



## Apolipoprotein B

### Alternative Names

APOB  
APOB100  
APOB48  
Abetalipoproteinemia, Normotriglyceridemic,  
Steinberg Type  
Apolipoprotein B Allotypes  
Ag Lipoprotein Types  
Low Density Lipoprotein Cholesterol Level  
Quantitative Trait Locus 4  
LDLCQ4  
Hypobetalipoproteinemia, Familial  
FHBL  
Hypobetalipoproteinemia, Familial, 1  
FHBL1  
Hypobetalipoproteinemia, Normotriglyceridemic  
Acanthocytosis with Hypobetalipoproteinemia

### Record Category

Gene locus

### WHO-ICD

N.B.: Classification not applicable to gene loci.

### Incidence per 100,000 Live Births

N/A to gene loci

### OMIM Number

107730

### Mode of Inheritance

Autosomal dominant

### Gene Map Locus

2p24

### Description

Apolipoprotein B is the main apolipoprotein of chylomicrons and low density lipoproteins (LDL). It occurs in the plasma in 2 main forms, apoB48 and apoB100. The first is synthesized exclusively by the gut, the second by the liver. ApoB48 is present in chylomicrons and chylomicron remnants and plays an essential role in the intestinal absorption of dietary fats. ApoB100 is a component of VLDL, intermediate density

lipoprotein (IDL) and low density lipoproteins (LDL). ApoB100 contributes to hepatic and peripheral tissue uptake of LDL by receptor recognition.

### Molecular Genetics

The Apolipoprotein B gene has been mapped on the short arm of chromosome 2, with an approximate length of 43 kilobases and 29 exons. The LDL-binding domain of the molecule is proposed to be located between the residues 3129 and 3532.

### Epidemiology in the Arab World

In an Arab patient with hypobetalipoproteinemia and absent plasma apolipoprotein B and resulting from a consanguineous marriage, Huang et al. (1989) demonstrated deletion of the entire ApoB gene exon 21 (211 basepairs coding for amino acids 1014 to 1084).

### Kuwait

~~Al Muhtaseb et al. (1989) studied a group of non-obese non insulin dependent diabetic (NIDDM) Arab women requiring insulin and found significantly elevated concentrations of apolipoprotein B, LDL apoB, and apoB/apoA1 in diabetic women compared with control subjects.~~

~~Moussa et al. (1998) conducted a case control study of 460 Kuwaiti obese children aged 6-13 years, matched by age and sex to 460 normal weight controls to evaluate the relation among apolipoproteins (Apos) B and A-I and the degree of obesity, glucose and insulin levels, body fat distribution, and serum lipids. The determination of obese children was performed through an illustrative cross sectional study of 2,400 school children. The Apo B mean concentrations was found to be higher in obese males and females ( $p < 0.001$ ), whereas the A:I:B ratio was found to be significantly lower in obese children ( $p < 0.001$ ). The A:I levels were found to be significantly lower in obese females ( $p < 0.01$ ) while it was alike for both obese and non obese males. The study demonstrated a positive correlation among Apo B levels and insulin and insulin:glucose ratio in obese~~



~~children and a positive association among Apo A I levels and total cholesterol, high and low density lipoprotein cholesterol. Moussa et al. (1998) documented poor Apo profile among obese Kuwaiti children and proposed early diagnosis of Apo changes throughout childhood for prevention of early-onset atherogenesis in adulthood.~~

Al-Bustan et al. (2009) investigated the possible association of clinical variables and apolipoprotein (APOE, APOCI and APOB) polymorphisms with the development of myocardial infarction (MI) and coronary heart disease (CHD) in Kuwaitis. APOB genotype was determined in 143 Kuwaiti CHD patients with (n = 88) and without (n = 55) MI and in 122 controls matched for gender and age. In this study, Al-Bustan et al. (2009) determined APOB genotype by studying a polymorphic segment in the promoter region codes for a signal peptide with two common alleles, a 27- amino-acid insertion allele (I) and a 24-amino-acid deletion allele (D). A statistically significant association was found between CHD and medical history of diabetes mellitus, hypertension, high cholesterol and family history of CHD. A highly significant association was found for family history and the development of MI. No significant differences were found for allele or genotype frequencies between CHD patients and controls. Al-Bustan et al. (2009) concluded that the strong effect of family history suggests a major genetic component for the development of CHD in Kuwaitis, but this association does not appear to be related to the APO genes investigated in this study. They also suggested that the results in this study encourage future research into these and other polymorphisms and their potential association with MI and CHD in the Kuwaiti population.

#### **Lebanon**

Alberto et al. (1999) determined the molecular basis of familial hypercholesterolemia in 59 patients from 31 unrelated Brazilian families. All patients were screened for the Lebanese mutation, gross abnormalities of the LDLr gene, and the point mutation in the codon 3500 of the apolipoprotein B-100 gene. None of the 59 patients presented the apoB-3500 mutation, suggesting that familial defective ApoB-100 is not a major cause of inherited hypercholesterolemia in Brazil.

#### **Syria**

~~Al Kateb et al. (1998) studied 192 male patients with suspected coronary artery disease, who underwent catheterization. They noticed that patients without coronary artery disease were slightly younger, thinner, smoked less, and had lower cholesterol, low density lipoprotein~~

~~cholesterol, and apolipoprotein B levels than did those who had coronary artery disease.~~

#### **United Arab Emirates**

Frossard et al. (1995) analyzed a random sample of 101 nationals from the United Arab Emirates for the VNTR located in the 3' region of the human ApoB gene. A total of 11 alleles (33, 35, 37, 39, 41, 43, 45, 47, 49, 51, and 53 repeats) were observed in the general population. However, the 43 and 41 repeats were the most observed alleles. Frossard and colleagues suggested that the peculiar pattern of alleles in the population of the United Arab Emirates can be quite useful for DNA fingerprinting and population genetic studies (Frossard et al., 1995).

Frossard and Lestringant (1999) used a polymerase chain reaction-based assay to investigate the allele and genotype frequency distributions of the alleles of a hypervariable region located in the 3' of the human apolipoprotein B (apoB) gene. The analysis of DNA samples of 367 unrelated UAE nationals (201 males and 166 females) revealed the presence of 18 different alleles, ranging from 21 to 55 repeats, making up 51 genotypes occurring in Hardy-Weinberg proportions with a heterozygosity index of 80.9%. This observation leads to the conclusion that this marker is very informative in the Emirati population and may be very useful for UAE-specific DNA fingerprinting as well as to assess the role of the apoB gene in cardiovascular diseases. Frossard et al. (1999) investigated the associations between genetic variations of the apoB gene and clinical diagnosis of essential hypertension. They compared the distribution of the alleles of a highly polymorphic variable number of tandem repeats localized 3' to the human apoB gene, the apoB 3' hypervariable region (HVR), in a group of normotensive and a group of hypertensive individuals. DNA samples from 437 unrelated UAE nationals (215 normotensives and 222 hypertensives) were collected. The apoB 3' HVR allele and genotype status were determined using a polymerase chain reaction-based assay. In the UAE population, Frossard and colleagues (1999) found 18 alleles underlying a total of 51 genotypes. The distribution of these alleles is significantly different between normotensive and hypertensive UAE nationals. The main peak of the distributions occurred at 35 repeats among hypertensives (with a relative frequency of 25.7% versus 19.6% in normotensives) and at 37 repeats among normotensives (28.8% versus 20.3% in hypertensives). Alleles with 21, 23, 25, 49, and 55 repeats are found in hypertensives only (with a combined relative frequency of 7.6%). Frossard et al. (1999) concluded that variations of the apoB gene, or of a nearby gene, that may be in linkage



disequilibrium with these alleles play a role in the development of essential hypertension in UAE nationals.

## References

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Huang LS, Ripps ME, Korman SH, Deckelbaum RJ, Breslow JL. Hypobetalipoproteinemia due to an apolipoprotein B gene exon 21 deletion derived by Alu-Alu recombination. *J Biol Chem.* 1989; 264(19):11394-400. PMID: 2567736

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## Related CTGA Records

Apolipoprotein C-I

Apolipoprotein E

Essential Hypertension

## External Links

<http://herkules.oulu.fi/isbn9514251598/html/x203.html>

<http://www.gpnotebook.co.uk/cache/x20030127061759665170.htm>

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