can be expected with good nutrition. Notably, in people who do NOT have the genetic disorder hypobetalipoproteinemia, a low cholesterol level may be a marker for poor nutrition, wasting disease, cancer, hyperthyroidism, and liver disease. ([From the Wikipedia article Hypobetalipoproteinemia](#).)



Recent Publications on Hypobetalipoproteinemia:

- [\[Clinical and genetic heterogeneity in hypobetalipoproteinemia\]](#)
Familial hypobetalipoproteinemia (FHBL) is one of the causes of... 1st October, 2009
- Med Clin (Barc). 2009 Jun 13;133(2):61-2. Epub 2009 May 14. ([DOI Direct Link](#))
- [\[Familial hypobetalipoproteinemia: clinical characterization of a new mutation in the APOB gene\]](#)
BACKGROUND AND OBJETIVE: Familial hypobetalipoproteinemia (FHB) is usually... 1st October, 2009
Servicio de Endocrinologia, Hospital General, Segovia, Espana.- Med Clin (Barc). 2009 Jun 13;133(2):57-60. Epub 2009 May 14. ([DOI Direct Link](#))
- [\[Apolipoprotein B gene mutations and fatty liver in Japanese hypobetalipoproteinemia.\]](#)
BACKGROUND: Familial hypobetalipoproteinemia (FHBL) is a hereditary... 25th September, 2009
Department of Internal Medicine, Graduate School of Medical Science,- Clin Chim Acta. 2009 Jan;399(1-2):64-8. Epub 2008 Sep 24. ([DOI Direct Link](#))
- [\[Familial hypobetalipoproteinemia: early neurological, hematological, and ocular manifestations in two affected twins responding to vitamin supplementation.\]](#)
Familial hypobetalipoproteinemia is a disorder of lipid metabolism... 24th September, 2009
aDepartments of Pediatric Medicine, Rheumatology, and Child- Curr Opin Pediatr. 2009 Dec;21(6):824-827. ([DOI Direct Link](#))
- [\[PCSK9 Dominant Negative Mutant Results in Increased LDL Catabolic Rate and Familial Hypobetalipoproteinemia.\]](#)
OBJECTIVE: Proprotein convertase subtilisin/kexin type 9 (PCSK9) is a... 19th September, 2009
IRT-UN l'institut du Thorax LI7R415. 8, Quai Moncoussu BP70721, 44007- Arterioscler Thromb Vasc Biol. 2009 Dec;29(12):2191-2197. Epub 2009 Sep ([DOI Direct Link](#))
- [\[Recurrent familial hypobetalipoproteinemia-induced nonalcoholic fatty liver disease after living donor liver transplantation.\]](#)
Familial hypobetalipoproteinemia (FHBL) is one of the causes of... 11th September, 2009

Department of Surgery and Medical Science, Graduate School of Medical- Liver Transpl. 2009 Jul;15(7):806-9. ([DOI Direct Link](#))

- [Cryptogenic cirrhosis in a patient with familial hypocholesterolemia due to a new truncated form of apolipoprotein B.](#)
Familial hypobetalipoproteinemia (FHBL) is an autosomal codominantly... 10th September, 2009
Functional Unit of Biochemistry of Metabolic Diseases, Laboratory of- Eur J Gastroenterol Hepatol. 2009 Jan;21(1):104-8. ([DOI Direct Link](#))
- [Do diabetes and obesity promote hepatic fibrosis in familial heterozygous hypobetalipoproteinemia?](#)
BACKGROUND: Familial hypobetalipoproteinemia (FHBL) is a hereditary... 4th August, 2009
- Intern Emerg Med. 2009 Feb;4(1):71-3. Epub 2008 Jul 17. ([DOI Direct Link](#))
- [Mutations and polymorphisms in the proprotein convertase subtilisin kexin 9 \(PCSK9\) gene in cholesterol metabolism and disease.](#)
Hypercholesterolemia is one of the major causes of coronary heart disease... 7th July, 2009
Institut Nationale de la Sante et de la Recherche Medicale (INSERM), U781,- Hum Mutat. 2009 Apr;30(4):520-9. ([DOI Direct Link](#))
- [Dysmorphic findings in two cases of abeta/hypobetalipoproteinemia.](#)
OBJECTIVE: Familial hypobetalipoproteinemia (FHBL) is characterized by... 29th May, 2009
Medical Genetics Branch, National Human Genome Research Institute,- Clin Dymorphol. 2009 Apr;18(2):90-1. ([DOI Direct Link](#))
- [Familial hypobetalipoproteinemia due to apolipoprotein B R463W mutation causes intestinal fat accumulation and low postprandial lipemia.](#)
OBJECTIVE: Familial hypobetalipoproteinemia (FHBL) is characterized by... 7th April, 2009
Department of Clinical Medicine and Emerging Diseases, University of- Atherosclerosis. 2009 Sep;206(1):193-8. Epub 2009 Feb 2. ([DOI Direct Link](#))
- [Identification of patients with abetalipoproteinemia and homozygous familial hypobetalipoproteinemia in Tunisia.](#)
BACKGROUND: Abetalipoproteinemia (ABL) and Homozygous Familial... 13th March, 2009
Research Unit of Genetic and Biologic Factors of Atherosclerosis, Faculty- Clin Chim Acta. 2009 Mar;401(1-2):51-6. Epub 2008 Nov 21. ([DOI Direct Link](#))
- [Functional analysis of two novel splice site mutations of APOB gene in familial hypobetalipoproteinemia.](#)
Familial hypobetalipoproteinemia (FHBL) is a co-dominant disorder... 3rd March, 2009
Department of Biomedical Sciences, University of Modena and Reggio Emilia,- Mol Genet Metab. 2009 Feb;96(2):66-72. Epub 2008 Dec 11. ([DOI Direct Link](#))
- [Fatty liver and insulin resistance: not always linked.](#)
One significant clinical symptom of familial hypobetalipoproteinemia... 14th November, 2008
Washington University School of Medicine, Department of Internal Medicine,- Trans Am Clin Climatol Assoc. 2008;119:217-23; discussion 223-4.
- [\[Neuroacanthocytosis update\]](#)
Neuroacanthocytosis is an inclusive term for a genetically heterogeneous... 13th August, 2008
Department of Psychiatry, Kagoshima University Graduate School of Medical- Brain Nerve. 2008 Jun;60(6):635-41.

Hypobetalipoproteinemia Patents:



- 4281013- [Antiatherosclerotic use of khellin and khellinin](#)
- 4581462- [Pyrrolizidine-3-ones](#)
- 5395846- [Amino Bi- and tri-carbocyclic aklane bis-aryl squalene synthase inhibitors](#)
- 5441946- [Phosphonate derivatives of lipophilic amines](#)
- 5556990- [Polyarylcarbamoylaza- and -carbamoylalkanedioic acids](#)
- 5563128- [Phosphonate derivatives of lipophilic amines](#)
- 5595872- [Nucleic acids encoding microsomal triglyceride transfer protein](#)
- 5596020- [Amino bi- and tri-carbocyclic alkane bis-aryl squalene synthase inhibitors](#)
- 5789197- [Microsomal triglyceride transfer protein](#)
- 6093699- [Method for gene therapy involving suppression of an immune response](#)
- 4542228- [4-Hydroxy-furochromone intermediates for antiatherosclerotic compounds](#)
- 4540798- [Antiatherosclerotic aminoethenyl furochromones](#)
- 4438274- [Antiatherosclerotic furochromones](#)
- 4284569- [Antiatherosclerotic furochromones](#)
- 4304722- [Benzo\[1,2-b:5,4-b'\]difuran-3\(2H\)-ones](#)
- 4313881- [5-Hydroxyfurochromones](#)
- 4313882- [7-Methyl-6-methylthio-, methylsulfanyl-, and methylsulfonyl-methyl-furochromones](#)
- 4313883- [Halofurochromones](#)
- 4399146- [Treatment of atherosclerosis with khellin-related furochromones](#)
- 4407821- [Lipidic compositions for use in dietetics, reanimation and therapeutics](#)
- 4412071- [Antiatherosclerotic compositions](#)
- 4434295- [Anti-atherosclerotic 6,7-dihydro-7,7-disubstituted-khellin analogs](#)
- 6150514- [14 Kilobase deletion in the promoter region of BRCA1 in a breast cancer family](#)
- 6225451- [Chromosome 11-linked coronary heart disease susceptibility gene CHD1](#)
- 7052855- [Anti-APO-B-48 monoclonal antibody, hybridoma producing the same, and method for measuring APO-B-48 using the same](#)
- 7101838- [Method to prevent accelerated atherosclerosis using \(sRAGE\) soluble receptor for advanced glycation endproducts](#)
- 7223591- [Strain of lactic acid bacterium and edible compositions, drugs and veterinary products containing it](#)
- 7232667- [Keratinocyte growth factor-2 polynucleotides](#)
- 7407943- [Antisense modulation of apolipoprotein B expression](#)
- 7419950- [Methods of inhibiting ectopic calcification](#)
- 7494972- [Method for inhibiting accelerated atherosclerosis in a subject suffering from hypercholesterolemia or hypertriglyceridemia](#)
- 7511131- [Antisense modulation of apolipoprotein B expression](#)
- 7521234- [Mammalian immortalized liver cell](#)
- 7033744- [Method for proliferating a liver cell, a liver cell obtained thereby, and use thereof](#)
- 6942967- [Target for treating atherosclerosis, obesity and type II diabetes](#)
- 6887712- [Methods and compositions to lower plasma cholesterol levels](#)
- 6288048- [Cholesterol and hedgehog signaling](#)
- 6288234- [Multibinding inhibitors of microsomal triglyceride transferase protein](#)
- 6309844- [Anti-apo-B-48 monoclonal antibody, hybridoma producing the same, and method for measuring apo-B-48 using the same](#)
- 6492365- [Microsomal triglyceride transfer protein](#)
- 6524613- [Hepatocellular chimeraplasty](#)
- 6551990- [Methods of inhibiting ectopic calcification](#)
- 6555651- [Ligand binding site of rage and uses thereof](#)
- 6616926- [Methods of modulating lipid metabolism and storage](#)
- 6693077- [Keratinocyte growth factor-2](#)
- 7527962- [Human diacylglycerol acyltransferase 2 \(DGAT2\) family members and uses therefor](#)

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