Table 1. Genetic classification of dyslipidemia.

Classification	Examples of Genetic Diseases* (Frequency)	Genetic Defect	Common Lipid Abnormalities**	Clinical Findings
Type I 'Chylomicronemia syndrome' (increased triglyceride levels and chylomicron particles)	Lipoprotein lipase deficiency	Genetic defect of LPL gene – autosomal recessive	TG levels in the 1000's, with as high as over 10,000 mg/dl Partial genetic defects may have TG levels > 500 mg/dL	Eruptive xanthomas Pancreatitis
	ApoCII deficiency (1:1,000,000 for either of the above)	Genetic defect of ApoCII gene – autosomal recessive		
Type IIa (increased LDL cholesterol levels and/or particles)	Polygenic hypercholesterolemia (1:20)	Multiple genetic defects – inheritance likely dependent upon underlying genetic abnormality	LDL-C levels > 130 mg/dL	Increased risk of ASCVD
	Heterozygous familial hypercholesterolemia (1:500)	Dysfunction or absence of LDL receptor – autosomal dominant	LDL-C levels > 190 mg/dL	Increased risk of premature ASCVD Tendon xanthoma
	Homozygous familial hypercholesterolemia (1:1,000,000)		LDL-C levels sometimes > 350 mg/dL, but most often 500 - 1000 mg/dL	Increased risk of premature ASCVD Tendon xanthoma
Type IIb (increased triglyceride levels, and increased LDL and VLDL cholesterol levels and/or particles)	Familial combined hyperlipidemia (1:50-1:200)	Multiple genetic defects of various apolipoproteins and/or LPL genes - ? autosomal dominant	LDL-C levels > 160 mg/dL TG levels> 300 mg/dL	Increased risk of ASCVD
Type III (increased triglyceride levels and increased intermediate-density lipoprotein cholesterol levels and/or particles)	Familial dysbetalipoproteinemia (1:1000-1:5000)	Genetic defect of apoE gene – autosomal recessive, or more rarely, autosomal dominant	LDL-C > 220 mg/dL TG levels > 300 mg/dL	Palmar xanthomata (orange discoloration of skin creases, tuberoeruptive xantomata of elbows and knees Increased risk of premature ASCVD
Type IV (increased triglyceride levels and increased VLDL cholesterol levels and/or particles)	Familial hypertriglyceridemia (1:50-1:100)	Unknown genetic defect – autosomal dominant	TG levels > 150 mg/dL	Unclear if increased risk of ASCVD
Type V (increased triglyceride levels and increased chylomicron and VLDL cholesterol levels and/or particles)	Hyperprebetalipoproteinemia (unknown frequency, very rare)	Unknown genetic defect – possibly due to an LPL inhibitor – unknown inheritance	TG levels > 500 mg/dL LDL-C levels > 130 mg/dL	Eruptive xanthomas Pancreatitis Increased risk of ASCVD

LDL = low-density lipoprotein; VLDL = very low-density lipoprotein; Apo = apolipoprotein; LPL = lipoprotein lipase.

*In addition to genetic causes, many of these hyperlipoproteinemias may be acquired or secondary to high carbohydrate diets, medications, and/or underlying diseases. Hence, the frequencies listed above only refer to the number of patients with the genetic abnormality specified and do not reflect the total frequency by which the types of lipid phenotypes are observed in clinical practice.

** The listed lipid values for genetic abnormalities should be considered common lipid values, intended to provide general values that might be encountered in clinical practice. These lipid values are not intended to be diagnostic. Lipid values can vary widely among individual patients with genetic abnormalities, especially when combined with secondary causes of dyslipidemia and other genetic abnormalities.